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GENOME FINLAND

From Rare Diseases to Data Economy

HUP HELSINKI UNIVERSITY PRESS

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Abbreviations

AI	Artificial Intelligence
BBMRI	The Biobanking and Biomolecular Resources Research Infrastructure
BBMRI-ERIC	ERIC here signifies a European Research Infrastructure Consortium
BRCA	BReast CAncer gene
CNS	Congenital Nephrotic Syndrome
CVD	Cardiovascular disease(s)
DIPP	The Type 1 Diabetes Prediction and Prevention Project
DNA	Deoxyribonucleic acid
DTC	Direct-to-consumer
EHDS	European Health Data Space
EHR	Electronic health record
EMBL	European Molecular Biology Laboratory
EPO	Erythropoietin (a hormone)
ERC	European Research Council
ETENE	Finnish National Advisory Board on Research
EU	European Union
FDH	Finnish Disease Heritage
Fimea	Finnish Medicines Agency
FMC	Finnish Maternity Cohort
FIMM	Institute for Molecular Medicine Finland
FINBB	Finnish Biobank Cooperative
GDPR	General Data Protection Regulation
GMO	Genetically modified organism
GWAS	Genome-Wide Association Studies
HeLa	Cell line based on Henrietta Lacks's cancer
HGP	Human Genome Project
HSD	Health Sector Database

HUNT	Trøndelag Health Study, HUNT biobank
HVA	Well-Being Services County/Counties (hyvinvointialue)
ICT	Information and communications technology
IPR	Intellectual Property Rights
KTL	National Public Health Institute (Kansanterveyslaitos)
MEE	Ministry of Economic Affairs and Employment
MIT	Massachusetts Institute of Technology (USA)
MSH	Ministry of Social Affairs and Health
NIH	National Institute of Health (USA)
NHS	National Health Service (UK)
OECD	Organisation for Economic Co-operation and Development
PCR	Polymerase chain reaction
PIN	Personal identification number
PRS	Polygenic risk score(s)
R&D	Research and development
RNA	Ribonucleic acid
SHOK	Centre for Strategic Excellence (Strategisen huippuosaamisen keskittymä)
SISu	Sequencing Initiative Suomi
Sitra	Finnish Innovation Fund (Suomen itsenäisyyden juhlarahasto)
SNP	Single nucleotide polymorphism
Stakes	National Research and Development Centre for Welfare and Health (Sosiaali- ja terveysalan tutkimus- ja kehittämislaitos)
T1D	Type 1 Diabetes
Tekes	Finnish Funding Agency for Technology and Innovation (Tekniikan kehittämiskeskus)
TEO	National Authority for Medicolegal Affairs (Terveydenhuollon oikeusturvakeskus)
THL	Finnish Institute for Health and Welfare (Terveyden ja hyvinvoinnin laitos)
Tukija	National Committee on Medical Research Ethics

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CHAPTER 1

Introduction

In 2004, Leena Peltonen (1952–2010),[1] a renowned Finnish medical geneticist, claimed in an interview that 'Finland is 10–15 years ahead of biobanks under construction elsewhere in the world' (*Bioteknologia* 4/2004). Her words exemplify the conviction that Finland is—or rather could become—the Eldorado for cutting-edge genetics as a result of its unique population, excellent public databases and data sourcing infrastructure, and world-class exper-tise in biomedical research. This ambition has been the lodestar for frontline biomedical scientists, governmental officials, politi-cians, and business promoters involved in innovation policy for some thirty years. Today, the view that Finland could belong in the vanguard of research in medical genetics is more solid and promising than ever, and intensely advocated at home and abroad. The Finnish Ministry of Social Affairs and Health effectively summarized this self-image when it published Finland's national Genome Strategy, stating:

> Finland is particularly well placed to utilize genomic data. From a global perspective, Finland's strengths include a high standard of healthcare, uniform treatment practices, reliable healthcare registers, a long tradition of high-quality genetic research, and the willingness of the population to participate in scientific research. … Finns are genetically relatively homogenous. This provides special opportunities to combine genomic and health data. As a result, genetic mechanisms targeted by drugs can be identified in a manner that is difficult, if not impossible, elsewhere. (Ministry of Social Affairs and Health 2015a, 12–13)

This book is a thorough examination of Genome Finland, outlined in the extract above. By 'Genome Finland' we refer to the predominant presentation of Finnish genetics as a success story, and Finland as a milieu of unique excellence for biomedical research. In this book, we study the content of the success story and the image, how they came about, and what lies beneath the surface. We present a historical narrative of the rise of medical genetics in Finland by describing phases of the development and tracing background events and initiatives back to the late 1960s. However, our purpose is not to write a detailed chronicle. Instead, our analysis provides a mapping of the multiple paths and pursuits whereby the trajectory of genetics has evolved in Finland, resulting in an enabling environment for innovation. In a nutshell, this book follows paths from 'heritage' of rare biological and medical traits to an asset in scientific and commercial competition in global biomedicine.

While doing this, we sort out the variety of stakeholders and driving forces in science, politics, and the economy influencing the development. We unpack the manifold dynamics between the stakeholders, their interests, and pursuits. Furthermore, we situate the development and alleged success of Finnish genetics in an international landscape of developments in the science, social concerns, and business connected with new genetics and genomics.[2] Our narrative from the 1960s to the present day closely reflects our own research interests and undertakings over the past 20 years, bringing together findings from our empirical published and unpublished sociological research conducted in a number of research projects. As such, we are aware of unavoidable omissions in our historical recounting of the development of Genome Finland.

Genes in society: Two configurations

At the turn of the millennium, when the human genome was sequenced, the promise of genetics to medicine in terms of scientific breakthroughs, improved diagnostics, new cures, and the

boosting of medical business became more pronounced than ever. Obviously, Finland was not the only country where expectations of the revolution in healthcare associated with advances in medical genetics prevailed. From a global perspective, some commentators have even said that Finland is more of a peripheral curiosity in this field (Wheelwright 2005).

Specifically, molecular biology focused on DNA and RNA spearheaded the development of biology into Big Science, especially in the USA since the 1960s (Hood and Rowen 2013; Sampat 2012). In the late 1980s, the National Institute of Health (NIH), the biggest public medical science funder in the USA, launched a grand project for mapping the human genome, directed by two big names in molecular genetics, James Watson and Francis Collins. As this Human Genome Project (HGP) approached its massive sequencing phase at the end of the 20th century, a private entrepreneur scientist Craig Venter and his company Celera Genomics challenged the NIH project and started its own mapping and sequencing effort. This famous race was a transnational endeavour, surrounded by tremendous hype about unprecedented improvements in medicine. Lofty expectations circulated around HGP and human genomics in general (Douglas 2005). According to many, the possibilities of genomics appeared limitless in terms of diagnosis, treatment, and even cures, particularly after the publication of the first map of the human genome in 2003. No longer restricted to single genes, studies in genetics targeting the whole range of the human genome—the interactions and co-occurrences of the tiny biochemical building blocks of humans—were to open 'the book of life' and reveal the secrets of health and disease. Commercially, this was also a major watershed moment for venture capital investors and start-ups in biomedicine, since genomics was expected to provide plenty of new opportunities for patenting, licensing, and other forms of commercialization (Parthasarathy 2011; Styhre 2015).

In the late 1990s and early 2000s, the excitement facilitated the launch of large-scale initiatives to boost research in medical genetics—renamed 'genomics' and, later, 'post-genomics'—and

build research infrastructures serving that purpose all over the world. Although HGP opened up new horizons and established the practices and networks of transnational biomedical science, the aforementioned initiatives were mostly national, with the few exceptions of regional projects like CARTaGENE in Québec and Generation Scotland. These initiatives received generous funding from governments and public innovation agencies, private charities dedicated to science funding, and medical companies, especially big pharmaceutical enterprises. They were launched with great expectations of boosting science and hopes of bringing benefits to healthcare and public health in the future. In addition, governmental and corporate funders in particular underlined that investments in medical genomics might create business opportunities in an emerging high-tech biomedical market and thus benefit the national economy. The establishment of an Icelandic biobank run by a private company, deCODE Genetics, and the founding of the UK Biobank have received perhaps the most public and scholarly attention (e.g. Busby and Martin 2006; Hoeyer and Tutton 2005; Pálsson 2007; Fortun 2008). Yet parallel efforts were launched simultaneously in Estonia, Singapore, Japan, and Latvia (Swede et al. 2007), which based their operations in relatively small populations claimed to be distinctly unique in their genetic composition. The initiatives in Finland belong in this category.

The turn of the millennium, with its genomics hype and boom, was not the first time genetics became a significant social and political issue. The first configuration in which scientific interest in biological heredity became entwined with political and social issues took shape in the first half of the 20th century. At that time, many scientists, public debaters, politicians, and social activists all over the Western world claimed that genetics as an emerging paradigm of biology would provide the scientific basis for eugenic politics and social reform. In eugenics reasoning, genetics—focused mostly on experimental and biostatistical studies of the biological mechanisms of Mendelian laws—was enmeshed with late 19th-century doctrines of racial biology and anthropology, theories

of the biological inheritance of mental deficiency and abnormality, and Galtonian views of the inheritance of mental capabilities (Weindling 1989; Kevles 1995). Politically, such eugenic science was deployed to alert the public and governments to the biological and mental 'degeneration' of individuals, populations, nations, and the whole human race looming in the near future, as, it was said, 'genetically inferior' individuals, classes, and races procreated extensively.

Pioneering scientists in genetics like J.B.S. Haldane, Julian Huxley, Lancelot Hogben, and Herbert Jennings distanced themselves from eugenics as science and politics as early as the 1920s and 1930s (Kevles 1995, 122–147). They refuted the core claims of eugenics: the genetic origin of race and racial hierarchy, genetic causes of mental deficiency, mental illness and immorality, and the related view of the expanding degeneration of the population. Despite scientific rejection, eugenic ideology and policy gained firm ground in most Western countries in the 1920s and 1930s, with widespread popular advocacy of policies and legislation fighting the alleged genetic deterioration of the nation and its population. The focus and intensity of coercive eugenic measures, such as marriage bans and sterilizations, varied from one country to another, but everywhere they were social-class biased, gendered, and racialized, targeting predominantly marginalized individuals and groups, poor working-class people, people classified as non-white, and women (e.g. Broberg and Roll-Hansen 2005; Koch 2000; SOU 2000; Stern 2005).

After the Second World War, eugenics fell out of grace altogether. The abuses of science, especially the experimental biology and medicine associated with the genocidal atrocities that Nazi Germany conducted before and during the war, were revealed to the world (see Proctor 1988; Kuntz 2004), and eugenics was seen as identical to Nazi racism. In this context, genetics more broadly also came to be viewed with more suspicion, and as morally questionable.

In the aftermath of the revelations about 'Nazi science', the re-codification of medical and research ethics, especially in the

natural and life sciences, was ignited as an international effort. The rules and codes of proper conduct and procedures, and the indicators of misconduct in treatment and experimental research with human subjects, were negotiated, explicitly declared, and agreed by professional and Western research communities in documents ranging from the Nuremberg Code in 1947 to subsequent versions of the Helsinki Declaration between 1964 and 2013 (Faden and Beauchamp 1986; Gallin and Bedzow 2022; Weindling 2022). The protection of patients and other persons involved in experimental trials or other research has been the main purpose of the ethical codes. In the latter half of the 20th century, genetics has constantly been the subject of newly codified research ethics—especially studies in human and medical genetics—and ethical alertness and heightened concern over proper conduct of research and its ethical consequences that has been partly due to the eugenic past associated with the contemporary field (e.g. Wertz 1998; Dyck 1997). In addition, the idea of the exceptional character of knowledge about human genes—of individual persons, groups of people, and populations—has facilitated the growth of ethical concerns (e.g. Murray 1997; see Chapter 5). The intensification of ethical reflection on genetics happened side by side with advances in molecular biology, spearheaded by studies in genetics, as it became the epistemic paradigm and predominant research technology in biology that was maturing into Big Science. Accompanying this development, human genetics evolved and facilitated the growth of genetics into a major domain of medicine, and molecular genetics engendered some of the most influential all-round biotechnology in the world today.

In Finland, the trajectory of eugenics—or 'racial hygiene' as it was referred to in the Finnish discussion—followed the path taken in many other European countries. In the first decades of the 20th century, concern about degeneration, ideas of inborn and genetic defects of mental capabilities or morality, and a hierarchical taxonomy of races spread from European and Scandinavian discussions to certain circles of Finnish academics and the cultural elite (Mattila 1999; Hietala 2005). During the 1920s and

1930s—the first two decades of the new nation—eugenics as science and politics gained a strong foothold in the country. This trend was caused by assumptions of the rapid procreation of the 'lower elements' of the population, referring to the working class and the poor. The discussion facilitated widespread civic activism and political demands for eugenic reforms and legislation to fight such a threat to the vital strength of the nation and *folket* (kansa in Finnish). The milestone of eugenics advocacy in Finland was the Sterilization Act that allowed the sterilization of 'idiots', 'imbeciles', and individuals with mental defects, as well as persons with 'asocial' behaviour, if there was a reason to believe that their defect traits were inheritable. The law was passed in 1935 with almost no dissonance in public and parliamentary discussion.

Following the European trend, eugenics also lost most of its ideological and political appeal in Finland after the war; however, this did not result in the dismantling of existing legislation and administrative procedures established during its heyday (Mattila 1999). The sterilization policy epitomized this continuity, and a new sterilization law was passed in Finland in 1950. The overwhelming majority of individuals subjected to sterilisation were women, often poor single mothers, a practice that continued in the country until the late 1960s.

For genetics as a science, a new era started after the Second World War. In the period from the path-breaking studies that established the double helix structure of DNA (deoxyribonucleic acid) in 1953 to the publication of the map of the whole human genome in 2003, molecular genetics became the vanguard and backbone of the life sciences. During this period, scientific and lay advocates of expansive genetics struggled with the eugenic past, including the explicit and implicit racist tendencies of the early 20th-century discussions on biological inheritance (Kevles 1995, 269–290; Kerr et al. 1998; Kay 2000, 279, 291–292; Meskus 2009, 58–64). These attempts, mostly rather ambivalent, to distance the new genetics from its scientifically, socially, and ethically dubious legacy happened in a milieu in which worldwide codification of medical and research ethics was under way, and

bioethics as a special branch of philosophy emerged and was established. Advances in human genetics toward the end of the century increasingly raised critical concerns about the social and ethical consequences of the code-breaking effort that would allow the secrets of the 'book of life' to be known and socially, politically, or economically appropriated. Many social scientists were involved in addressing those concerns in the 1980s and 1990s (see below).

Even today, the above debate continues in many forms. However, the way the new genetics and its applications in medicine and elsewhere are assembled in society—i.e. the second configuration for genes in society—is significantly different from eugenics prior to the Second World War. The social significance of eugenics as science and ideology was essentially *reactive* in the sense that it articulated a concern about the degeneration of the population and mobilized government and civil society to fight that threat with coercive policy measures targeting marginalized individuals or groups of people who were assumed to be 'genetically' defective or inferior. By contrast, the core and most pronounced message of the new genetics—in current terms, genomics or post-genomics—has been *proactive*, especially in the context of medicine and healthcare; it promises to enhance the possibilities of disease prevention and cure and the promotion of healthy lifestyles, which would greatly benefit public health and the economy. The scientific and technological breakthroughs in medical genomics in the 21st century—for example, gene tests for BRCA (BReast CAncer gene) cancers, lactose intolerance, and the adverse effect of certain drugs, whole genome sequencing for Lynch syndrome, advances in the exact diagnosis of rare genetic diseases—have made hopes and expectations about the revolution in medicine more intense and expansive. Advances in biomedicine based on molecular genomics and gene technology were seen to introduce radical changes in Western medical knowledge of causes, biological mechanisms, and cures for diseases, thus opening—according to its advocates—'unprecedented' opportunities to find treatments for incurable conditions, better medicines, more precise diagnoses, and more efficient prevention of illness.

To a great extent, the promise of health benefits that the new genomics will bring in the near future has been emphatically individual-focused, and, since the late 1990s, the praise and promotion of medical genomics has been associated with 'personalized medicine', today often also called 'precision medicine'. The term refers to a variety of visions of personally tailored biomedical care in which diagnosis and treatments based on knowledge of average patients will be replaced by individually tailored diagnoses, risk assessment, and medical care based on personal mapping of genomes and biomarkers of health risks and susceptibility to diseases (e.g. National Academy of Sciences 2011; Tutton 2014). The advocates of personalized medicine underline its predictive and preventive character; personally tailored care would include a programme for health maintenance including diet and lifestyle instruction, health checks and, if needed, medication, all embedded in the analysis of the person's genetic and other biological make-up (see Chapters 3 and 6). Anu Wartiovaara, a prominent Finnish neuroscientist and geneticist, summarized this vision of 'the enormous steps forward in treatment and prevention of diseases' in the following way:

> If I knew my own DNA code, it would enable the identification of my inherited personal susceptibilities [to diseases], and thus it would be possible to focus lifestyle counselling on the areas that would benefit me the most. In addition, most of the adverse effects of medicines could be avoided, and the efficacy of medication could be enhanced. (Wartiovaara 2005, 67, own translation)

Genomics through the looking glass of social sciences

Since the turn of the millennium, historians, philosophers, bioethicists, and scholars of law and political studies have eagerly examined the events and topics related to the promise and making of the genomic revolution. The social sciences are not an exception to this. The first wave of social science scholarship on

the new genetics took off at approximately the same time as the NIH launched HGP at the end of 1980s. Critical discussion of the social, ethical, and political consequences of the expansion of genetic knowledge dominated the social science research agenda throughout the Western world in the 1990s (see e.g. Kevles and Hood 1993; Nelkin and Lindee 1995). The topics of the studies were widely varied; many focused on the issue of possible genetic discrimination and its multiple dimensions, and there were numerous discussions of 'backdoor eugenics', a term that referred to an implicit and subtle racial discrimination the new genetics may indirectly reinforce (Duster 1990). Prenatal and genetic screenings and their significant expansion at that time arising from new medical technology were perhaps the main topic that social scientists in different Western countries studied from historical, ethnographic, ethical, and policymaking perspectives, problematizing and challenging the rather straightforwardly affirmative views of the medical professionals (e.g. Rothman 1996; Rapp 2004; Jallinoja 2002; Meskus 2009). Quite a few studies were also conducted on the impact and reception of knowledge on genetic disease or genetic susceptibility to disease by people subjected to genetic testing (e.g. Marteau and Richards 1999; Konrad 2005; Haga et al. 2013). They emphasized that the testing procedure, the results, and their interpretation tend to engender ambivalence and worries in the people involved, and they also underlined the importance of the social context in which the testing and reception of personal genetic knowledge take place.

In the wake of HGP and related hype, and the technological advances in molecular biology, a wide range of topics related to these developments began to complement and, gradually, overshadow the previous agenda. Furthermore, quite a few researchers focused on studying the social, political, and economic contexts and consequences of the genomics revolution in medicine, healthcare, and other walks of life, such as, for example, forensics, while research funding considerably increased during the 2000s, especially in the USA and in the UK.

Many social scientists claimed that the 21st-century genomics and post-genomics—terms which refer, on the one hand, to the shift in study focus from single genes to large sequences or even whole genomes and, on the other, to gene functions in protein coding and a variety of other physiological processes—will facilitate and enable profound and unprecedented changes not only in medicine and healthcare, but also in society and culture at large. British sociologist Nikolas Rose argued that we are entering the age of the 'politics of life itself', as ruling and governing people by political authorities, and political and human rights, are now 'concerned with our growing capacities to control, manage, engineer, reshape, and modulate the very vital capacities of human beings as living creatures' (Rose 2007, 3), which, in turn, may bring about novel issues concerning human and political rights. Californian sociologist Jenny Reardon (2017) claimed that rapidly expanding Western genomics—as a research and medical endeavour, with the global data sourcing it requires—does not live up to the liberal values it claims to cherish, especially justice and equality. This demands profound rethinking of social justice as a concept and social practice under the 'post-genomic condition' in which we find ourselves. These two works exemplify analyses which focus on the putting of new genomic knowledge, data, and related technology to multiple uses in society. These kinds of studies discuss the general consequences of a biotechnological revolution in terms of changes in our worldviews and self-conception, new modes of social and political power and control, the reshaping of rights, justice, and inequalities, and the redefinition of social relationships and belonging in regard to family and parenthood, kinship, ethnicity, and race. A wide range of trends is highlighted in this literature as the essence of the future in the making. At the one end, Rose and many others emphasize the promise of the new medical genomics to widen the possibilities of biological modification, enhancement of personal life, and control over one's own health and vital functions, even at the cellular and molecular levels. This would open a future landscape of unprecedented individual choice over the facts and requirements of life itself (Parens 1999;

Novas and Rose 2000; Elliott 2003; Rose 2007). At the other end, studies like *The Social Life of DNA* (2016) by American sociologist Alondra Nelson and *Population Genetics and Belonging* (2017) by Finnish anthropologist Venla Oikkonen focus on new constellations of prejudice and injustice, contestation over race and ethnicity, and ambivalence about ancestry, which the expansion of new technologies for mapping the human genome and genomic variation introduces and facilitates.

Many studies have adopted a more restricted approach to the emerging post-genomic era, focusing on changes in the life sciences and medical research. Some studies have critically analysed the 'geneticization' and 'biomedicalization' of medicine as research and practice (e.g. Conrad 2005; Clarke et al. 2010; for an overview, Weiner et al. 2016). Others have focused on the transformation of biology and biomedicine into techno-sciences, as all research seems to have become dependent on and embedded in complex technical equipment and machinery (e.g. Rheinberger 1995; Lock et al. 2000; Burri and Dumit 2007; Clarke et al. 2010). Many analyses have addressed the changes in the organization of scientific research into multi-centred networks or platforms crossing national, institutional, and disciplinary boundaries, and linking academic researchers to private companies, civic organizations, and other non-academic stakeholders (e.g. Gottweis 1998; Keating and Cambrosio 2003; Kleinman 2003). These studies have discussed shifts in epistemology in the life sciences and biomedicine, the requirement of increasing standardization and its impact, and the effects of the negotiation and reconciliation of interests in research practices.

In discussions of the transformation of the life sciences and biomedicine after HGP, a focal topic was the increasingly pressing requirement to use massive numbers of samples and health-related data from ever-bigger research populations. This required more sequencing and other laboratory technology and, especially, advanced information and communications technology (ICT) capable of analysing such data (e.g. *BioSocieties* 2013; Cambrosio et al. 2014; Douglas 2014; Mackenzie et al. 2016). Some scholars

claim that the life sciences and biomedicine are becoming increasingly data-intensive, statistical, and even algorithmic, which trend is profoundly changing the epistemology of biology and medicine (Leonelli 2014; 2016; Mazzocchi 2015). Data mining—sorting out massive data sets using advanced algorithms or artificial intelligence (AI)—highlights correlations and collative patterns as the desired outcome of scientific study (Mayer-Schönberger and Cukier 2013), a development that Chris Anderson, former editor of *Wired*, has commented on:

> Petabytes allow us to say: 'correlation is enough'. We can stop looking for models. We can analyze the data without hypotheses about what it might show. We can throw the numbers into the biggest computing clusters the world has ever seen and let statistical algorithms find patterns where science cannot. … Correlation supersedes causation, and science can advance even without coherent models, unified theories, or really any mechanistic explanation at all. (Anderson 2008)

Addressing the changes in research in the post-genomic era (discussed above), social scientists have also been interested in the reorganization of research into big transnational research consortia and supporting infrastructures for collecting and storing tissue samples and other data sourcing. A number of studies have focused on the general governance of research or infrastructure endeavours, which are characterized by a multiplicity and heterogeneity of stakeholders and an environment in which the demands of national and international legislation and regulations are very diverse (e.g. Gottweis 1998). Biobanks and biobank networks have been a favourite subject of these governance studies (e.g. Tutton and Corrigan 2004; Gottweis and Petersen 2008; Hoeyer et al. 2017).

Because data-intensive research in medical genomics requires masses of sample and patient data from a variety of sources and populations, data sourcing is a crucial element in research governance. Social scientists have addressed several critical aspects of data sourcing for biomedical research by discussing privacy and

data protection (Tanner 2017; Kaye 2012) and the trust and willingness of people in relation to donation of tissue samples and personal data to biobanks and biomedical research (e.g. Levitt and Weldon 2005; Dabrock et al. 2012; Critchley et al. 2015). Social scientists, *vis-à-vis* bioethicists and legal scholars, have actively contributed to discussions on informed consent, the most debated and controversial topic related to sample donation and data sourcing for the new genetics (Allen et al. 2013; Hoeyer et al. 2005). Many social scientists have been critical of mainstream bioethics because of its emphasis on individual choice and a negligence concerning the social context of informed consent (e.g. Hoeyer 2010). Some have pointed out that informed consent has become merely a procedure and formality that provides an ethical safety belt for data sourcing and research, indicating a sort of 'empty ethics' in post-genomics (Corrigan 2003). A few studies have discussed the active participation of donors and patients in data sourcing, and their opportunities to exert active influence over data sourcing procedures and the use of data in research beyond informed consent. These studies are closely connected with social science discussions of patient activism, as well as disease advocacy groups and alliances in the context of medical genomics and biomedicine (e.g. Gibbon and Novas 2008), as exemplified by studies on the Généthon biobank, founded and run by a patient organization in France (Callon and Rabeharisoa 2007; Mayrhofer 2008).

Not surprisingly, the economic and commercial dimensions of genomics and biomedicine in general have been a major social science topic over the past 25 years. From the early 2000s, social scientists studying organ donation, sperm and ovum donation for assisted reproduction, and donation and other sourcing of tissue samples for biomedical research (e.g. Andrews and Nelkin 2001) have critically discussed the entanglement of advanced biomedicine with commercial pursuits and exploitation. In this context, Catherine Waldby and Robert Mitchell introduced the concept of 'tissue economies' to address global 'systems for maximizing the productivity [of biomaterials] through strategies of circulation, leverage, diversification, and recuperation' (Waldby and Mitchell 2006, 31).

Early studies on the Iceland biobank project, initiated by entrepreneur scientist Kari Steffensson and his company deCODE Genetics with the help of Big Pharma financing, opened a perspective onto biobanks and other forms of data sourcing in the service of large genome sequencing projects as an essential part of the global tissue economy (Rose 2001; Pálsson and Rabinow 2001; Fortun 2008). Ever since, social scientists have studied and discussed a wide range of topics related to the economy of the genomic revolution in medicine.

A plethora of studies have focused on the commercialization of scientific findings, data, or data management services (e.g. Tanner 2017; Jarvenpaa and Markus 2018); direct-to-consumer (DTC) genetic tests by private companies like 23andMe and CureTogether and related data sourcing have also been popular topics (e.g. Nelson and Robinson 2014). Other studies have focused on the commercial aspects of biobanking (Caulfield et al. 2014) and biobanks' financing issues (Simeon-Dubach and Henderson 2014; Chalmers et al. 2016). Furthermore, many studies have analysed commercial activities connected with biomedicine and medical genomics as examples of the marketization of academic science, the formation of innovation business clusters or ecosystems, and the dynamics of a knowledge-based economy (McMeeking and Harvey 2002; Sunder Rajan 2006; Cooke 2007; Bicudo 2018). Medical genomics and cutting-edge biomedicine as an investment domain for venture capital, and the significant impact of venture capital in boosting and creating new opportunities within the field of genomics, have also been explored (Dibner et al. 2003; Howell et al. 2003). Medical genomics as the flagship of national or regional innovation policy programmes and booster of high-tech business has been analysed in a number of case studies (see below), while substantial research has concentrated on demonstrating how current biomedical science—its organization, interests in 'knowledge production', and epistemology—is entangled with the dynamics of contemporary capitalism, especially neoliberalism and financialization. The latter topic is discussed in terms of 'bioeconomy' (Cooper 2008; Birch and Tyfield 2013), 'biocapital' (Sunder Rajan

2006), or 'technoscientific capitalism' (Muniesa and Birch 2020). Overall, it is fair to say that most social science studies of 21st-century medical genomics and biomedicine have addressed their economic and commercial aspects in one way or another.

As noted above, the mapping of the human genome and related advanced biotechnology allowed identification of genetic variations at the level of a single nucleotide throughout the genome. This advance promised to make medicine unprecedentedly precise and accurate, even in the realms of clinics and prevention. In the late 1990s, market analysts specializing in the emerging genomics market coined the term 'personalized medicine' for this future development; since then, 'precision medicine' and 'stratified medicine' have also been used to signify the promise (e.g. Tutton 2014). According to a recent definition, the term refers

> to a medical model using characterisation of individuals' phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention (European Commission 2014).

Quite early on, social scientists became interested in the advocacy and implementation of medical genomics as medical practice and even beyond, in the guise of personalized medicine. British sociologist Adam Hedgecoe's study (2004) of pharmacogenetics in the diagnosis of Alzheimer's disease and breast cancer was one of the first analyses of this topic. Ever since, social scientists have studied and discussed a variety of settings and aspects of personalized medicine: sociotechnical imaginary and expectations related to the concept; economic and commercial aspects and uses; DTC genetic tests; patient activism and disease advocacy; the configuration and implementation of the idea in different medical specialities; and the diffusion of the meaning of personalized medicine more broadly. British sociologist Richard Tutton's *Genomics and the Reimagining of Personalized Medicine* (2014) provides an overview of this multidimensional social science discussion. It also

shows that social science research has become more intense, running parallel with the production of influential policy reports and biomedical innovation initiatives on personalized medicine (e.g. National Academy of Sciences 2011; European Science Foundation 2012) in the first two decades of the 21st century. In the 2010s, the advocates of personalized medicine began to suggest that big data, advanced data mining algorithms, and AI were showing the way to proceed with personalized medicine and would make its promise come true (e.g. Hood and Friend 2011; Topol 2011; Swan 2012; Pentland et al. 2013). They promoted the idea that algorithmic and computerized mining of massive amounts of health-related population and personal data will help to replace medicine based on statistical averages and risk groups with 'precision medicine': defining health-promoting lifestyles, preventive or anticipatory medical measures, and treatment person-by-person based on accurate anticipatory calculations. Again, many social scientists have analysed and discussed this turn toward data-driven, personalized medicine, the related intensification of data sourcing, and their epistemological (Leonelli 2014; 2016), social and ethical (Prainsack 2017; Reardon 2017), and economic (Hoeyer 2019; Wadmann and Hauge 2021) implications and repercussions, which are highly diverse. Among the concerns of social scientists is the trend—or rather the potential—of data-driven personalized medicine to direct healthcare practices towards predictive modelling, risk assessment, and diagnosis based on data mining, thus transforming medical care. This approach suggests that 'algorithms [will] replace human experience, intuition and contact', facilitating the outsourcing of clinical consultation to smartphones or website portals and eclipsing 'human contact and durable social relations' (Prainsack 2017, 184–185).

Numerous social science studies have concentrated on cases of national or regional projects that advance domestic medical genomics and its infrastructure and take advantage of the medical biotechnology boom in terms of science, healthcare, and—especially—the economy. The Icelandic Health Sector Database (HSD) has received a lot of attention (Rose 2001; Pálsson

and Rabinow 2001; Pálsson 2007; Fortun 2008), and a number of analyses have been published about the UK biobank (Busby and Martin 2006; Petersen 2005; Tutton et al. 2004). In addition, social scientists have examined biobanking and related extensive health data sourcing in a number of local settings, from Sweden (Hoeyer 2004b; Cool 2016), Denmark (Wadmann and Hoeyer 2018; Hoeyer 2023), and Estonia (Fletcher 2004) to regional projects in India (Sunder Rajan 2006, 83–103) and several countries in Southeast Asia (Ong 2016; Sun 2017).

Anthropologist Aihwa Ong's *Fungible Life* (2016) is an example of this type of study. In it, she presents the case of the foundation and consolidation of Biopolis, a centre for biomedicine in Singapore. Biopolis is a state-driven project, founded by the Singaporean innovation agency in 2003, with lavish governmental funding and the ambition to become the leading hub of medical genomics and biomedical science in Southeast Asia, which would also boost Singapore's economy. Ong conducted ethnographic research at Biopolis for nine years, interviewing scientists, managers, governmental officials, and politicians involved in the project, and observing in labs and on campuses. The focus of the study is the entanglement of global biomedical science with national and regional specificities, requirements, and interpretations, as scientists at Biopolis were mapping gene variants, disease risks, and biomarkers by mobilizing the 'Asian' genome, bodies, and health data for biomedical research. By differentiating between Chinese, Indian, and Malay DNA, the project aimed to compile ethnic-stratified databases out of Singapore's population, which would represent the majority populations in Asia and could be utilized in scientific and commercial R&D (research and development) throughout the world.

Ong does not focus on science only; on the contrary, her analysis primarily addresses the ways that the big scientific initiative and actual biomedical research are intertwined with national and regional politics and the economy. She shows how Biopolis is considered predominantly a national project with multiple objectives. It aims to put the distinctiveness and excellence of 'Asian'

genomes, populations, and science on the map of global genomics, appropriating the scientific and commercial asset that 'Asian' population and genome offer and preventing their exploitation by 'Western' pharmaceutical companies and associated scientists. This is envisaged as creating opportunities for scientific and commercial collaboration with Western partners on equal terms and, simultaneously, as guarding the perquisites of 'Asian' people. Ong analyses how global biomedicine and genomics, impregnated by an entrepreneurial ethos that sees scientific and commercial pursuits as indistinguishable, are modified and redefined to serve the national cause and public good, articulated both in terms of national and ethnic uniqueness, and as competitiveness in global biomedical science and business.

Ong herself says that the topic of the book is future-making in which people and institutions involved in genomic science and biomedicine mobilize, utilize, and tackle uncertainties and unknowns related to trajectories and their outcomes yet to come. Her study, and many similar studies, underline that grand endeavours of future-making like Biopolis inevitably take place in specific local, regional, or national settings and locations while being simultaneously networked globally. Furthermore, *Fungible Life* demonstrates that in such future-making, the forces and features of the particular national or regional context reconfigure and redefine the genome, the biomedical science, and the cultural, political, and economic factors that enable and condition big genomic initiatives. In many ways, national biobank initiatives reflect calls to regard the DNA of the nation-state-bound population as a national resource, thus laying political sovereign claim over nature (Tupasela and Tamminen 2015; Hinterberger and Porter 2015; Rabinow 2002).

Studying genomics in Finnish society

The authors of this book have participated in and contributed to the above-described social science research on the genomics turn in medicine for over two decades. We have participated in more

than ten research projects, both domestic and international, most of them multidisciplinary, with both public science funding and funding from governmental innovation agencies.[3] In the course of project work, we have engaged in collaboration, dialogue, and even debate with other social scientists, bioethicists, lawyers, biomedical scientists, data scientists, clinicians, patient organizations, biobank executives and experts, innovation policy officials, politicians, and lay people as members of the public and as our interviewees and informants. Over the years, we have participated in over 100 conferences, seminars, workshops, and meetings, and followed innumerable public presentations and PowerPoint shows on the topic. Moreover, we have also read thousands of pages of documentary material. We have come across multiple faces of the genomics revolution in many settings, and the range of topics we have analysed and written and given talks about over the years is wide. This book, *Genome Finland*, is an outcome of this journey of ours as individual scholars and as a team, and is based mainly on what we have published,[4] but also on what we have experienced and witnessed along the way.

Our book is about biomedical science *in* society. More precisely, we focus on medical genetics and genomics, which we think of not as a world of its own in relation to society but as being and happening in society. This means that we approach genetics and genomics as being developed in certain social, political, and economic circumstances, and within a specific epistemic culture (see Knorr Cetina 1999). The latter has enabled and shaped the formation of genetics and genomics as science and a medical specialty, which, in turn, has given genetics and biomedicine the consistence and power to be of social, political, or economic influence in society. 'Society', in this case, refers to a particular society, namely, Finland, which simultaneously invokes many entities: a nation, the state, the national economy, the population, a people. When we analyse and discuss genetics in Finland, we think of our topic as a constellation formed by biomedical science and its institutions; governmental and other political stakeholders; economic actors and institutions (e.g. private companies, entrepreneurs,

and investors); and the public. Yet we do not merely examine the reciprocal relationships between science, politics, business, and the public in the domestic arena, but also analyse genetics in Finland by emphasizing the dynamics and dependencies between local, national, and transnational (or 'global') developments and arrangements.

This book is not a history or sociology of science, as we do not concentrate on the historical changes, progress, or paradigm shifts of medical genetics. Obviously, changes in the concept of the gene (Fox Keller 2000; Kay 2000; Torgersen 2009), the becoming of the 'whole genome' as the focus of genetics (Dupre 2004), the development of PCR (polymerase chain reaction) and sequencing technology (Rabinow 1996), and the growth of research data and populations (Leonelli 2016) are significant background factors in our story, but our focus is not predominantly on the scientific context of genetics. Instead, we examine genetics in Finland insofar as it exemplifies biomedicine as societal or even political practice. This means that we discuss the interlaced social, political, economic, and technological conditions within which medical genetics has developed, transformed into genomics, and reached its status in Finland. In addition, we analyse views and ideas about the utility and benefits of biomedical research and attempts to utilize scientific research and its results socially, politically, and economically—including the various initiatives and organizational changes accompanying this drive. Regarding the latter topic, we emphasize that its potential and expectations are central in the promotion and utilization of cutting-edge biomedicine. Furthermore, this book unpacks Genome Finland—that is, the success story of Finnish genetics and genomics and the making of the narrative and related imaginary—and its emergence and consolidation as a heterogeneous and multidimensional assemblage, characterized by non-coherence, unexpected turns, and complexity rather than unity and straightforward plans and progress.

In many respects, our book resonates with the studies of national or regional initiatives for biobanking, genome sequencing, or personalized medicine discussed above. What makes the

Finnish case especially interesting is the Nordic welfare-state context within which the shaping of medical genetics as a national endeavour has taken place. Probably the most salient characteristic of the Finnish welfare state is the entitlement of all citizens—and, in fact, all permanent residents in the country—to healthcare, health insurance, and other social services and benefits, including education. Public authorities—the state and regional authorities—have an obligation to provide healthcare services, which are funded through taxation and mostly supplied by public institutions. Public healthcare and social security that cover the whole population are beneficial to biomedical research because they engender vast amounts of standardized, health-related data from practically all Finnish residents as individuals and populations stored in regional EHR (electronic health record) databases and national registers which are maintained and supervised by the public authorities (see Chapters 3 and 4). Furthermore, Finnish people either tend to have quite positive attitudes toward or do not pay much attention to routine data collection by the authorities because they see such data sourcing as necessary for the functioning of the healthcare and social services that benefit them. Such 'data solidarity' (Snell et al. 2021) is seen to manifest among Finnish people as an exceptionally positive attitude toward and trust in scientific research, and a willingness to comply with scientists' requests to participate or donate data, which the Finnish welfare state is seen to facilitate (see Chapters 4 and 5).

In Finland and in the other Nordic countries, universities and scientific research facilities are public institutions and predominantly funded by the state. Consequently, spearhead science carried out in Finland tends to be seen and framed as a national pursuit and servant of the public. In other words, scientific research is supposed to serve a common national cause and bring glory and other benefits not only to the scientists themselves but also to Finland and the Finns. As we demonstrate throughout this book, this ethos of a national science impregnated the development of Finnish medical genetics and genomics in the society.

Scientific efforts in Finnish medical genetics and research institutions have also had primarily national significance in another context. Since the late 1990s, governmental innovation agencies and national innovation policy have keenly focused on and supported genomics and biomedicine. In this setting, the goal of advancing biomedical science became entangled with expectations concerning the national economy and biomedical business opportunities; indeed, the task of becoming and staying competitive was assigned to Finnish medical genomics by the advocates of innovation policy, not only scientifically but also commercially (see Chapter 6).

These characteristics of the welfare state as a milieu for pursuing new genetics—universal public healthcare and a social security system, well-organized health and population data repositories and routine data collection by public authorities, national science in public academic institutions, and national innovation policy's facilitation of the commercialization of research—can be found in all Nordic countries, although there are considerable differences between them. Expert and policy advocates of Finnish genetics and genomics have claimed and underlined that there is one thing that distinguishes Finland from the other Nordic countries: the Finnish population is 'genetically homogeneous', which makes some features of its genetic composition rare and extraordinarily suitable for studying genetic causes and correlates of diseases (see Chapter 2). As we demonstrate, the figuration of 'genetic uniqueness' is a core element in the making of Genome Finland.

In sum, we portray and analyse the patchwork of efforts by which the institutions and interlocutors of biomedical science of a small but quite prosperous nation try to manage—and succeed—in a harsh environment of global scientific and commercial competition. The tale of Genome Finland shares many common characteristics with the genome initiatives in small countries like Iceland, Denmark, Sweden, Estonia, and Singapore, yet the features listed above make the Finnish case specific. Our book captures both these similarities and the specificity of the trajectories

by which biological heritage useful for science was transformed into an asset in global biomedical business in Finland.

Outline of the book

In the chapters that follow, we describe the success story of Finnish medical genetics by looking at one historical trajectory at a time. Chapter 2, 'From Finnish Disease Heritage to genome-wide association studies', traces the narrative of the Finnish population's unique genetic homogeneity as it relates to genetics, and the advantages that Finnish genetic heritage has been seen to offer to international biomedical and genetic research. It also discusses what the homogeneous Finnish genetic heritage has meant for the study of monogenetic and Mendelian diseases since the 1960s, but especially in the 1980s and 1990s. Furthermore, it shows how the research trajectory in Finland has moved from the study of rare diseases in families to population studies of multifactorial diseases and today's research settings, which utilize genome-wide association studies (GWAS), epitomising the change from genetics to genomics.

Chapter 3, titled 'Building up biobanking', discusses Finnish sample collections in relation to the international enthusiasm for the founding of biobanks at the beginning of the new millennium. The main topic of our analysis is the shift from an idea of sample collections to biodata repositories in biomedical data collection and storage. The chapter does not just concentrate on the novelty of biobanks, but also highlights local continuities, especially those concerning the older sample collections on which most of the current Finnish biobank tissue repositories are based. In addition to situating biobanks in the landscape of sample collections and biomedical research, the chapter examines them in the context of another key, early 21st-century trend, namely, personalized medicine. Discussion of biobanks in this chapter also teases out attempts besides biobanking to create coordination and infrastructure for the utilization of many kinds of health data reservoirs and biomedical resources for knowledge production

in biomedicine. In Finland, these efforts have been coordinated scientifically, politically, and economically under the idea of fostering personalized medicine, resulting in several overlapping and even competing endeavours.

The narratives of Chapters 2 and 3 are continued in Chapter 4, titled 'A unique population: Registered, recorded, research friendly', which examines the reasoning and practices behind Finland's portrayal as a unique environment for biomedical research and development. The focus is on the shift from conceiving of the Finnish population in terms of genetics to a view emphasizing population as a health data resource and the willingness of people to participate in biomedical research. The chapter proceeds by demonstrating how additional layers are added on top of each other in the narrative of Finnish uniqueness as a biomedical R&D environment. The starting point is the view of the extraordinary and rare genetic composition of the Finnish population. Often, the advocates of medical genomics associate this with another beneficial population factor, namely, the positive attitude of Finnish people toward research. Our analysis then moves on to data sourcing. As genomics research requires not only samples and participants with a positive attitude, but also their health-related data, Finland has also been perceived to have an advantage in terms of the availability of, and access to, such data. In Finland, there are tens of well-kept electronic national population and health data repositories interoperable through universal personal identification numbers (see Alastalo and Helén 2022), which adds another layer to the uniqueness of Finland as a milieu for biomedical R&D. We end the chapter by showing how, by the 2020s, the whole environment of healthcare institutions, data sourcing infrastructure, and enabling legal and regulatory frameworks became increasingly marketed as a test bed for international collaborators.

'Challenging informed consent' is the title of Chapter 5, which traces the Finnish debates and practices of informed consent and contextualizes them in international developments. The main topic is the readdressing of informed consent as the focus shifts from protection of research subjects' bodily integrity to a

bottleneck for research and development. First, the chapter outlines the principles of informed consent in international medical research ethics and discusses the impact of genomics and biobanks on the debate. The discussion then moves on to the main topic, namely, biobank consent as it was adopted and reformulated in Finland. It was regarded as a pioneering solution for sample collection from individuals, although in practice it has been applied only to a fraction of samples utilized in biomedical research. Against the background of the Finnish consent model, the chapter then provides an analysis of the origins of the consent debate, which demonstrates that historical practices of public health research and register-keeping have framed the approaches to informed consent in Finland. Overall, this chapter offers an analysis of the different paths by which individuals' genetic information becomes part of biomedical R&D infrastructures. It also shows that consent practices have remained anything but clear, uniform, and transparent in the process.

Chapter 6, titled 'Good business?', discusses the commercial aspect of Finnish medical genomics. Building on the analyses and discussions of the previous chapters, it concentrates on assumptions of economic gains and commercial prospects related to biobanks, and research utilizing their interlinked data repositories, such as population registers and patient record repositories. The analysis discloses how national innovation policy became the primary framework for the discussion and development of biobanks and biobank research, and how economic expectations and commercialization activities started to dominate the domain of medical genomics in Finland. In sum, the main topic is the becoming of wealth as the priority of Genome Finland, instead of science and health. Throughout the chapter, the developments in Finland are juxtaposed with similar trends and activities in other countries.

Chapter 7 presents the summary and conclusion of the book. We highlight our main argument that although the vision and even the mission of Genome Finland appear solid, closer examination reveals that the view is both historically and at present

dispersed, consisting of fragmentations, frictions, and contestations, and it looks very different depending on the stakeholder's or observer's perspective. In addition, we summarize what our analysis reveals to be the basic tendencies of constructing and maintaining Genome Finland: a persistent pursuit to consolidate 'one voice' and a unified image of successful and unique Finnish genetics and genomics (see Tarkkala and Snell 2022); almost non-existent public discussion on biobank and genome initiatives and their social consequences; a distinctive appeal to pursue the institutionalization of data sourcing through legislation, which resulted in endless modifications and preparation for legal regulation; and the dominance of economic and commercial expectations of medical genomics into the 21st century. Finally, we discuss whether the expectations and assumptions about Genome Finland's success, assets, and competitive advantage are accurate in the context of contemporary global scientific and commercial competition in biomedicine and the health data economy. We also point out that Genome Finland is coming to a crossroads, which requires from the stakeholders new insight and decisions on which way to proceed.

CHAPTER 2

From Finnish Disease Heritage to genome-wide association studies

Introduction

At the turn of the millennium, Finnish researchers, policymakers, and research funders alike witnessed the emergence of exceptional new possibilities from the genomic research explosion (see Chapter 1). In Finland, the envisioned opportunities were largely based on genetic research, which had been taking place in Finland since the late 1950s and early 1960s, a trend that followed in the footsteps of international research and had begun by looking at blood groups and chromosomal changes. At the beginning of the 1960s, however, Finnish researchers, particularly within paediatrics, noticed some rare hereditary features in Finnish families. This observation would later have a significant impact on the international contributions made by Finnish genetic research.

This chapter tells the story of how the study of rare diseases in Finland expanded into an internationally recognized research field examining disease risk at the population level. Starting with the work of paediatricians in the field of what has come to be known as Finnish Disease Heritage (FDH), we describe the significant impact it had on Finnish genetics in general. From there we move on to discuss what the Finnish genetic heritage has meant for the study of monogenetic and Mendelian diseases, especially in the 1980s and 1990s, and later for multifactorial diseases and today's research settings, which utilize genome-wide association

studies (GWAS). This trajectory serves as a roadmap to a better understanding of how Finnish genomic research has developed. The study of families with rare diseases has provided a historical account of disease aetiology in Finland, which can still be seen in today's genomic research, while the narratives of disease origin continue to play an important role in explaining why the Finnish population is so well suited for studying genetic diseases. This chapter, therefore, describes the development of Finnish genetic research from the early 1960s to its current situation, which focuses on genomics and personalized medicine.

The perspective we adopt in this approach also highlights the transition from clinical genetics and counselling to population-based calculation and research of disease risk in the Finnish context. This transition reflects changes in the relationship between the medical profession and genetics over time, from a field where individuals and families were the primary target of medical intervention (i.e. studying rare, hereditary conditions in individuals and families) to studying disease risk at the population level and then calculating risk scores for individuals as well. This paradigmatic shift has not been without its problems, but as we discuss in other chapters, it reflects broader scientific and societal changes in the expectations and hopes associated with genetics and subsequently genomics; furthermore, it is part of a change in how the medical community and researchers approach the question of disease in general. In this historical arc, we see a move away from patient–doctor relationships mediated by care to a relationship mediated more by a logic whereby everyone is a potential patient who is at risk.

Finnish Disease Heritage

In 1966, a young Finnish paediatrician, Reijo Norio, defended his PhD thesis at the University of Helsinki. Entitled *Heredity in the Congenital Nephrotic Syndrome* (Norio 1966), it was a genetic study of 57 Finnish families who were carriers of the rare, hereditary genetic condition. Almost 35 years later, he published a book titled

Suomi-neidon geenit (The Genes of the Finnish Fair Maiden). The Finnish maiden is a symbolic representation of the Finnish nation utilized since the end of the 19th century to promote nationalism and independence. In the book, Norio reviewed what became known as Finnish Disease Heritage (FDH), a collection of over 30 hereditary conditions over-represented in the Finnish population; some of these can be found elsewhere, whereas others are found only in Finland (Norio 2003a). FDH is not a diagnostic category, but an umbrella term used to describe a group of conditions that tend to fall into one of five broad categories. According to Norio,

> FDH comprises monogenic, mostly autosomal recessive disorders, which are markedly overrepresented in Finland. Some of them have been detected in Finland, and later some scattered cases have been reported elsewhere. In some, the number of known cases is greater in Finland than in all other parts of the world put together. But even 10% of all known patients can be considered as overrepresentation bearing in mind that the number of Finns is less than one thousandth of the world's population and only about 0.5% of the summed populations of Europe and North America. One prerequisite for a disease to be included in FDH is that a minimum of ten families must be known. Any new disorder must be studied properly so as to be sure that it is a homogeneous clinical entity. In many cases, the Finnish clinical genetic community has, by quiet mutual agreement, accepted the disorder into FDH. (Norio 2003a, 443)

One of the criteria for inclusion in the five categories is that the condition is overrepresented within the Finnish population, as there are only a few rare diseases within FDH that are found exclusively in Finland. In fact, it has never been a straightforward process to decide which diseases to include as part of FDH, something Norio himself has noted. In many cases, there is a lack of reliable data on rare conditions from other countries to make such a claim (Norio 2000, 95). In essence, the five categories represent disease groups that have very different backgrounds and causes. For the purposes of the paediatric community in Finland,

however, they have provided important scaffolding on which research into rare diseases could rely.

The work that began in the early 1960s has had a significant impact on Finnish genetics in general. More importantly, the work on rare diseases has also provided an important historico-cultural rooting for genetics (Tupasela 2016), linking the genetic peculiarities of Finns with the migration history of early settlers and their subsequent struggles with famines and diseases. These hardships have, according to geneticists, produced important founder populations and bottlenecks, which according to its proponents, are constitutive of today's Finnish population. We discuss the significance of this to contemporary Finnish genomics in Chapter 4.

Reijo Norio's research in the 1960s, which sought to identify the causal mechanism behind Congenital Nephrotic Syndrome (CNS), became the starting point for a much broader research programme within the Finnish medical community. Norio recounts the decision made by his superiors to send him to contact families that had been identified with the condition. In *Suomi-neidon geenit* he wrote:

> The wise had decided that someone must travel around the country to interview all 39 families affected with congenital nephrosis and who were known at the clinic. The Finnish cause for this disease had to be known, something all these families would share' (Norio 2000, 14, own translation).

Consequently, in 1963 Norio began to study the various families across Finland that had been diagnosed with CNS to gain a better understanding of its causes: meeting and interviewing, but also using Finnish church records to identify family members who were potential carriers. This archival approach proved very useful. Prior to computerization and the development of the Finnish welfare state, church records—some dating back to the 17th century—provided the best source of information on migration to and from regions, and the marriages, births, and deaths within a given parish. The approach of using church records to identify family members and related lineages has also proved very effective

in subsequent studies of rare diseases, as well as other hereditary conditions in Finland. Church records in Finland were the main repository of family pedigrees until 1969, when the population information system was set up, and then digitalized two years later in 1971. For decades before that, Finland had a decentralized system in which the Evangelical Lutheran Church and the Orthodox Church maintained data on their own parish members, while the Civil Register maintained data on individuals who did not belong to a church from 1919 to 1970. The centralization of the registers and their subsequent digitalization had a significant impact on the possibilities that were afforded not just to record-keeping on the population but also to research. This feature of administration in Finland (as well as in all other Nordic countries) would have significant relevance in terms of current research practices using biobanks and healthcare information.

The registers maintained by the churches contained not only valuable information on marriages and births, but often also on the cause of death, which, although not medically based, nonetheless could provide important clues. The following excerpt from *Discover Magazine* in 2005 describes the process of discovery related to Northern epilepsy:

> When Aune Hirvasniemi, a pediatric neurologist at the local hospital, began to track the disease in the late 1980s, she found 19 patients in a handful of families. No one had connected the cases before. Hirvasniemi consulted the records of the Lutheran Church, which for 250 years had written down the comings and goings of Finns in each parish. Creating a medical pedigree for Northern epilepsy, she followed it all the way back to its founder, Matti. She published her discovery of the epilepsy in 1994, the same year that researchers in Finland identified its gene on chromosome 8. (Wheelwright 2005)

The historical descriptions of family movements and intermarriages provided researchers with insights into how these rare diseases came about in different areas of Finland and contributed to elaborating a general theory of rare disease development in

Finland. As more rare diseases were studied, and the causal mechanism was identified as hereditary mutations, the diseases became known as Finnish Disease Heritage (FDH) if they met its general criteria. The term was first introduced to the Finnish medical community in 1972 in a special issue of the Finnish medical journal *Duodecim*, edited by paediatrician Jaakko Perheentupa. A year later the term appeared for the first time in English in the *Annals of Clinical Research* in an article titled 'Hereditary diseases in Finland; rare flora in rare soil' (Norio et al. 1973). Since then, 36 different hereditary conditions have been included within the classification, the incidence of which is quite low, usually in the range of 1:10,000–1:100,000. Although individually they occur quite rarely, as a group of conditions they impact almost 1 per cent of the total births in Finland (Norio 2000, 23).

Over the decades, the number of diseases identified and associated as being part of the Finnish Disease Heritage were visually represented in what has come to be known as Perheentupa's steps (Figure 1). Like Reijo Norio, Jaakko Perheentupa was a paediatrician interested in studying hereditary conditions in the Finnish population; his contribution lay in identifying the gene associated with Mulibrey nanism in the early 1970s. The image of Perheentupa's steps shows both the condition and the year in which the gene associated with the condition was identified. It represents both the development and the identification of FDH conditions in Finland and the substantial work and effort that Finnish paediatricians have put into the study of the diseases which comprise it. The genes and founder mutations of these diseases have nearly always been detected by Finnish researchers.

The corpus of the work surrounding FDH is presented on the FinDis website (FinDis n.d.), which describes the various diseases and their backgrounds. What is important in relation to FDH is the historical and cultural narrative that is provided to describe the birth and development of the various conditions and FDH itself, which has been explained with reference to several compounding causes. The first of these is population isolation; in other words, given Finland's peripheral position in relation to large population

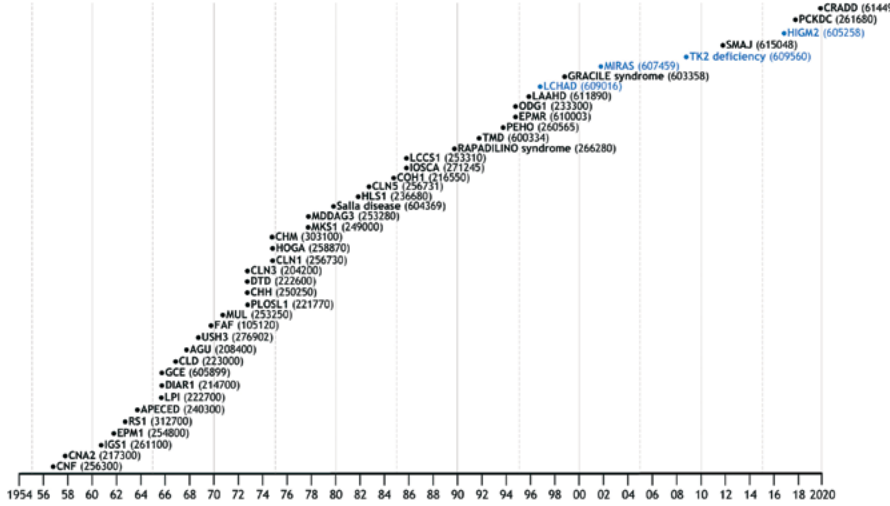

Figure 1: Perheentupa's steps (Uusimaa et al. 2022). The figure is an illustration of when a specific gene related to a rare disease was identified and isolated, thus representing the progression of the genetic discoveries related to FDH. Released under the license CC BY 4.0.

migrations, the number of individuals migrating to or through Finland has been limited over the centuries. The second explanation for the emergence of FDH has been regional isolation, whereby a substantial number of healthy carriers move and live in close geographical proximity to each other, meaning that over the years the likelihood of two carriers reproducing increases. The third is called the founder effect, in which a few mutation-carrying families move to a specific region and thus introduce their gene pool to that area. This has led to genetic drift in several areas in Finland, with specific genetic mutations being carried from one generation to another. All these elements have played a role in the emergence of the various conditions that constitute FDH.

Population and molecular geneticists in Finland who studied diseases at the population level (as opposed to among families) later adopted the historical narratives used to describe the development of rare diseases. Explanations such as isolation, early settler migration, and bottlenecks helped to provide the basis for

understanding the genetic structure of the Finnish population. In an article published in 2000, for example, Leena Peltonen and her colleagues (Peltonen et al. 2000) argued that the study of population isolates was useful in mapping complex traits. The shift from studying isolates and families to studying complex genetic traits of diseases marked a significant turn in Finnish genetics. What is noteworthy, however, was the key role that population history related to rare diseases continued to play in relation to complex traits.

Besides population-level studies of more complex genetic traits, other notable lines of research emerged, including studies of mitochondrial DNA by professor of forensic medicine Antti Sajantila and colleagues (Sajantila et al. 1995; Sajantila et al. 1996), which aimed for a better understanding of the development of European populations. These comparative studies were significant international collaborations which helped to establish not only the place of Finnish genetics research in the international research community but also the notion of the Finnish population as exceptional and homogeneous. In these studies, the 'Finnish population' always represented a distinct entity that was compared with other European populations. The comparative aspect of population genetics and genetic structure was also a significant research topic for the PhD students who came after the paediatricians (Lappalainen et al. 2008; Jakkula et al. 2008), who have continued to utilize Finnish population samples.

Genetic research and the institutionalization of medical genetics in Finland

The work on Finnish Disease Heritage also laid the foundations for the first institutionalized genetic counselling service, which started in 1971 at Väestöliitto (Family Federation of Finland). Väestöliitto was founded in 1941 to promote population growth and the well-being of families with children in Finland; it also had concerns about illegal abortions in the country and about the quality of the national population, in the spirit of pre-war eugenics

(see Chapter 1). In 1951, the Federation founded a genetic counselling board in close collaboration with the University of Helsinki (Meskus 2009, 59–62). The goal was to provide expert advice to the National Board of Health and municipal authorities on issues related to 'eugenic' grounds for sterilization and abortion, as defined in the 1950 abortion law and the amendments of the 1935 sterilization law. In addition, the board provided advice to individual citizens on hereditary diseases and mental illness. In the 1950s and 1960s, the board's genetic counselling remained a 'small scale' activity (von Koskull and Salonen 1997) focused on hereditary diseases (Meskus 2009, 61–63).

Reijo Norio, who had been working with Finnish Disease Heritage, was able to establish a new unit at Väestöliitto in the beginning of the 1970s, at a time when Finnish medical genetics was searching for a clinical home in Finland. Väestöliitto was a suitable host, since it had experience and expertise in services related to genetics through its small-scale counselling, and there were people there who were interested in hereditary diseases. It was therefore institutionally easier to get a rapid start for the new service than it would have been in the national healthcare system and its hospital units. The service differed significantly from the work done previously at Väestöliitto. There was a radical shift in focus away from sterilization and the control of women's reproduction to clinical work, counselling on hereditary diseases, and support of the families concerned. Reijo Norio was the first doctor to head the brand-new clinic concentrating on rare hereditary diseases, particularly Finnish Disease Heritage, with his job as the 'first full-time medical geneticist' in the country starting in 1971 (von Koskull and Salonen 1997, 70).

The Väestöliitto clinic, which grew slowly during its first years, became essential in forming relationships between patients, families, and doctors instead of just issuing expert opinions and assessments (Meskus 2009). The doctors involved participated in international networks, worked at the clinic, and researched the affected families they met through appointments, although, of course, the patients and affected families did not show up from

nowhere. Before the founding of the clinic, and for many years still to come, most clinical genetics and genetic studies were conducted in paediatric clinics where Norio himself had also worked at the beginning of his career. For example, Pertti Aula, who eventually became a professor of medical genetics, founded the first chromosome laboratory at the paediatric hospital in Helsinki and started to do genetic tests through amniocentesis when it became possible (Helsingin Sanomat 2021). Paediatric clinics were often the first port of call for families with FDH-related disease burdens or other rare conditions, since many of these rare diseases presented themselves in early childhood—or even at birth.

Eventually clinical genetics found its way into university hospitals all around the country. In 1972, a year after Norio started to work at Väestöliitto, a chair for medical genetics was founded at the University of Helsinki. The first professor, Albert de la Chapelle, was appointed two years later, and finally, in 1976, the department of medical genetics was established (von Koskull and Salonen 1997, 70). By the time of his appointment, de la Chapelle had already had a profound impact on Finnish genetics research. While still working full-time as a clinician in the 1960s, he had published several important papers with renowned international researchers, including Paul Polani and Ruth Sanger, on sex chromosome abnormalities (de la Chapelle et al. 1964; Harper 2011; Kääriäinen and Aittomäki 2021). His particular interest lay in male chromosomal abnormalities such as Kleinefelter's syndrome. Between 1966 and 1968, he spent time as a post-doctoral researcher at Columbia University, where he was drawn towards work with haemoglobin. The early interactions and communication that he had with international researchers proved to be important in later collaborations and helped to internationalize Finnish research in genetics. The study of chromosomal abnormalities also provided an important base for future screening programmes for conditions such as Down's syndrome (Meskus 2009).

During the 1970s and 1980s, university hospitals around Finland steadily established genetics clinics. The first was in Helsinki, and then came the clinics at Turku and Oulu, which were

followed by Tampere and Kuopio. By the 1980s, the time was right for clinical genetics to become its own special subfield of Finnish medicine, and it founded its own association, the Finnish Society for Medical Genetics (Suomen lääketieteellisen genetiikan yhdistys, SLGY). The society, established at the end of the 1970s by doctors practising medical genetics themselves, had an impact on the founding of the specialized clinical genetics units and laboratories in Finland, as well as on clinical genetics becoming its own specialty in the first place (SLGY 2018a). By the end of the 1990s, there were eight clinics in regional hospitals around Finland offering genetic clinical services (von Koskull and Salonen 1997, 70), meaning that the need for a specific clinic at Väestöliitto started to diminish. As medical genomics developed further, the work surrounding the identification of rare diseases and genetic mutations became easier, with international databases helping in the identification of mutations.

In 2013, the Norio Centre of Rare Diseases was established, commencing a new era for the unit that had its roots at Väestöliitto, as the need for support and services for families affected with rare or even still undiagnosed diseases was identified as persisting, even though diagnostics and clinical care was now mainly done elsewhere. The new Norio Centre combined this support for families and patients with the activities of the Rinnekoti foundation, which had been offering therapy for children with disabilities and once had its own genetics unit as well. Nowadays, the Norio Centre concentrates on the dissemination of information on rare and hereditary diseases and on the education of professionals on the topic. Medical genetics continues as its own specialty, although genomic tests and analyses are increasingly becoming part of other medical fields such as oncology.

The genes of the Finnish Fair Maiden

As discussed above, Reijo Norio's book *Suomi-neidon geenit* (The Genes of the Finnish Fair Maiden) aimed to popularize FDH to the broader public. It also served as the basis for three scientific articles,

which Norio published a few years later in *Human Genetics*. Titled 'Finnish Disease Heritage I' (Norio 2003a), 'Finnish Disease Heritage II: Population prehistory and genetic roots of Finns' (Norio 2003b), and 'Finnish Disease Heritage III: The individual diseases' (Norio 2003c), these articles were directed towards the international scientific community to raise awareness of the work done in Finland on rare diseases. The impetus to publish the three articles came from Norio's British colleague Professor Peter Harper, a doctor and academic, who was aware of the work conducted in Finland and felt it was important that the separate work and articles by the various researchers studying FDH over the past thirty years be collected in a coherent set of publications.

The significance of the imagery of the Finnish fair maiden used in the title of the book suggests that the work on FDH was not just scientific, but also drew on a much broader spectrum of cultural and symbolic imagery. The figure of the young, blonde, blue-eyed woman has strong connotations with the Finnish romantic period from the mid-1800s to the early 1900s, which was characterized by a focus on developing a national self-image with roots in the Finnish language, folklore, art, and architecture. Indeed, the work of artists, writers, composers, and architects helped to bolster and politically develop a particularly Finnish identity rooted in cultural imagery. More recently, technological development, including in medicine and genetics, has provided another way in which national identity can be connected to contemporary endeavours and developments. Concomitantly, the work of Finnish geneticists and paediatricians on identifying both genetic and historical causes of rare diseases has provided an important narrative related to human migration and disease development in Finland. Since the science of genetics drew heavily on historical explanations of human migration and marriage, the two sides—scientific and cultural imagery—have become closely intertwined (Tupasela 2016).

One example of this interlinkage can be seen in the comparisons that Finnish geneticists often make between the Finnish population (and its genes) and the Sampo—an artifact in the national epic poem *Kalevala*—which is akin to a horn of plenty, providing

mythical resources and producing wealth and prosperity for those who control it. The significance of the *Kalevala* is even more instructive as a reference in Finnish genetics, since it represents a collection of Finnish oral traditions made by Elias Lönnrot and his contemporaries during the 1800s. Lönnrot, who was also a medical doctor, travelled widely in Finland and Karelia (a region of eastern Finland, part of which was lost to Russia in the Second World War), where he collected traditional songs and verses from the local people. Later, Lönnrot organized and compiled these different stories to form the corpus of the *Kalevala*, an epic that became one of the cornerstones of Finnish national identity, providing a mythical origin narrative that bonded the nation and people through language and culture. Indeed, the compilations of the *Kalevala* and FDH have many similarities: both have helped to formulate and define an important part of Finland's national identity; both corpora (linguistic/poetic and genetic) have helped to bind the imaginaries of nation and population together through different media. The parallels between Lönnrot and Norio are also striking, as both are doctors who travelled across Finland and visited different parishes to collect stories; Lönnrot's tales were sung, while Norio's storylines were recorded in church archives and decoded from blood samples of family members (Tupasela 2016).

The link between FDH and national romanticism has played a significant role in the contextualization of more contemporary Finnish genomics. After the mapping of the human genome, Finnish genomic research began to see a stronger link between genomics in Finland and its possible contribution to international research. Finnish researchers such as Leena Peltonen began to pave the way to utilizing Finnish genetic resources (including sample and research data collections and registers) in international collaborations that were not just seen to be scientific in nature but also viewed as new economic opportunities that would benefit Finland as a whole. As she wrote in an article in 2004 with her husband Aarno Palotie:

> The information produced from the analysis of the material would most likely have a great impact on the national economy. The achieved results could create the opportunity to utilize funds invested into the Finnish healthcare system to commercializing the new knowledge and even offer the possibility to partially finance the healthcare system of tomorrow. (Palotie and Peltonen-Palotie 2004, 1712, own translation)

The linking of Finnish genetic material and data to the national economy has become a cornerstone of Finnish innovation policy and rhetoric (see Chapter 6). The connection between national romanticism and its modern twist is even more obvious when Finnish biobank collections and registries are described as a Sampo—the mythical machine that produces wealth in different material forms (see Tupasela 2016).

Modern national romanticism was also part of a campaign commissioned by the national innovation-funding agency, Business Finland, and its subdivision Visit Finland, which markets Finland to foreign business. The campaign's core was a composition called 'Symphony of extremes' by a successful Finnish metal band Apocalyptica (2021), described as 'born out of Finnish DNA', as some DNA sequences were made into sounds. The composition and music video were said to unite art, science, and travel in a new way. The commercial significance and its emergence in the Finnish context are discussed in more detail in the following chapters.

Building up national capacities

The significant up-turn of commercial expectations would not have been possible, however, without the significant efforts to modernize and internationalize Finnish biomedical research. As we have already pointed out, Finnish doctors and researchers have engaged in significant international collaboration for decades, a process that became more pronounced at the turn of the millennium. At the same time, support was emerging in the Finnish science and innovation policy for the internationalization of Finnish

research: for example, the Academy of Finland launched a centre of excellence programme to provide more critical mass and support to research clusters (Academy of Finland 1997). Efforts were also made to evaluate Finnish research (Academy of Finland 2002; 2003b) and to develop a more coherent research programme strategy (Academy of Finland 2003a; 2003c).

Finland even made an attempt to establish a Genome Information Centre in Helsinki to support genetics in the country at the beginning of the millennium (Technomedicum 2004). Although it had a rather short shelf life due to lack of backing, during its operation the centre specialized in the use of micro-array analysis and provided genotyping services to research groups all over Finland. Illustrating the rapid development of genetics locally, in 2001 it genotyped 400,000 samples, whereas by 2003 it was already genotyping over one million (Muilu 2004, personal communication; see also Tupasela 2004, 4163). The increase in genotyped samples signified the rapid emergence and up-take of sequencing technologies in Finland and the need to keep abreast of international developments. It also marked the development of a more professionalized approach to sample management and analysis on an increasingly larger scale. The new sequencing technologies would prove even more important later when combining sequence data with other data—from population and healthcare registers, for example—became more prevalent.

Subsequently, the Ministry of Education began its own process of enquiry with the establishment of a molecular medicine, genetics, and epidemiological research institute in 2005 (Opetusministeriö 2005). Both the genome information centre and the research institute suggested that there was a need to develop more critical mass and focus within the biomedical research community, which would also make Finland a more attractive location for international collaboration. The internationalization of Finnish research was an important theme espoused by other government funding agencies at the time, such as Tekes, which was tasked with funding more applied research projects involving commercial partners (Tekes 2003); these focused on specific areas, such

as pharmaceutical R&D (Tekes 2001) and bioinformatics (Tekes 2002). Much like the Academy of Finland, Tekes also sought to improve and bolster the internationalization of Finnish research collaboration (Tekes 2004).

In addition to the development of new funding mechanisms and programmes and the setting up of specific institutes and programmes (see Chapter 3), Finnish legislation was revised to support the new needs of researchers using ever-increasing numbers of samples and associated health and register data (see Ministry of Social Affairs and Health 2007). We discuss these needs in more detail in Chapters 3 and 5, where we look at the legislative changes surrounding biobanking and informed consent in Finland. The role of the study of rare diseases and its subsequent naming as Finnish Disease Heritage cannot be emphasized enough as the foundation upon which Finnish medical genetics in its current state has been built.

The scientific focus of medical genetics, however, moved from clinical work on rare diseases in families to the study of common, multifactorial diseases and even polygenic risk scores (PRS) toward the end of the 1990s. Here it is necessary to say a few words about the fate of FDH during this shift. In 2019, two Finnish researchers, Helena Kääriäinen and Teppo Varilo, published a piece about whether FDH was changing (Kääriäinen and Varilo 2019). They wrote that a slow decrease of FDH-related diseases was to be expected, since nowadays people are more mobile both within the country and between countries, which 'dilutes the genetic isolates' (Kääriäinen and Varilo 2019, 878a), thereby also reducing the incidence of diseases related to isolation. The genetics of the population living in Finland was thus identified as becoming increasingly diverse, and the rare diseases treated at the genetics clinics are in practice becoming increasingly varied as well—not just the rare conditions related to the 36 FDH diseases identified so far. In addition, patients have new mutations that are showing up for the first time in their families, and the diseases do not necessarily follow the family lineage. Therefore, Finnish genetics, and its peculiarities and strengths, presents a new kind

of landscape compared with the 1960s or even the 1990s. A new kind of narrative about what Finland has to offer through its population and the environment and society in which its people live is the topic of Chapter 4.

A recent article paid specific attention to 'genetic mixing within current human populations' in Finland (Kerminen et al. 2021, 4), noting that the rapid urbanization and industrialization which Finland experienced after the Second World War has meant that the previously immobile population has begun to mix more. This mixing, as well as the growing number of immigrants in Finland, has increased the so-called genetic 'noise', which makes it more difficult to identify disease-causing genes with small sample sizes. For geneticists, this has meant that genetic 'Finnishness' and its specific features have diminished, leading to a need to identify only those individuals as genetically 'authentic' Finns whose grandparents, for example, were born in the same Finnish parish in a certain area of the country (Tupasela 2022a; 2016; Tupasela and Tamminen 2015). Thus, the inclusion criteria for genetical Finnishness have remained the same despite the increased mixing of population living in the country.

The Human Genome Project: Opening a new era for biomedical research

After the completion of the Human Genome Project with its first map of the human genome in 2003, one might have thought that Finland was in an advantageous position to participate in this new era. High-quality research had been conducted, the Finnish population structure was known among scientists as 'one of the best-studied genetic isolates' (Peltonen et al. 1999, 1913), and Finnish researchers had built their international networks and reputations. Finland, 'out of proportion for its size, has by example shaped research in human disease genetics' (Kere 2001, 103), declared Juha Kere, a prominent geneticist and researcher, marking out the future. However, it soon became evident that the genomic revolution required bigger collections of samples and more extensive

collaboration between scientists from various backgrounds. Since the new dawn came with new analytical techniques enabling genome-wide association studies and with an understanding of multiple factors behind common diseases, Finnish strengths did not seem to be such powerful currency anymore. The correlations that were now sought in diseases with multiple causes were to be identified and confirmed only from a bigger pool of samples.

The emergent field of medical genomics set forth such requirements that larger tissue sample collections were needed to gain statistical significance. Small collections were not enough to identify weak genetic correlations among thousands and even tens or hundreds of thousands of sequenced samples. The Finnish Genome Information Centre at Meilahti Biomedicum in Helsinki was a manifestation of the need to combine smaller collections to provide larger tissue sample sets for the new science (see Chapters 3 and 6). The Icelandic case had supplied an important blueprint for how genetics and genomics could be studied at the population level, and subsequently numerous large national or regional biobank ventures were initiated, including the UK Biobank. Finland already had data stored from many population-level cohort and epidemiological studies, and mapping commenced on the sample and data collections that had been established—and their purposes. This process helped to identify what was further needed to attain large enough samples and data collections to conduct large-scale genetic and genomic research. We discuss these activities and their results in more detail in Chapter 3, and the challenges this development posed in terms of informed consent in Chapter 5.

Several important projects were launched which reflected this need to combine existing samples and data collections, not just within Finland, but also internationally. In 1994, for example, a large childhood type 1 diabetes study (the Type 1 Diabetes Prediction and Prevention project—DIPP) began. According to its website,

> [The] DIPP study has screened more than 200 000 newborns for genetic risk of T1D and about 17 000 children have participated in the follow-up that includes regular visits, interviews and collection of various kinds of biological samples until the age of 15 or diagnosis of T1D. The recruitment started in 1994 and is still continuing constituting a data and sample repository of dynamic birth cohort that includes a large number of children at different stages of the disease process. (DIPP 2022)

The DIPP project collaborates with numerous international studies on childhood diabetes, such as the TEDDY study, TrialNet and Innodia. A second important Finnish study has been the ongoing Botnia project that is similarly linked to diabetes and focuses on the genetic and metabolic characterization of diabetes. This study, started in 1990, has been investigating 'connections between diabetes risk factors and environmental factors' (THL n.d.), initially in Ostrobothnia and later in other parts of Finland as well as in southern Sweden. Today, the Finnish Institute for Health and Welfare (THL) reports that the collection consists of samples from more than 15,000 donors and is 'one of the largest diabetes family collections in the world'; the materials linked with the original Botnia study as well as its follow-up projects can be found at the THL biobank (THL n.d.)

A third example of the development and internationalization of Finnish genetic research was GenomEUtwin (Peltonen 2003), a four-year project between 2002 and 2006 initiated by two prominent Finnish researchers, Leena Peltonen and Jaakko Kaprio. The project combined samples and data collected on 6,000 sets of twins from 13 countries in the Finnish National Public Health Institute (KTL). The samples had originally been collected for epidemiological research, but with the sequencing of the whole genome they could now be used to investigate genes and gene variations associated with disease. With a specific interest in studying obesity, migraines, cardiovascular disease, and stroke, the study was not just a new venture into scientific research; it also served as an infrastructure template for future work, utilizing a federated

database approach that allowed researchers access to the data from other groups only if they contributed data to the study themselves. This approach also helped to establish the groundwork for European-wide research collaboration on tissue samples using data sharing. Its significance can also be seen in the realization that existing collections were not large enough to detect disease-associated genes, as such detection required exceptionally large tissue collections and data sets to provide analytical power.

GenomEUtwin was just a small example of development that had already begun in broader biobanking efforts in Europe. The need to collaborate internationally, and to standardize, harmonize, and professionalize biobanking in general, was a major impetus in Finland joining the Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) network early in the 2000s. BBMRI (later BBMRI-ERIC, see Chapter 3) is a pan-European network of biobanks that seeks to develop common standards to facilitate the exchange and dissemination of information and tissues between them (BBMRI 2008; see also COGENE 2003; ECVAM 2002). By establishing a national node, Finland was able to constitute itself as an important source of not just samples and information, but expertise in genetics and genomics as well. According to the Finnish BBMRI website in 2015, '[t]he major goal of BBMRI is to develop a research infrastructure that will facilitate high quality research use of comprehensive collections of biological samples and associated data' (see Tarkkala 2019, 124).

The BBMRI infrastructure has also played a key role in establishing a Finnish network of biobanks, which has further helped to internationalize Finnish research. This has brought with it increased attention and interest in technical aspects of standardization and harmonization of data and data management, as well as in ethical, legal, and social issues connected with tissue collection and use (developments discussed in more detail in the following chapters). With the increase in international collaboration giving rise to the sharing and comparing of extensive population data sets, approaches to studying diseases in terms of genetics

were also realigned. Within this broader transformation, the role and function of Finnish uniqueness also began to change.

One prominent example of the use of samples collected in Finland was the way in which they have been deployed in comparative genetic studies of various populations. The use of 'Finnish genes' was not a new concept in population genetics (Cavalli-Sforza et al. 1991), but Finnish researchers were increasingly able to participate in comparative studies. These studies were not focused on studying disease-associated genes, however; rather, their purpose was to reach a better understanding of the development and migration of populations over the centuries by using genetics as the medium of analysis. Notably, a major criticism of some of the projects in which Finnish samples were used came from the social sciences for reifying so-called isolated and indigenous populations (Lipphardt 2014; Whitmarsh and Jones 2010; M'charek 2005; Duster 2005). These critiques pointed out that studies in population genetics were relying on *a priori* notions of populations—mostly political, administrative, or cultural—which might not necessarily have anything to do with genetic relatedness or might inadvertently exclude individuals who are related but not seen to belong to a given population.

One such example is the genetic Atlas of Europe. In an article in *Current Biology* (Lao et al. 2008), researchers from across Europe sought to compare European populations. The study compared DNA samples (autosomal, non-gender-related) from 23 European countries and mapped the DNA differences onto a graph; differences from bottom to top signified genetic differences along the north–south axis, while differences from left to right denoted genetic differences between the east and west of Europe. The relative size of the area allocated to different countries depicted the relative difference within the samples of a given country. Figure 2 is an image from that article that visually represents the differences and commonalities between the different populations. In this mapping, Finns are portrayed as a population that differs significantly from other European populations.

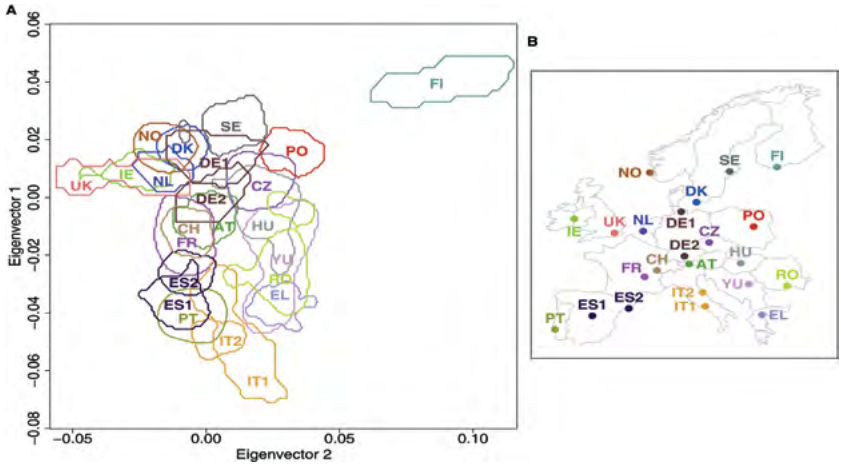

Figure 2: SNP-Based principal component analysis of 2,514 European individuals from 23 subpopulations (Lao et al. 2008). The image on the left shows the relative genetic differentiation between the various sub-populations (described as being small), which correlates with geographic distances between the populations. Finland and Finns being geographically more distant the relative distance to other populations is greater. Published with permission from Cell Press. All rights reserved.

The use of Finnish samples in comparative genetic population studies has not only had an important impact on the internationalization of Finnish research, but has also highlighted some of the challenges with which Finnish research has had to contend in terms of justifying how a unique and different population can also be relevant for other populations. The plot also represents only one way of calculating difference. Other studies have compared, for example, differences in mitochondrial DNA, which have provided different interpretations of relatedness and similarity.

To highlight this feature of comparative population genetics, a team of Finnish researchers published an article in which they compared samples taken from Finns living in various parts of the country, then added samples taken from Swedish donors to see what this would do to the comparison. They plotted their results on a graph, noting the following:

> Interestingly, in the MDS plots the Finnish-Swedes stood out from the rest of Western Finland *only when* Sweden was included in the analysis, which highlights the importance of *relevant reference populations* also when detecting patterns of variation within a country (Salmela et al. 2008, 6; emphasis added).

This observation highlights the way in which differences and similarities can be made obvious depending on what other populations are included in an analysis. Given that there is no common yardstick for comparison of genomic differences, the inclusion and exclusion of samples from different populations will always impact how similarities and differences are made. Compared with the study of 23 European populations, relative distances can change between populations depending on which populations are sampled and compared.

One of the more recent developments in the internationalization of Finnish population and genomic research has been the release of an online search engine that allows people to search for data on sequence variants in Finns. Called the Sequencing Initiative Suomi (SISu), the database allows people to

> examine the attributes and appearance of different variants in Finnish cohorts and see their aggregate distribution in Finland visualized on a map. In the current version users can search for summary data on single nucleotide variants and indels from exomes of over 10,000 individuals sequenced in disease-specific and population genetic studies. The SISu project is an international collaboration between multiple research groups aiming to build tools for genomic medicine. The first version of the SISu search engine was released in 2014. (SISu 2022)

The SISu initiative is another important example of how samples collected and analysed from the Finnish population are being leveraged onto the international research community to provide a tool for analysis and comparison. Looking at the broader perspective, it can be seen that the role and function of Finnish genetic and genomic data has evolved in many respects, reflecting

how medical research is conducted, not just in Finland but internationally too. It also reflects a change in the context in which patients are understood, studied, and treated. While the study of FDH was contextualized by clinical work between treating physicians and families, current approaches focus far more on searching for statistical significance and statistical power in thousands of samples. This shift has also had a significant impact on the role and rights that patient and sample donors are seen to have in relation to medical research.

Realigning Finnish genetic heritage with GWAS studies

Throughout the 1980s and 1990s, the Finnish population had been identified as offering advantages for the study of monogenetic diseases. The benefits of a homogeneous population were utilized, and successful research programmes were built upon knowing these methods and approaches. For example, during the 1990s, the usefulness of studying long linkage disequilibrium related to the homogeneous population isolate had been forcefully demonstrated in the study of monogenetic diseases. At this time, prominent scholars like Leena Peltonen established their laboratories both in Finland and abroad, with professionalized sample processing that piloted the standardized operating procedures that would later come to characterize biobanking.

The discussions on the homogeneity of the population had a particular meaning in the Finnish context. For Finnish researchers, homogeneity means that there is less genetic variation in the samples, which makes certain associations stand out clearly if you know what you are looking for and you already have a notion of the approximate location on which to focus. In practice, this has meant that finding disease-associated genes in the Finnish population has been possible with a smaller number of samples than elsewhere, since there is less genetic 'noise'. Utilizing the advantage of long linkage disequilibrium in a homogeneous population was particularly helpful in mapping genes associated with diseases or

other characteristics, and especially powerful in the localization of disease-associated genes. In 1992, Johanna Hästbacka and colleagues stated in an article in *Nature* that 'Finland represents an ideal population for linkage disequilibrium mapping' (Hästbacka et al. 1992, 204). In these studies, a single disease-associated allele was often sought from a specific area. The researchers stated that 'the relevant question in an isolated founder population is whether there is a single allele with a significantly higher frequency on disease-bearing chromosomes than on normal chromosomes' (Hästbacka et al. 1992, 210). Or, as it was put in *Wired* in 1999 about the homogeneous Icelandic population and its advantages,

> It's significantly easier for genetic researchers to isolate genes in a homogenous population, such as Iceland's, than it is for them to study a more varied population, such as the United States. Anomalies, including disease genes, are easier to pick out when the gene maps of individuals are similar (Philipkoski 1999).

This same logic applied—and still applies—to Finland. For example, in 1999, a group of prominent Finnish scholars wrote that they had efficiently used 'special strategies taking advantage of linkage disequilibrium', especially in the 'mapping and restriction of Finnish disease loci' (Peltonen et al. 1999, 1913). These studies not only provided information about Finnish Disease Heritage, but also suggested wider international relevance. For the Finnish research community, the international relevance of Finnish collections has been important, since it provides a vital gateway to international collaboration, meaning that Finnish samples and data had to be made relevant for international collaborators. As Juha Kere, a noted Finnish geneticist, wrote:

> A population of about 5 million at the northern corner of Europe is unlikely to arouse the attention of the human genetics community, unless it offers something useful for others to learn. A combination of coincidences has finally made this population one that, out of proportion for its size, has by example shaped research in human disease genetics. (Kere 2001, 103)

In one paper, researchers conclude that they have not only gained knowledge about Finnish disease loci, but also about 'biological processes and metabolic pathways essential for normal development and function of human cells and tissues' (Peltonen et al. 1999, 1913). This last part exemplifies how the wider relevance of results gained in a Finnish population is always needed to match the population material with the broader knowledge needs of biomedical research and its current interests, technologies, and methods.

Further important advantages that Finnish collections and research have had in the international research community are speed and efficiency. When presenting some of the advantages of Finnish genetics for analytical methods, Leena Peltonen (1997, 554) mentions efficiency. In Finnish population data, the sufficient number of samples for meaningful research analysis is often lower than with other study populations; thus, the research techniques utilized in the study of FDH were already seen as promising in the 'search for complex, polygenic diseases' also (Peltonen 1997, 555).

Over the years, Finnish genetics scholars have put a lot of effort into framing their homogeneous population as offering great potential for biomedical research and enabling the achievement of results of more general relevance. Despite this, the Finnish homogeneous genetic heritage, backed up by a small population size, has not unanimously been considered only an advantage. With the 21st century, genome-wide association studies (GWAS) arose as a model for studying disease in terms of genetics, and interest in monogenetic diseases has been gradually fading. Both developments worked against the advantages that homogeneous populations with long linkage disequilibrium had to offer, although Finnish researchers continually emphasized the wider implications of their results in their papers. Thus, at a time when Finland started to get its sample collections coordinated to make them more widely usable by international biobank networks (see Chapter 3), the need to convince international biomedical audiences of the continued relevance of Finnish samples became ever more urgent. This meant that the sample collections and populations

had to be framed as not being too idiosyncratic to be included in international collaborative efforts, pooling vast numbers of samples from different populations. In order to fit in, the results based on Finnish samples needed to apply to other populations too (see Tarkkala 2019; Tarkkala and Tupasela 2018); status as merely 'an obscure outlier' (Tarkkala and Tupasela 2018) would exclude Finns from international collaboration. This, in turn, would hinder the development of scientific research in Finland and, in the worst case, compromise the health benefits that Finnish people could get from these studies in the long term. The uniqueness and idiosyncrasies that were originally identified in relation to FDH now had to be increasingly downplayed to remain internationally relevant in light of the new GWAS methods and their requirements.

In recent years, although a lot of emphasis has been placed on the need for large quantities of samples and for Finnish samples to fit into these bigger, pooled sample collections, there have also been other attempts to frame the use of this population material in research. Some explicitly highlight the specific characteristics of the population and concentrate less on showing what a good fit the material is with other populations. Research papers were published during the 2010s to convince readers that using the Finnish population can be both efficient and cheap in certain research settings, despite the genetic homogeneity. The same cost-efficiency argument has been deployed as before, as it is claimed that by using certain techniques, full whole-genome sequencing can be avoided. Instead, samples are only genotyped, after which mathematically imputed data can be added to cover the areas not included in the selective genotyping itself. Using statistical imputation to calculate and fill the information gaps would, therefore, equal fully sequenced genomic data. This saves a lot of time and money, as homogeneity allows for this sort of imputation, thereby hinging operations on the Finnish genetic heritage, which allows such mathematical modelling to be done accurately (Tarkkala 2019, 62). This emphasis, however, simultaneously locks the idea of the Finnish population to the people descending from the

original bottleneck founder population that carries the homogeneity allowing imputation.

Another potential advantage hinging on Finland's genetic homogeneity was foregrounded and advertised in the late 2010s and early 2020s. The so-called 'loss of function' gene variants found in the Finnish population were identified as potential targets for drug development, as it is believed that through loss of function variants one can learn why some people remain healthy despite being genetically susceptible to a certain disease (see e.g. Lim et al. 2014). This, then, could show researchers what a potential drug should target to keep those people healthy. These individuals might share the same genes or some of the same genes, but do not have the protection of loss of function variants which are believed to offer a direct route into what medicine should address and tackle to keep others healthy as well. This approach is emphasized in the FinnGen study, a big national research initiative, taking place in Finland from 2017 onwards and aiming to utilize and collect 500,000 samples.

On the FinnGen homepage, it is stated that 'thanks to the genetically unique Finns, genomics data is faster to analyse, and the probability of findings is higher than in genetically heterogenic populations' (FinnGen 2022a). The same strategy has been utilized in Iceland, where 'instead of picking a disease or disorder and then fishing around in the genome for significant variants, scientists can first identify a genetic slip-up and then see exactly what happens in humans who have it—just like in a lab, with mice' (Palmer 2015). The drawback of this approach is that what you are looking for must be present in the specific population, and small populations have less variation; if the variant does not occur in the population at all, data from it obviously cannot be used for the study. To put it another way, increased variation in the population under study increases the questions that can be asked and the settings in which the answers can be sought.

The homogeneity of the Finnish population is not always straightforward, as discussed earlier. Even though Finns can be described as genetically homogeneous, and this claim is well

grounded in the scientific literature, the opposite is also true. Inside this homogeneity, Finns are very different. Heterogeneity follows from the strong division of the population into western and eastern groups, and when these are compared with other populations used in scientific studies, western Finns might be closer to Swedes, for example, than to eastern Finns (e.g. Salmela et al. 2008). Moreover, research populations are always formed based on very specific practices and criteria, which means that the population designated as 'Finnish' in many scientific studies does not include, for example, Samí, Roma, or immigrant populations. Currently, while the Finnish sample collections are promoted through the lens of a homogeneous population and the efficiency this brings to research, in practice, samples and data are collected for research in Finland from all those inhabitants receiving services in public healthcare. Consequently, the current collections include samples from individuals who are not considered part of the homogeneous Finnish population. The population that has been genetically carrying FDH will not remain the same forever, as the genetic make-up of the inhabitants of Finland is changing and becoming more heterogeneous as a result of internal and international mobility.

Conclusion

When looking at current developments in large-scale genomic research in Finland, one needs a sound understanding of the history and background of medical research in the country to contextualize the slow yet steady growth and internationalization of its research. In terms of Finnish medical and biomedical research, this has also involved a slow move from clinical research with a small number of families with rare monogenic disorders towards large-scale genomic studies using hundreds and thousands of samples and related healthcare data. An important part in this development has been the institutionalization of medical genetics and counselling around Finland and the role that the study of FDH has played in that development.

The change in scale and focus has had a significant bearing on the relationship between the medical research community and patients and sample donors. Current trends in big data analytics do not seek clinical validity but rather statistical significance from large quantities of data. Although work with families with rare conditions continues, the focus of large-scale genomics projects in Finland and elsewhere has sought to realign research agendas towards personalized medicine. We discuss the impact and significance of these changes in subsequent chapters, since they play a key role in the development of Finnish genomics.

The rise of Finnish genetics through the specialty of paediatrics and the study of rare diseases has also played an important part in making genetic research into Finnish genes and heredity a symbol of national identity and origin. Notions such as genetic romanticism (Tupasela 2016), evolutionary nostalgia (Oikkonen 2018), and tethering (Hinterberger and Porter 2015) have been used to describe the different ways genetics and genes are used to link the present to the past. This linking can work in many ways, but in the case of Finnish population genetics, it has served to provide a level of national authenticity and provenance through which Finnish researchers can claim authenticity and uniqueness. As we have discussed above, however, this uniqueness has required negotiations whereby Finnish researchers have needed to show that despite their uniqueness, Finnish genes have relevance for international studies as well (see also Tarkkala and Tupasela 2018).

CHAPTER 3

Building up biobanking

Introduction

Biobanks are often defined as collections or repositories of human biological material such as tissue or blood samples, as well as personal data associated with the samples. Of course, sample collections existed before institutionalized biobanking; individual researchers, research groups, and institutions have been collecting tissue samples for both research and diagnostic uses for decades. Towards the end of the 20th century, however, the belief emerged that collecting samples solely for the use of one research group or project was an inefficient and outdated way to work in the life sciences. Instead, samples needed to be made available for the wider research community and various research uses in biology and biomedicine. For this, they needed to be collected and stored in bigger collections together with their associated data. This idea led to the founding of biobanks, both internationally and in Finland, at the turn of the millennium and in the decades that followed.

Within biomedical research policy, biobanks were expected to become an essential resource and infrastructure for biomedical research and development, and especially for human genomics and personalized medicine. Thus, from the beginning, biobanks were tied to the development and expectations of genomics and how genomics can transform medicine. In Finland, policy discourse on biomedical research also identified biobanks as crucial elements in making the new era of medicine become reality. As early as 2004, Finnish experts wrote that biobanks are 'at

the centre of this whirl of change', referring to the ongoing rapid development in genomics (Käpyaho et al. 2004, 6). Today, 'whirl of change' continues to describe the landscape of genomics and biobanks in Finland.

In this chapter we discuss this twisting and swirling by exploring how biobanking was officially established in Finland as an institutional practice in the 2010s. Finnish biobanks did not start from scratch; rather, they were built on previous sample collections and on the existing notion of the special nature and offerings of Finnish research cohorts and collections. At the same time, the biobank samples and the way samples are organized and made available are also part of national and international continuities, both historically and today. This chapter describes how biobanks and their sample collections were constituted as national flagships and integral parts of the success story of genomics and biomedicine in Finland that has been in the making in recent decades. It demonstrates that ideas about the nature and purpose of biobanks and biobanking are not fixed in this constellation. The justifications for biobanking, as well as its role and aims, have been adjusted constantly to new situations and developments in research and innovation policy, legislation, and science and technology. Biobanks have also been connected to various other developments in the field of genomics, as they have been seen to provide a necessary infrastructure and service for research. Even the role of biobanks as the flagships of Finnish genomics has been contested by other visions and rapid developments in the field—especially those related to the growing capacity to generate and analyse genetic data from samples and connect information from samples to other types of data, such as vast register data. Thus, instead of becoming the national spearhead of medical genomics, biobanks have become only one part of a bigger constellation of health data infrastructure.

The shifting focus from the tissue samples themselves to data is also an essential element and development tracked in this chapter. The samples in biobanks have increasingly been understood as sources of data, and biobanks have been developing into data

depositories instead of institutions collecting and storing biological samples in their freezers, which at first glimpse might seem to have been their main task. Although samples can be analysed and turned into data in many ways, biobank data most frequently refers to genetic or genomic data. The idea of biobanking is cumulative, meaning that the data resulting from an analysis is returned to the biobank to be used by other researchers. Indeed, the research of today does not necessarily need the 'wet samples' if data on them is already available (Tarkkala 2019). Moreover, the samples—whether already analysed or as material—are potentially usable for research only when accompanied by information on the sample donor, which adds another layer of data to the picture. Sample-related data might, for example, contain diagnoses, personal details such as the sex and age of the person, medicines received, or information about disease outcomes.

Collecting samples, collecting data—a very brief history

Biological sample collections are not new entities as such. Pathological and anatomical collections date back to the 18th century (Tybjerg 2015), and the oldest of these might still be portrayed in medical museums. These collections have been repositories of knowledge of their own time, as are the current ones; however, not all collecting has taken place in the contexts of museums or clinics, nor has its sole purpose been portrayal, preservation, or exhibition. Throughout the 20th century, researchers have gathered different kinds of samples and information for their own research purposes, which are collections of materials meaningful for answering the specific questions at hand and managed by individual researchers or research groups. Samples have formed the core of the collections, but many of these have been accompanied by data, such as family lineage and incidence of diseases, as an essential component. A well-known example of this is the collection of samples and data from Family G, a German immigrant family in Michigan, which includes a detailed cancer genealogy

dating back to 1895 with samples that helped to reveal Lynch syndrome, indicating a particular susceptibility to cancer (e.g. Kay 2019).[5]

There is a long tradition of collecting samples for research purposes and for the needs of genetic research in Finland (see Chapter 2), customs and practices that have directly paved the way for current Finnish biobank collections and biomedical research. Finnish biomedical research was successful in studying and finding associations in monogenetic diseases, as the discussion on the Finnish Disease Heritage (FDH) in the previous chapter has demonstrated. Record-keeping of the population and individuals dates to the early 18th century when Finland was part of Sweden and the Swedish state obliged every parish in the kingdom to keep records of the births, deaths, and marriages within its jurisdiction, and arranged the first nationwide census. In the late 19th and early 20th centuries, collection and storage of such data became more extensive and systematic with the establishment of the national statistical office, which started regular censuses in major towns and conducted statistical investigations of various social and economic topics, health and healthcare included. During the first three decades of the 20th century, the first health and medical registers were also established, related especially to contagious diseases and maternal and infant health, while nationwide health inspections for schoolchildren piled up further data to be registered. All this had political and administrative goals, because the ostensible purpose of data gathering was to serve the efforts of the state and other public authorities to maintain the population in good order, including initiatives and reforms to improve public health.

The long tradition of state-governed, systematic, comprehensive, and routine data collection from individual citizens, residents, and clients of the public services is seen as a distinctive characteristic of the Nordics (Snell et al. 2021; Tupasela 2021). Ingrained in local statecraft, these practices became even more intense in all the Nordic countries from the 1950s to the 1980s, as the public authorities and associated researchers were collecting

data on people's lives during that period to be used in the construction and maintenance of the welfare state's institutions and services: pensions, social insurance, education, and healthcare, among others. In Finland, a major boost to establishing systematic national registers and statistics facilitating the planning, provision, and administration of welfare services came in the early 1960s with national pension reform and the introduction of universal health insurance. These reforms required systematic collection, storage, and management of data on a much larger scale than before, because the data now needed to include information on every citizen, resident, and client of services in the country. This development enabled the introduction of the personal identification number (PIN), which allowed universal identification across all the services. Besides the PIN, the transformation of the data into electronic form between the late 1960s and the 1980s enhanced the coverage and usability of Finnish public registers and databases (Alastalo 2009; Alastalo and Helén 2022).

Alongside the administrative needs and rationales of the welfare state (Alastalo 2009), the extensive coverage of the Finnish registers and the abundance of their data provided opportunities for a wide range of research, both scientific and administrative. The PIN further expanded research possibilities because, as a universal identifier, it enabled detailed population-level studies across multiple administrative sectors and databases (Alastalo and Helén 2022), the results of which have been available and widely used in Finnish public health, social science, and demographic research since the 1970s (Gissler and Haukka 2004). These well-ordered repositories, run by the public authorities, also supported Finnish medical research—from epidemiology to biomedicine—by providing a variety of data about, for example, causes of death, medical records, prescriptions, cancers, and related information on employment, pensions, or income support, family relations, housing, and so on. The PINs are used in other Nordic countries as well. Their use in identifying individuals and connecting their data is framed as a special strength not only in Finland, but, for example,

in Norway, Iceland, and Denmark too (see Tupasela 2021; Brumpton et al. 2022; Holm and Ploug 2017; Tarkkala 2019; Rose 2003).

From early on, Finnish human genetics benefited from the state-driven collection and storage of population and health data. Thus, the rise of genetics and other biomedical fields from the 1970s onwards was closely tied to the national modernization project of establishing a Nordic welfare state in Finland (Tupasela 2016). In this respect, Finland was like other Nordic countries. Building up the welfare state and its data sourcing practices and institutions also provided a crucial backdrop for many research cohorts and clinical sample collections that biomedical research was to utilize.

Western medical science faced an epidemiological turn in the 1950s, spearheaded by extensive projects for studying the prevalence of cardiovascular diseases (CVD) and related morbidity in various populations (Aronowitz 2011; Giroux 2011; Jauho and Helén 2018). Finnish researchers also jumped on this bandwagon, taking part in the Seven Countries Study, one of the most prominent projects in comparative epidemiology on CVD since the late 1950s (Karvonen et al. 1994; Jauho 2021; Jauho and Helén 2018). The exceptionally high CVD morbidity in Finland was of interest to international scientists and of considerable domestic concern—a concern that motivated public health action and directed medical research on the problem. Consequently, domestic epidemiological research on CVD and other common diseases became entangled with the building up of public healthcare and health insurance as part of Finland's welfare services. For example, the Mobile Clinic Health Survey was launched in the late 1960s to serve the planning of public healthcare reform and consisted of samples from over 60,000 research participants from 1965 onwards. The North Karelian Project, an extensive health promotion and study endeavouring to further CVD prevention, sparked the launch of FINRISKI as a nationwide follow-up epidemiological study. FINRISKI was executed regularly every five years between 1972 and 2012 and has collected a cohort of 38,000 participants covering a 40-year period. These studies are milestones in public

health research and instrumental in the improvement of public health and health promotion in Finland. In addition, they also started producing sample and health data collections that, stored and managed at the National Public Health Institute (KTL), later became a precious resource for research.

Thus, KTL and its successor, the Finnish Institute for Health and Welfare (THL), are integrally tied to the history of collecting samples from Finns. THL was formed in 2009 when two governmental research institutions—KTL and the National Research and Development Centre for Welfare and Health (Stakes)—were merged into a single organization, meaning that many of the national registers containing health and social service data are now controlled by THL. Apart from these large institutional sample and data collections and research cohorts at KTL/THL and various universities—such as the 'Twinstudy' cohort from 1975 onwards, which was collected with the University of Helsinki and consists of longitudinal data from over 14,000 research participants (e.g. Käpyaho et al. 2004)—many researchers followed a more conventional and less centralized paradigm and collected samples for their own use in line with the topic on which they were personally working. These smaller sample collections were in the hands of relatively few researchers, with access granted to their own research groups or collaborators. This was exactly the kind of practice that was deemed inefficient and of inconsistent quality in the arguments supporting the movement towards larger, biobank-like depositories.

Another context in which samples had been piling up were the university hospitals and hospital districts around the country. Over the years, the pathology departments had collected diagnostic samples that could also be potentially interesting research materials. According to a report by the Ministry of Social Affairs and Health: 'There are millions of samples taken for diagnostics, care or causes of death at the pathology department. There are samples from the end of 19th century onwards, and the amount is increasing by hundreds of thousands annually' (Ministry of Social Affairs and Health 2007, 14, own translation).

In terms of the discussion in this chapter, it is important to note that biobanks are built on two elements: first, on already existing sample collections and their continued use, and, second, on new ideas that have expanded the scale of sample and data collection operations, reorganized the collection and storage of the samples, and reformed the access policy. For the latter, enabling interplay between research purposes and the institutions of the welfare state has been a crucial prerequisite, which we discuss more thoroughly in Chapter 4. In this framework, the Finnish biobanks have turned the access to research materials into a new kind of service and infrastructure for biomedical research.

New value and initiatives from old sample collections

Towards the end of the 20th century, a consensus emerged in the research community that many diseases are multifactorial and genetically complex. To understand them, big sample collections would be needed. A single sample collection by one researcher or one institute was no longer considered big enough for contemporary research. Instead, it was clear that samples would have to be pooled from multiple collections—locally, nationally, and internationally—thereby stimulating international interest in big sample collections, which would also entail research becoming international and collaborative. This was important, as it was becoming increasingly difficult to publish research findings if they were based on only one cohort or population. International journals began to require that findings be validated in other population groups as well. The new landscape was outlined by medical geneticist Juha Kere in an editorial in the journal of the Finnish Medical Association:

> It is clear that the value of sample collections associated with extensive and reliable data has increased, but at the same time the value of individual data in terms of research has decreased. Nowadays, it is difficult to publish gene association studies concerning

> multifactorial diseases, unless the conclusions are based on combining several different sample data, preferably from different countries. … Thus, both clinical-epidemiological expertise and international cooperation are emphasized in current research. (Kere 2007, 864, own translation)

In this context, the gaze was turned on already existing depositories of both samples and data and the promise that lay in combining them, triggering reorganization in many countries to make these resources available efficiently. The founding of Iceland's biobank in 1996 had made headlines and become a benchmark in the field, as Iceland had offered the private company deCODE Genetics monopoly access to population and national health data collections and genealogies dating back centuries. Around this time, the word 'biobank' emerged in the scientific literature (Hewitt and Watson 2013), along with DNA banks, genetic banks, and databases. Biobanks came to be understood as vast, necessary sample pools with well-characterized data for establishing associations between diseases and genomic profiles—vitally important for advanced biomedical research (Zika et al. 2010, v). The field of biobanking itself also started to show signs of professionalization and disciplinary clarity; new journals were established, such as *Cell and Tissue Banking* in 2000 and *Biopreservation and Biobanking* in 2002, and international societies were founded, including the International Society of Biological and Environmental Repositories (ISBER), which was established in 1999.

In this kind of vision, then, samples would be made easily available to researchers through organized, well-governed collections called biobanks. This positioned samples as a resource for research that could be obtained through a service provided for all researchers in an efficient and open manner. One would only have to contact these sample providers, which would then deliver the needed research materials, instead of a researcher or research group having to collect everything personally. The biobanks would have standardized collections, dedicated professionals, and suitable liquid nitrogen freezers where samples would be stored

at a consistent temperature. This clear and consistent model of operations based on standardized operating procedures was from the beginning also identified as a specialty of professionalized biobanking in Finland (Tarkkala 2019).

Establishing national and regional biobanks represented an opportunity to stay in the game in international research and to be an internationally interesting partner, and, naturally, biobanks were an important resource for the study of diseases. Along with deCODE Genetics in Iceland, which had operated since 1996, among the first biobanking infrastructures were the UK Biobank in the United Kingdom, and CARTaGENE in Quebéc, Canada, both of which were founded in 1999. The Estonian Biobank followed soon after in 2001 and the Trøndelag Health Study (HUNT)—a large population study in Norway running since 1984—became known as a biobank in the early 2000s. The establishment of biobanks was not, however, solely connected to the needs and expectations of biomedical research. Biobanking was also associated with wider ambitions, tying the needs of the life sciences to the expectations of economic gains resulting from biomedical innovations in fields like pharmacogenomics and personalized medicine.

This new era placed genes and genomics within the purview of business and national economic development, a perspective strongly promoted by international developments and several feasibility studies and reports that sought to elaborate on the new model and its role in conducting genetic and genomic research. Consequently, the founding of biobanks at the beginning of the 2010s was not the first attempt in Finland to organize sample collections and data for research and business. For example, during the early part of the 2000s, the idea emerged of developing a national Genome Information Centre in Helsinki whose goal would be, in part, to commercialize the new findings being made in genomic research.

The uses and storage of genetic material were also discussed in parliament's Committee for the Future, which made a statement about the 'social and legal challenges of human genome and stem cell research' wherein various aspects of the human genome

were discussed, including how it could be stored and utilized in Finland. The idea of the Genome Information Centre was also mooted. The committee recognized the importance of genomics to economic growth, but emphasized the need for public discussion and acknowledgement of the interests of Finnish people.

> The Committee for the Future is of the opinion that investigation should be carried out to discover a solution which best ensures the reasonable use of internationally unique genetic material already collected and the further collection of this material in a way that is of best use to all Finnish people. Discussion on the issue should be continued with expert talks and public debate, enabling thorough examination of ethical issues related to gene material collection and its use. A discussion report should be drawn up for this purpose, examining the various views affecting the founding of a Genome Information Centre in a way that is easily understood by the public. (Kuusi and Parvinen 2003, xvi, own translation.)

The blueprint for the centre was sketched in a feasibility study published in 2004 (Käpyaho et al. 2004). It exemplified the idea of Finnish tissue material and data becoming resources, which can be used to collaborate and engage with both big and small pharmaceutical companies. The Finnish Genome Information Centre would have worked as a non-profit centre facilitating the translation of new information to the commercial sector. This model does not clearly explicate how commercial value would be generated except through the general idea that commercial actors would develop new products. The proposal is, however, representative of the thinking regarding national resources and commercialization which was becoming increasingly pervasive in Finland (see Chapter 6). It is also one way of imagining the reorganization of these resources for new purposes and for new kinds of uses and users (Tupasela 2008).

Not everyone was in favour of the plan. Many researchers objected to the centre's being located and run by researchers in Helsinki, while some noted that the expectations were simply not realistic. For example, renowned geneticist Petter Portin

commented that 'in principle the plan is worth supporting, but it is too grandiose and directed too much towards the production of economic profits' (Portin 2005, 39). Despite the expectations, the proposal and development of the centre fizzled out and disappeared from the policy radar. While this was due in part to opposition by the research community, the scientific findings also increasingly suggested that the one gene/one disease paradigm was fallacious and that the causes of diseases and other medical conditions were far more complex and difficult to patent and commercialize than originally thought (Tupasela 2006a).

In another example of proposed infrastructure, the Institute for Molecular Medicine Finland (FIMM) was founded in Helsinki in 2007 to match the European Molecular Biology Laboratory (EMBL) concept. The aim was to establish a specifically Nordic version of EMBL, whose strengths, according to an Academy of Finland report (2003c, 16), would lie not only in the combined population of the Nordic countries' 24 million inhabitants but in their genetic characteristics, population registries, and databases as well. Importantly, this institution and its founding were seen as necessary to foster the internationalization of Finnish biomedical research, improve its attractiveness and competitiveness, and build infrastructure in the country to 'provide researchers with better resources for working in an international environment' (Academy of Finland 2003c, 16). Funding and its continuity were, nevertheless, insecure, which seems to be an ongoing companion of these projects and openings and has also been an issue for Finnish biobanks and their continued existence.

Although health, both of the individuals and the population, remained a key factor in justifying biomedical research, the argument about economic benefits was intensifying. In an article written by project manager Kirsti Käpyaho and colleagues (2004) on the economic potential of genomics, it was suggested that Finnish resources should be put to better use and their commercial potential exploited. Otherwise, taxpayers' money was not being well spent, and resources were being wasted. Many of the arguments and opinions which began to be expressed during this period

reflect a strong moral reasoning behind funding biomedical research that emphasized the role genomics and commercialization would play in saving the Finnish economy. This perspective was not, however, adopted unconditionally by the research community. Some researchers felt that the emerging plans promoting the link between genomics and business were inflated (Tupasela 2006a).

As mentioned earlier, the Icelandic example became the embodiment of expectations about possible scientific breakthroughs and innovations on the road to more personalized medicine and the application of genomics in medical care. While the first version of the Finnish Genome Information Centre (presented above) was being planned, the biobank in Iceland had already been founded. DeCODE Genetics—the company with exclusive rights to study Icelandic genetics—and the Icelandic Health Sector Database (HSD) planned alongside it, were attracting media attention (Berger 1999; Chadwick 1999; Gulcher and Stefansson 1999). Many popular newspaper articles celebrated the opportunities offered to genomics by the study of a whole nation (e.g. Philipkoski 1999). However, the founding of deCODE Genetics and plans for the HSD also raised concerns, controversy, and debate, particularly over the privacy of Icelandic citizens and the ownership of the collection and data. The latter became especially pronounced, since the deCODE biobank has had bankruptcies since its founding and therefore also several owners over the years (e.g. Reardon 2017; Pálsson 2008, Fortun 2008). This has created wider questions about whether national DNA should be an object of commercialization and what might be the consequences.

As early as 1999, a newspaper article in *The Washington Post* began with the worrying statement: 'Iceland has decided to become the first country in the world to sell the rights to the entire population's genetic code to a biotechnology company' (Schwartz 1999). These kinds of issues and tensions between private and public, health and wealth, privacy and openness, scientific research and innovations, have followed biobanks and the use of genomic and health data ever since. The tensions between

research requirements, the distribution of benefits, and the rights of the people whose samples and data are being used have remained an ongoing concern without easy solution, as we show in Chapter 5.

To sum up, despite the problems, and in line with international developments and optimistic expectations, Finnish researchers, research organizations, and public funding bodies increasingly saw national and regional sample collections as biobanks at the turn of the millennium. The epidemiological cohorts and pathological sample collections were perceived as a national resource that could facilitate the development of scientific discoveries and new innovations, especially in concert with other Nordic countries (Academy of Finland 2003c). All these developments finally contributed to calls to coordinate and organize Finnish samples—those in both clinical hospital collections and research institutions—into actual, modern biobanks.

From sample collections to modern biobanks

In the early years of the 2000s, many prominent researchers had already written in different forums about the need to advertise and utilize already existing, internationally exceptional, Finnish sample collections and to unite them by forming them into biobanks (e.g. Palotie and Palotie-Peltonen 2004). Finnish collections were listed and presented in reports and documents on genomics, and the numbers of samples and participants were counted to show their size and potential (e.g. Halme 2005; Käpyaho et al. 2004, see Figure 3).

From a terminological perspective, Finland's approach to what constitutes a biobank is unique, with 2013 marking a clear watershed between two eras. Before 2013, when the Finnish Act on biobanking came into effect, any collection of tissues could be considered a biobank. In fact, existing Finnish collections of samples were often described as biobanks, both by Finnish actors and in international reports (e.g. Zika et al. 2010; National Biobanks 2012). For example, in 2006 a news piece on the founding of

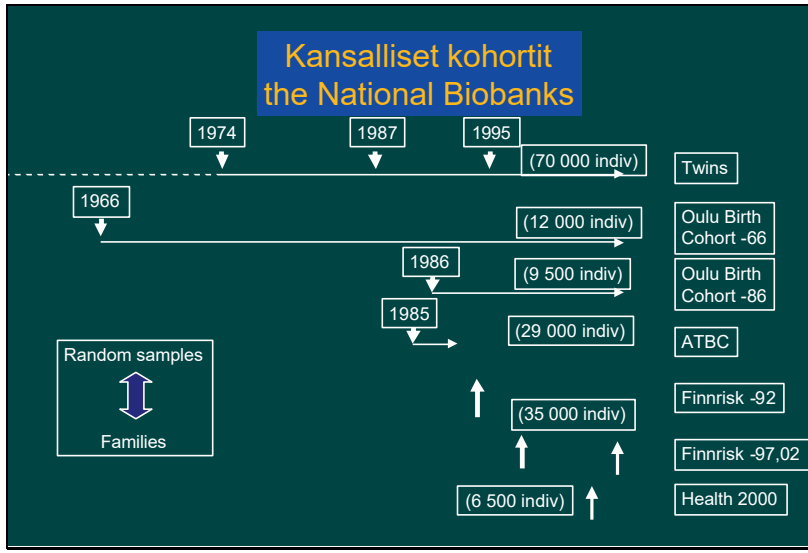

Figure 3: Kansalliset kohortit—the National Biobanks. In this slide from a presentation by leading Finnish medical geneticist Leena Peltonen in 2008, the title reads 'national cohorts' in Finnish, yet is translated to 'the National Biobanks' in English. This peculiar translation from cohorts to biobanks already hints the change in envisioning previous research cohorts now as resources more broadly. It also underlines that conceptually biobank as a term was in Finland at this point more flexible than after the Biobank Act.

Slide source: Tutkas 2008. All rights reserved.

biobanks in Finland noted that biobank activities at a population level were already underway, as KTL was collecting serum samples from pregnant women and had been doing so since 1983 (Suvilehto 2006). In a European report from 2010 that surveyed over 170 biobanks in Europe (Zika et al. 2010, 42–43), five Finnish biobanks were identified, with the role of KTL particularly highlighted:

> All samples collected in KTL projects are stored in a centralized biobank, which today contains DNA and serum/plasma samples from 200,000 Finns (approximately 5% of the population). This biobank constitutes an important national resource, increasingly

> used in international research projects in which Finnish researchers from KTL and other institutions are involved. (Zika et al. 2010, 43)

Thus, the existing collections and practices were at times regarded as biobanks and as matching with the general descriptions and definitions of biobanks that were circulating internationally. With the introduction of the national Biobank Act, however, biobanking became legally institutionalized. A tissue collection could only be called a biobank if it had been granted an official biobanking permit by Valvira, the National Supervisory Authority for Welfare and Health. (Later, Valvira's mandate was transferred to the Finnish Medicines Agency Fimea.) This meant that to be a biobank one had to meet certain official criteria and standards, both institutional and quality-wise, a change that reflected calls by the research community to coordinate and develop sample collection and storage in line with more transparent and professional standards. This can be considered the modernization of Finnish biobanking (Tarkkala 2019).

Before the legal institutionalization of biobanks, sample collecting and storage were mostly project-bound and often under the control of a particular hospital, researcher, or research institution. The new biobanks were to change this, yet the already existing and operating collections and ways of collecting samples and data were foundational for the first official biobanks and their collections. The institutional connections and established practices of the existing collections were also fundamental for identification of the route Finland should take, and of where Finnish biobanking could be successful. The already existing collections were seen as an internationally exceptional basis for modern biobanking (Laitala 2011). According to one study, Finland had over 190,000 samples within ten of its most significant epidemiological cohort studies, and pathology collections in its hospitals were estimated to consist of well over 2 million samples (Tupasela et al. 2015; Ministry of Social Affairs and Health 2007). The latter were used routinely in medical practice for teaching and research, as well as

for comparative purposes if patients became rediagnosed with a new condition. Now, they were expected to gain a new, additional purpose within a biobank.

Before the Biobank Act in 2013, understandings of the definition of a biobank were not fixed, and they included the idea of utilizing old sample collections but also new standardized practices, as well as open promises for future research. Work and negotiations were needed to form a joint understanding of the kind of profile and organization Finnish biobanks would have. For example, possible commercial collaboration and access and permission policies had to be established. Biobank status would also be tied to the ability to provide what was described as 'high-quality materials' for research. A big part of institutionalizing biobanking in Finland was the work undertaken to convince public funders and decision makers of biobanking's relevance and usability for contemporary research, which took place on several fronts, performed by several stakeholders. For example, many researchers wrote popular articles about the uniqueness of Finnish sample collections and data, and how this could be harnessed to benefit both medical science and the national economy.

> Already, large Finnish data sets have brought international competitive research funding to our country, and they will certainly attract top researchers interested in analysing unique materials to Finland in the future, enabling the creation of a genuine international research centre in genetic epidemiology. The new information produced in the analysis of the data sets is most obviously also of great national economic importance. (Palotie and Peltonen-Palotie 2004, 1,712, own translation)

The awareness-raising paid off, and in 2006 the Ministry of Social Affairs and Health integrated a sentence about biobanking into its strategy aimed at 2015: 'Appropriate operating conditions are created for the use and collection of so-called biobanks' (Ministry of Social Affairs and Health 2006b, 11, own translation). To create these conditions, a need was perceived to define what a biobank was and to chart all the legislation that currently

regulated biomedical research in Finland and elsewhere. To this end, the Ministry of Social Affairs and Health set up a working group in 2006. The group had two tasks: first, to evaluate and promote the possibilities of utilizing sample collections, and second, to map the current state of regulation and anticipated renewals of human tissue sample collections and their use in biomedicine. The report by the working group (2007) proposed that the establishment of biobanks should be enabled in Finland, and it took a stance in defining biobanks as sample collections for future and undefined research uses. The objective of the working group was generally regarded by stakeholders to be of high importance. It was acknowledged that an Act—or at least clarifications of the existing Act on the Use of Human Organs and Tissues for Medical Purposes (2001/101)—was needed, because the current regulation left many questions open. However, some considered new legislation unnecessary altogether, as it was believed that the system was already working rather well (see Vierula 2010).

The report also took a stance on a highly debated issue: should Finland aim for one national biobank, or should there be several? The working group supported the idea that there should be many of them instead of one big, national biobank of Finland. The reasons for this proposition were manifold: first, as there were already samples collected from over two million donors, it would not make sense to start a sample collection from scratch (Ministry of Social Affairs and Health 2007, 20); second, the control over existing sample collections was scattered among many different institutions, which meant that pooling and reorganizing them into one big collection would be a very complex manoeuvre organizationally; and, third, it was unclear whether researchers and hospitals would be willing to transfer samples and data to a central biobank, since the collections also represented an important research resource for them and centralization was not imbued with any clear incentives to participate.

Privacy matters were also used as a justification for a less centralized approach. The report by the working group stated: 'To centralize information about donors and the samples into one

place increases the risks related to data protection' (Ministry of Social Affairs and Health 2007, 20). This concern over privacy continues to be a faithful companion of genetics and genomics. Finally, instead of promoting the idea of one national biobank, the midterm report (Ministry of Social Affairs and Health 2006b) of the working group particularly emphasized the need for a national registry of sample collections. In addition to maintaining the registry, the tasks of this planned national unit would also include providing permits for establishing a biobank. Thus, this official instrument would monitor biobanks but not provide any centralized database for samples, or access to biobanks.

In connection with the governmental working group report (Ministry of Social Affairs and Health 2007), a draft for a new Biobank Act was published, and stakeholders were invited to give opinions and comments. During the public hearing, questions were raised for discussion that included issues of informed consent, commercial interests, ownership of the biobank collections, division of labour in controlling biobanks, and the actual possibilities of receiving samples from biobanks. Some, mainly researchers, questioned whether it is a relevant regulation at all—especially in relation to population and cohort-based biobanks (e.g. Vierula 2010). A related critique voiced that as there was good regulation already in place, the new legislation was only introduced because other countries were developing or already had such legislation (Tupasela et al. 2015). Many commentators were also concerned about how public institutions could attain funding for biobanks. In fact, there were numerous disagreements about the content and scope of the law, including in relation to the commercial utilization possibilities of biobanks.

Based on the discussions and criticism, a second and revised draft for the Act was published in 2010. However, even this second Biobank Act draft never managed to reach parliamentary discussion, as there were other pressing legislative issues before the new elections in 2011. After the new parliament started to work, the third version of the Biobank Act was drafted, opened for comments, and finally processed and passed by parliament in 2012. It

came into force in September 2013, after a political and administrative preparation process that took several years.

The Biobank Act and the first institutionalized biobanks

The content of the Biobank Act is crucial to understanding biobanking in Finland. In contrast to many other countries, Finland chose to base the institutionalization of biobanks on separate and specific legislation that defines what a biobank is and does. The Biobank Act (2012) defines a biobank as

> 'a unit maintained by an operator engaging in biobanking activities for the purposes of collecting and storing samples and information associated with the samples for future biobank research'.

Biobank research, then, is something that

> 'utilis[es] the samples contained in a biobank or information associated with them for the purposes of promoting health, understanding the mechanisms of disease or developing the products and treatment practices used in health care and medical care' (Biobank Act, 2012).

This letter of the law guides and provides a frame for biobank activities in Finland. Separate legislation of this nature can also be seen as a solution to a challenge raised and identified internationally: that biobanks need clear governance and legislative frameworks to succeed (e.g. European Commission 2012).

The Finnish legislation requires that all biobanks are registered officially as well as approved by a national authority (Fimea, from 2019 onwards; before that, Valvira). This makes the coverage of the term biobank, in practice, quite limited and specific. It is simple to count how many biobanks there are, for example, since each of them is in the register. In a way, this solution ended the dispute over whether old collections are biobanks or if only newly founded collections can be regarded as such. As a result, 12 biobanks, as defined by the Act, were founded and registered

during the decade following the implementation of this law. Two have since merged, so at the time of writing this book there are 11 biobanks registered in the official Biobank Registry maintained by Fimea. In addition to the registered biobanks, there are still numerous sample and data collections, but they are not considered by definition biobanks and are not covered by this legislation. This means that the Biobank Act has not hindered the collection and use of samples outside biobanking.

In the early 2010s, two disease-specific cancer research projects had commenced to pilot biobanking, setting up prospective sample collecting and trying to foresee the criteria in the upcoming legislation. Eventually, they would officially become biobanks when the legislation was enacted (Tarkkala 2019). After the law came into force, hospital districts started to establish their own biobanks and slowly commenced the collection of new, prospective samples to complement their older pathology collections of diagnostic samples. At the beginning of 2014, the THL Biobank, a national, population-based biobank at the National Institute of Health and Welfare, and Auria Biobank, a regional clinical biobank associated with the Turku University Hospital and hospital district, were established and registered, and started to operate as the two first official biobanks in Finland. The THL Biobank was founded on the cornerstone of already existing sets of samples collected for research purposes, and included biobanking in their new research projects, instigated after the Act became operational (see Figure 4). The two disease-specific biobank pilots—the Finnish Haematology Register and Biobank and the Helsinki Urological Biobank—both also gained their status as biobanks in 2014.

While the new Biobank Act defined biobanks in a way that seemed to concentrate on establishing new collections and new infrastructure, the existing sample collections still played a significant role in these new biobanks. In fact, most of the biobanks were founded with existing tissue collections. A specific part of the Act enabled the transfer of old collections to a new biobank, and for this reason many of the very same collections and samples that had been listed as already existing 'biobank' collections

during the period while the law was being drafted now fit the legal definition. Before the transfer, however, registered individuals whose samples were involved were supposed to be notified of this. In practice, biobanks considered that asking every individual if they would allow their old diagnostic or research samples to be transferred into biobanks would be unreasonably burdensome or even impossible. Therefore, many biobanks resorted to the opportunity presented by the Act to transfer samples based on public announcement (Salokannel et al. 2019; see Tupasela 2021). We discuss the issue of 'legacy samples' in more detail in Chapter 5.

For example, the old samples from the clinical collections at Turku University Hospital were the foundation of Auria Biobank's sample collection. Although the collection of new, prospective

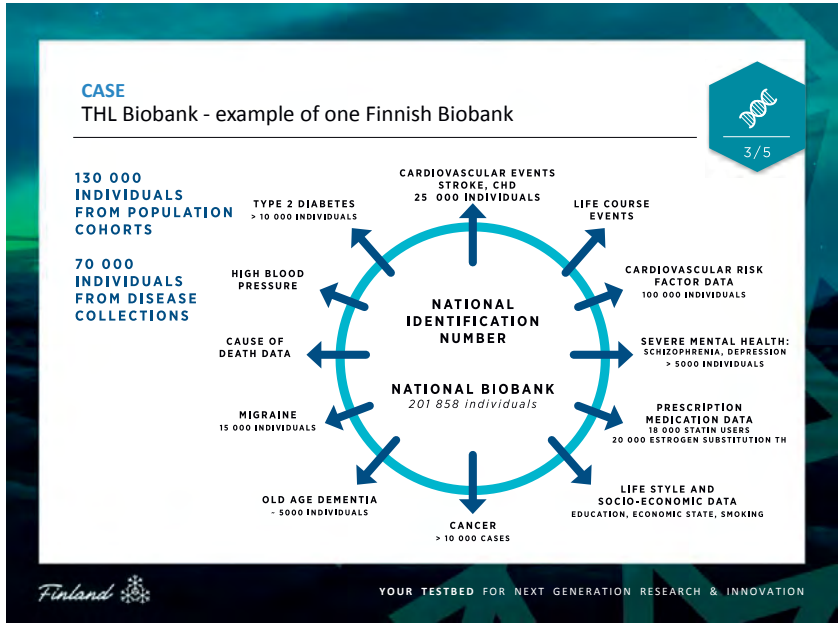

Figure 4: Case—THL Biobank. This slide from a promotional presentation shows how the already existing collections or so-called legacy samples formed the basis for Finnish biobanking. Interestingly, in this presentation the THL biobank was referred to as a 'national biobank' (Sitra 2015a). All rights reserved.

samples has been ongoing since 2015, the majority of Auria's samples are currently still old diagnostic material from the hospital collections, of which cancer-related samples are the largest proportion. The THL Biobank has been in the biobank register since 2014, having as its base the existing collections and cohorts of THL (see Figure 4). Indeed, the number of legacy samples in Finland, now transferred to biobanks, is over 10 million (Salokannel et al. 2019). To put this into perspective, the current population of Finland is about 5 million inhabitants and there are 500,000 new, non-legacy, samples. For many biobanks, the old legacy samples will form the core of their collection for years to come, and it will take a long time until the number of prospective samples outstrip the legacy samples. Of course, new and prospective samples have also been collected by the new biobanks; the disease-specific biobanks, such as the Haematological Biobank, and the biobank of the Finnish Red Cross Blood Service even rely solely on a new, prospective sample collection.

Implementing 'the best biobank Act in the world' and initial problems

After the lengthy process of preparing the Biobank Act, the first biobanks were established in 2014. The mood was optimistic, and positive statements, presentations, and public speeches about biobanking and the benefits of the new Act were rife. Whether the speaker was a ministry official, a representative of the Finnish pharma industry, or a biobank manager, the common statement in their public speeches was that Finland is and must continue to be united. As all actors, from ministry officials to healthcare providers and genetic researchers, are involved in shaping the biomedical future of Finland, everyone needs to tell the same story, speak with 'one voice', and share the same ambition (see also Tarkkala and Snell 2022).

Olli Carpén, appointed the first biobank professor in Finland in 2013, gave speeches stating that Finland now had the best biobank Act in the world (e.g. Carpén 2015). The reasoning behind this

statement was that as a small country, Finland cannot compete with the large numbers of samples. The Biobank Act, however, could lift Finland to the top of the biobanking world by creating an environment that was easy and smooth to navigate and operate. Having a legal framework and a specific model of operation in general was also seen as a factor that increases public trust in biobanking. In general, the Act was described as having good balance: securing the rights of the donors and enabling research and development at home and, importantly, across borders.

The first biobanks in full operation, especially the Auria Biobank, had the opportunity to interpret the legislation and establish practices that would also serve as examples for other actors in the field. They got to develop practices related to data management and collaboration with commercial actors, and also put together the first informed consent sheets and information leaflets about biobanking for donors and the public, allowing them a kind of head start in these matters. In 2015, after operating 1.5 years, Olli Carpén wrote in a blog text that the Biobank Act enabled the use of precious research materials. In addition, the start had been good:

> For Auria Biobank, the merging of samples and data has started successfully. During the past year-and-a-half, thirty new research projects have been started, the first of which have already been completed. (Carpén 2015)

Soon after the launch of the biobanks, however, it emerged that the new biobanks were finding it anything but simple to comply with the legislation (see also Soini 2012), which had to be interpreted on the go as the biobanks confronted new and unanticipated situations for the first time. Not all stakeholders, including biobanks themselves, were happy with the content and formulations of the Act, and there were complaints about how difficult the implementation period seemed to be. In addition, a number of experts raised concerns about the overall functioning of the Act. It was criticized, for example, for being too complex, and therefore it was difficult to understand what the legal requirements

actually meant in practice. Moreover, specific parts of the law were being questioned, such as the requirement to return research results and a disregard for the idea of combining research and care (see Tupasela and Liede 2016). Others were concerned about whether the Biobank Act was actually hindering some types of research, which would inevitably lead to their eventual decline. Moreover, the need to obtain consent in order to collect and store samples was identified as slowing down activities, and the idea of changing to the opt-out model was raised and discussed (Snell and Tarkkala 2019). In their report, an expert group stated on this topic that the sample collecting was a bottleneck for Finnish biobanks, 'which is complicated by the current consent process that has proved to be somewhat inefficient' (Ministry of Social Affairs and Health 2016, 9).

One of the most fundamental problems identified in the Act was that it was too sample centred (HE 2018). The consent model and the legal processing of biobank samples and data were connected to the actual existence of a sample, while collaborators and researchers might have been solely interested in the already existing data. Because of the consent and sampling process, it was possible for a situation to emerge where there was consent but not yet a sample. In this situation, a person's data could not be used as long as the sample was not taken. The idea was that people would give a sample to the biobank next time they came to the laboratory. However, for many generally healthy people this means that the sample could be taken months or even years after they have given their consent. Many biobank experts considered that this sample centredness was hindering the possibilities of biobanking, given that many biobanks were in fact aiming to be health data repositories, not only sample depositories (Tarkkala 2019).

The situation also highlighted how biobanking's perceived potential benefits had changed from the expectations when founding and planning them. Biobanks were no longer seen as attractive partners because of their samples, but because they also had clinical data in a standardized and accessible form, combined with precise sample or other biological data (Tarkkala 2019;

Lehtimäki et al. 2019). Biobank experts wrote in an article that the Finnish approach, which aims to go beyond sample collecting to focus on combining both clinical and register data with samples in biobanking, is special:

> This approach departs from the usual biobanking, in which the focus has been in sample collection and management without investing in the clinical data related to the samples. Yet the combination of sample-based biological data – 'the omics data' – and phenotype data from patient information systems and registers is what modern, clinical trials need and require. (Carpén and Helander 2017, 593, own translation).

During the first years of biobanking it became clear overall that the new biobank legislation was already outdated when it came into force (see also Soini 2012), so a process to update it started almost immediately. Parliament had required the government to monitor and evaluate the functioning of the Act, and, to meet this obligation, the government appointed a new working group—the steering group for the Biobank Act. In its midterm report (2015), the group suggested several amendments to the Act, some of which were rather technical while others implied more profound changes. As many problems with the biobank legislation were identified right away—and it was uncertain when the Act would be amended—the institutions and people involved in biobanking expected more guidance and interpretations of the Act as it currently stood from the Ministry of Social Affairs and Health and Valvira (Tarkkala 2019). For some biobanks, the early identification of the need for legislative reform and the wait for clarifications and new instructions meant postponing operations such as beginning the collection of new samples; biobanks did not want to take steps that might turn out to be juridically suspect or even illegal. Therefore, many started slowly and waited for upcoming guidance and updates. The legislation had created just the kind of uncertainty it was meant to expel, with biobanks seeing their operating environment as being in a state of flux rather than stable and unambiguous (e.g. Southerington et al. 2019). As

one biobank advocate described the first years of biobanking in a research report:

> We have got stuck in it, that first it took so long to get the Act, and they assigned the steering group, and the working group of civil servants that should give the recommendations, the statutes and possible recommendations. So we cannot do very much. We cannot say that let's do this kind of consent forms because we have to wait that the working group would say what they will require. (Tupasela et al. 2015)

Eventually the reform process became as prolonged as the original drafting of the biobank legislation. This could be taken as a sign of fundamental challenges in the field, including the introduction of the General Data Protection Regulation (GDPR) in the European Union. It is also indicative of rapid changes in what kind of regulation is required as scientific practices and technology evolve and move on. Two rounds of proposals for reformed biobank legislation were presented, one in 2018 and another in 2020, and both proposals were commented on by stakeholders and experts. Among the most discussed aspects of the new proposals were implementing the GDPR, what counts as data associated with a sample, and the relationships of biobanks to other organizations and authorities (discussed below). In 2022, one more government proposal (HE 247/2022) for reform was submitted to parliament, and the revised Act was eventually accepted in the parliament in February 2023. However, it seems that the Biobank Act was ultimately mainly updated to meet the stipulations of the GDPR, and for the large part otherwise left as was. The proposed changes have been described as 'technical' (Finnish Government 2022), but they include profound changes to the practice of informed consent, which we discuss in Chapter 5.

Mergers, networks, and service points

During the years when biobanking operations were being planned, formed, and kicking off, Finnish actors in the field were

participating simultaneously in international collaboration, programmes, and endeavours. There were numerous European-level efforts to coordinate practices regarding both samples and data, all in the name of fostering biomedical research and innovation and enabling collaboration. There were also many programmes in their preparatory phases of initiation, including the establishment of ELIXIR Europe in 2006, which aimed to manage, coordinate, and guide the resources of life science laboratories, organizations, and units to create a European research infrastructure. Similarly, a European-wide project called Bioshare ran from 2010 to 2015 with a goal of standardizing and harmonizing data across biobanking data sets. This covered, for example, data on lifestyle and social circumstances as well as the phenotypes associated with common diseases.

Under the European Union, the importance of networking and standardization also gained attention specifically in the context of biobanks. The BBMRI-ERIC (a European research infrastructure for biobanking) started to operate with the official status of a European Research Infrastructure Consortium (ERIC) in 2013, with the goal of coordinating biobanking in the EU. The aim was to facilitate access to biological resources and introduce common standards and harmonization—thereby fostering collaboration—across Europe. The preparations for and implementation of BBMRI-ERIC took place between 2008 and 2012, which coincided with the drafting of the Finnish Biobank Act and the piloting of the first Finnish biobanks. The process of creating this European network resulted in an infrastructure for biobanking in Europe with its own headquarters and national nodes that continue to foster collaboration, while also offering support in terms of ethical, legal, and societal issues. They have also developed models for public–private partnerships in R&D in the field. Finland became a member of BBMRI-ERIC in 2013 and subsequently established a national node. One of the explicit goals of BBMRI.fi was to contribute to commercialization:

> BBMRI-collaboration is particularly expected to foster research uses of samples that hospital districts have collected for decades. The results may enable the use of funds invested in Finnish healthcare to commercialize new knowledge (Finnish Government 2013, own translation).

Most of the Finnish biobanks were to become part of the national node BBMRI.fi. This was one of the first joint contexts for Finnish biobanks to build national cooperation and collaborative relationships with other European biobanks. Yet, for European-level collaboration to become possible, national collaboration had to be firm as well:

> Extensive national cooperation guarantees that the entire research community utilizing biobank materials will also have access to European research infrastructure and cooperation opportunities in research. The goal of the national biobank network is the introduction of common and compatible operating models, utilizing existing resources and investments as much as possible. (Ministry of Social Affairs and Health 2015b, 19, own translation)

An earlier report (Ministry of Social Affairs and Health 2007, 20), called 'Biobanks our common benefit', which was published together with the first proposal for a Biobank Act, had identified even then that there would be the need for top-down guidance as well as collaboration between biobanks in terms of establishing unified practices and IT structures. BBMRI.fi, then, was one form of such collaboration, and it was heavily encouraged by the Ministry of Social Affairs and Health. However, from the ministry perspective, soon after biobanking operations started it had become clear that the level of national coordination was insufficient and the field of biobanking in Finland was too scattered. The government and the Ministry of Social Affairs and Health kept an eye on biobanks and were interested in facilitating collaboration in biobanking to secure the identified potential benefits.

In an interview with Health Europe, the director-general of strategic affairs and a chief medical officer at the ministry,

Liisa-Maria Voipio-Pulkki, retrospectively describes the ministry's strong role in fostering the unification and standardization of the Finnish biobank field in those early years. She states that they had put pressure on biobanks to encourage a situation wherein biobanks had to collaborate in order to secure their funding. She describes the unification of operations as follows:

> It took a couple more years, and some government money, for their operations to become more uniform. Originally, we hoped that we could have just one national biobank, but that was not possible. Most biobanks are owned by independent hospital districts, but the government sent them a very strong message that they had better co-operate with each other and, if they were able to develop some common processes and standards, the government would support them with some seed money and also by writing new legislation if necessary. (Health Europa 2018)

So, the path chosen—that of having multiple regional biobanks—was also problematized by the ministry itself. The field developed slowly, as some biobanks were only in the starting stages; biobanks interpreted the legislation differently; and at the same time it was thought that modern medical science needed a larger pool of samples than individual Finnish biobanks could offer their collaborators. In addition, having multiple biobanks could not provide a smooth service, as those wanting access to samples and data would need to contact each biobank separately. In a report from 2015—drafted to follow up and evaluate the functioning of biobank legislation, and to point out the required reforms—it was stated that there was a need for a centralized service through which researchers and collaborators could check the availability of samples in all Finnish biobanks (Ministry of Social Affairs and Health 2015b, 20). Again, the ministry set up a working group to investigate the integration of Finnish biobanks and whether it should happen through mergers or closer cooperation, or by creating a new organization with legal status (Ministry of Social Affairs and Health 2016).

But it was not only the ministry that was calling for a more unified biobanking sector. Biobanks had also proactively started to reorganize themselves outside the BBMRI.fi, which had until then served as the national node of biobanks with all biobanks as members. For example, the disease-based Helsinki Urological Biobank, which had started as a biobank pilot project, merged with the bigger Helsinki Biobank. Meanwhile, the possibility of an even larger merger was being investigated, as first the regional biobanks of Turku and Tampere started to plan one, and later Oulu joined the plan (PSHP 2017). This planned merger was advertised as the first step to creating a national biobank, and a route to test how a merger would be possible legally and practically. The idea was to form a joint, centralized service operator for Auria, Tampere, and Borealis biobanks, but they did not go ahead with it. Instead, a national model for a Finnish biobank cooperative grew from their efforts, with an objective to 'gain an internationally credible position' (PSHP 2017).

Thus, the integration of biobanking activities was put forward from both sides—biobanks and the ministry—building on the working groups' memorandum. And, after the merger between the three biobanks had already been prepared (as noted above), the nationwide option emerged and the Finnish Biobanking cooperative FINBB was founded in 2017. The purpose of FINBB was to serve as a one-stop shop for Finnish biobanks' customers. The cooperative was a response to the need to integrate biobank activities, as well as part of the rising ethos in the field that Finland had to offer a centralized and searchable sample catalogue and easy access to data. It was not, however, an effort to merge local biobanks into one national biobank. As the integration report (Ministry of Social Affairs and Health 2016) states, this would have been too big a change and one that would not please all the local host institutions (such as hospital districts), hospital staff, and patients. Thus, a cooperative was a compromise that seemed to be accepted by the stakeholders, as it pooled resources but also left room for local innovations and decision-making. The cooperative states that its mission is 'to enhance the competitiveness

of Finnish health and biomedical research by providing researchers a centralized access to collections and services of the Finnish biobanks and their background organizations' (Fingenious n.d.). The centralization of a single access point for biobanking samples was also something that was being developed internationally at other biobanks, such as the Danish National Biobank, to facilitate easy access to samples (Tupasela 2021). FINBB cooperative is owned by regional healthcare providers (wellbeing services counties), universities, and THL, and it hosts eight of the eleven registered biobanks. From the beginning of 2020, FINBB took the responsibility for being the national coordinator of BBMRI-ERIC, and all the eleven biobanks are part of the network side of FINBB.

The different ideas and initiatives to integrate biobanking represent the dynamics in the field. On the one hand, the Ministry of Social Affairs and Health has taken an active role in guiding the sector, and the biobanks have also expected it to provide clear guidelines and signs of support. On the other hand, biobanks have also wanted to develop their activities independently, and the ministry has stated on many occasions that it is the responsibility of the biobanks to interpret the law and advance their own operations. It has not always been clear when the ministry and biobanks expect each other to take a stance on issues; thus, there has not been a unanimously shared consensus on how to proceed and divide responsibilities in recent years.

Biobanks in the service of personalized medicine

In parallel with the institutional integration of biobanks, the purpose of biobanking was also evolving. Biobanks had been established in Finland to serve the future needs of medical research. During the decade that it took from the first memos to implementation of the legislation, the purpose became more focused on genomics, with increasing emphasis on the idea of personalized medicine: medical treatment that would be preventive, personalized, predictive, and participatory (e.g. Hood and Flores 2012),

accurately serving individual patients with the right treatment at the right time or even preventing diseases altogether. For personalized medicine to succeed and become a reality, international collaboration and large data sets, in addition to large sample collections, were needed. Biobanks were portrayed as offering a route to the execution of this new paradigm, at the heart of which lies digital, standardized, and computable information about individual people in relation to large populations. The idea is that both efficacy and precision will follow, and outdated, unstructured data such as patient narratives and non-standardized medical records will be history.

Throughout the first decades of the millennium, in addition to biobanks there have been several other projects and programmes in Finland to establish, for example, networks, infrastructures, and databases to support the development of biomedicine and genomics. Thus, biobanks were not the only route by which personalized medicine was being advocated. The Ministry of Social Affairs and Health moved from supporting biobanks to constructing a broader vision of personalized medicine, under whose umbrella several other initiatives, infrastructures, and reforms were prepared and took place, such as the National Cancer Centre and a planned new version of the Genome Centre (not the Genome Information Centre described earlier in this chapter and in Chapter 2). Eventually, as we come to conclude in this section, rather than becoming exclusively the flagship of biomedicine and genomics, or the only element needed to foster personalized medicine in Finland, biobanks became just one component of a larger infrastructure and of larger political ideals of innovations and economic possibilities tied to health data and the healthcare sector (see also Chapters 4 and 6). Thus, although the visions of the early 2000s positioned biobanks as answering the needs of future medicine and innovation, 20 years later the biobanks as they became organized in Finland were not enough in themselves to fulfil such visions.

One of the game changers was the launching of the idea of building a national Genome Centre. The Ministry of Social Affairs and Health together with Sitra—a large think tank that operates

under the Finnish parliament—prepared a proposal for a national genome strategy that was published in the spring of 2015. Its principal aim was to set in place 'key measures for ensuring that, by 2020, genomic data will be effectively used in health care and in the promotion of health and well-being' (Ministry of Social Affairs and Health 2015a, 3). The strategy proposal was the result of a collaborative process involving an appointed working group and workshops open to medical, scientific, administrative, and commercial stakeholders. The goal was to prepare the strategy in time for it to make it into the programme of the upcoming new government. Eventually, however, the strategy was left out, which was a huge disappointment to the stakeholders involved in the drafting process. Despite this setback, the genome strategy continued to be actively promoted and lobbied for, and a year later government funding was directed at establishing a national Genome Centre. The preparations to plan and establish the centre and especially to draft new legislation for it started at the beginning of 2016. The purpose of the centre and its connection to biobanking was outlined as follows in a government statement:

> A Genome Centre will be established in Finland, aimed at developing Finland into a pioneer and internationally desired partner in healthcare, high-level research and global business utilising genome data. Public biobank activity will be enhanced by standardising operating methods and ensuring effective cooperation with the Genome Centre. (Finnish Government 2016)

A large number of stakeholders were included in the Genome Centre working group set up by the ministry—including representatives from biobanks, hospitals, and universities (and a social scientist)—to discuss what the centre should do to reach the goals outlined in the genome strategy. The central idea was that the Genome Centre would store the genome information of all Finns in one place, starting in the near future. But the practicalities of this proved to be a heated topic: What was considered genomic information? Can other public infrastructures also store genome data? Is it mandatory for all public and private actors to

store information in the Genome Centre and on what grounds? One of the disputed topics was the relationship between biobanks and the centre. During the first years of biobanking, it became clear that one of the main incentives for storing samples was that users of samples would perform a genome analysis on them. The operating idea of the biobanks was built on the logic that information derived from a sample in a research project—such as genome data—would be returned to the biobank, which would then store data that in itself could be utilized in further research projects (Tarkkala 2019).

The push to establish the Genome Centre complicated this picture and the division of labour between biobanks and the potential new infrastructure, which would not only be a source of data but a state authority regulated by yet another law. With these discussions, the role of biobanks became contested. If the Genome Centre became the access point for Finnish genome data, would the data-focused Finnish biobanks become mere sample depositories? As the preliminary documentation related to the Genome Centre was made available for comment by stakeholders, it was not only the representatives of biobanks that called for a clearer division of labour between biobanks, the biobank cooperative FINBB, and the proposed centre. Researchers, hospital districts, and patient organizations had also noted that these different organizations had parallel and overlapping objectives and missions, without a clear division of labour.

While the Genome Centre was being prepared and its connections to other national efforts were debated in Finland, there were new European developments as well. A large initiative called '1+ Million Genomes' was launched. Its aim is to enable secure, cross-border access to genomic data in Europe, safeguard privacy protection, and benefit the people. Finland signed the declaration in April 2018, but this came as a surprise to the Genome Centre working group, as the European initiative had been managed by a different group of people at the Ministry of Social Affairs and Health. Slowly 1+ Million Genomes merged into the landscape also, and became part of national developments from January

2021, when a seminar was organized to kick off Finnish activities related to the initiative, taking into consideration the ongoing national developments too (Ministry of Social Affairs and Health 2021).

Just like biobanking and the Biobank Act, the establishment of a Genome Centre and drafting of related legislation proved to be a similarly long and complicated process. There were four different rounds of comments between 2017 and 2021. Eventually the Genome Act was split into two parts: the first dealing with the establishment of the Genome Centre and the second dealing with storing and analysis of genome information. The first part reached parliamentary discussion in 2022 and not even a draft of the second part was available. This was one of the most important criticisms of the proposal, as many stakeholders found it difficult to comment on 'half of a law'. Others questioned the need to establish yet another infrastructure while existing institutions such as biobanks were under-resourced. The law was not passed, and it is still unclear whether the centre will become reality at some point. The idea of a national genome data repository is still present in some documents (Pentikäinen et al. 2023), and an updated version of the Genome Strategy (Ministry of Social Affairs and Health 2023) was published in 2023.

The proposed Genome Centre was not the only thing adding complexity to the space in which biobanks were developing and planning their operations. An unprecedented biomedical genome-mapping project was being prepared and piloted by the Institute for Molecular Medicine Finland (FIMM) at the University of Helsinki, with Professor Aarno Palotie as its director. Out of the project grew a consortium called FinnGen, Finland's national genomics initiative, which resembled the ideas related to biobanking and genealogical data in Iceland and the UK Biobank in the United Kingdom (Tupasela 2021, 113). FinnGen states that its main aim is 'to improve human health through genetic research, and ultimately identify new therapeutic targets and diagnostics for treating numerous diseases' (FinnGen n.d.). The study describes itself as unprecedented, as it is a collaborative project

that brings together 'Finnish universities, hospitals and hospital districts, THL, biobanks, international pharmaceutical companies and, hopefully hundreds of thousands of Finns' (FinnGen n.d.), and combines genome information with digital healthcare data from national health registries. The original aim was to collect samples and data from 500,000 Finns, which task was accomplished in autumn 2023.

FinnGen was officially launched in 2017, the same year as the biobank cooperative FINBB. It is important to note that FinnGen was not about building an infrastructure. Rather, it is an academy–industry collaboration and a precompetitive research project. Why it made such a difference for biobanks, however, was that its funding from Business Finland and pharmaceutical companies exceeded the public resources directed to biobanks, the Genome Centre, and FINBB in total, and part of this funding was allocated to gathering samples through biobanks. The funding for collecting and genotyping 500,000 samples within FinnGen provided biobanks with much-needed extra resources, as they were the institution actually doing the collecting and pooling of samples.

In a couple of years, FinnGen became not only the largest user of Finnish biobank data, but also the most important customer of Finnish biobanks and the main route to the Finnish tissue sample and health data repositories for foreign researchers and companies. Importantly, it also became the enabler of Finnish biobanking, since some of the smaller hospital district biobanks in particular had been struggling to find adequate funds to start sample collection. Now there was FinnGen to provide resources for this. In its own way, FinnGen took over biobanking in Finland, and, to an extent, the objective of FinnGen—'to produce comprehensive genome variant data of 500,000 biobank participants' (FinnGen n.d.)—became simultaneously the priority of Finnish biobanks. Thus, the objectives of biobanking were constructed not only in legislation and in relation to other infrastructures, but also in accordance with the available funding and resources.

Not surprisingly, FinnGen and the proposed Genome Centre were not perfectly aligned. At first, in 2015, when both FinnGen

and the centre were in the early planning stages, the representatives of FinnGen were arguing for the need to establish a reference database of Finnish genomes, and it would have to be done fast. The Genome Centre was seen as a route to this goal. Five years later, as the centre had still not been established, the FinnGen people were arguing that there is no need for it, as their project already serves as an example of good practices in storing and utilizing large amounts of genome data. Thus, this is yet another example of how during the first 20 years of the millennium, genomics has become increasingly an issue about data access, storage, and custody (see also Reardon 2017). FinnGen's major challenge is that it is a research project and does not have a legal mandate to continue operations after the project ends. The establishment of a Genome Centre, however, would provide infrastructure and have a legal mandate, making its long-term operations sustainable. As of the writing of this book, the fate of FinnGen in relation to the samples and data it has analysed remains unclear.

Biobanks and secondary use of health data

The 2010s were a decade during which it became evident that data itself is a currency and asset. While several stakeholders were advocating biobanks as part of personalized medicine, others started to envision expectations in terms of a data economy. Genomics also kept moving towards more and more data-intensive practices—becoming data-driven (Hoeyer 2023). But the relationships between samples and data are not clear-cut, and the emphasis on data has not disconnected it from samples (Tarkkala 2019). Besides, there is a popular understanding that a sample in itself is not valuable; it only becomes so with the associated data. Thus, information was needed from the sample donor, although it could also be generated from the samples themselves. Therefore, samples and the analyses based on them, as well as information on the donor, are all essential for R&D practices (e.g. Strasser 2019; Tarkkala 2019). In biobanking, the aim is to turn samples into data that can thereafter circulate with different uses, which is

sometimes presented in banking terms as samples gaining interest as new layers of data from previous analyses pile up in the bank despite the actual material sample diminishing (Tarkkala 2019). The sample/data nexus is a challenge for the regulation and organization of research resources, since samples and data are generally not seen as the same or interchangeable.

The differentiation between a sample and data has its background in the former always requiring some sort of intervention in the human body. The guidelines and regulations related to medicine and research apply special emphasis to bodily interventions; however, in practice a great deal of health data originates in the samples. To give an obvious example, the old diagnoses written in the health records might be based on blood or saliva. Even when this is old and a diagnostic sample has not been taken for research or biobank purposes, it is in the form of health data in the records that might be used for biobank research. But, as noted above, without a specific *biobank sample* having been collected, this data cannot be utilized from a biobank. The way this prevented the use of solely health data through biobanks was eye-opening for the whole research community and the Ministry of Social Affairs and Health. Not only did it reveal the need for legislative reform, it also demonstrated that there was a need for the data themselves, and they needed to be made available as quickly as possible to avoid the loss of competitive advantage. This, then, is one practical example of what, in Finland, led to enthusiasm for new openings providing access and service connected to health data. But instead of developing biobanks to be data providers also, both government and hospital districts started to develop their own services and access points eagerly. At the moment, many regional well-being counties (former hospital districts) have their own so-called 'data lakes' that are advertised for collaborators interested in clinical data. For example, the data lake of the largest regional public healthcare provider—HUS in the capital area—has data from 3.5 million people. The data include patient records, lab results, and pathology data. There has also been talk about storing genome data in the HUS data lake.

On a national level, a large initiative called Isaacus was initiated in 2015 to create a new operating model to collect national and local health and well-being data and distribute it to researchers and service developers. The aim of Isaacus—also called the Digital Health Hub—was to function as a one-stop shop where researchers, research institutes, companies, and other users could easily access register data from different regions and organizations with only one permit application. The vision of Isaacus overlapped temporally and conceptually with the efforts to integrate biobanks, as one of the motives for integrating biobank activities was to create a one-stop shop for biobanking. In practice, both biobanks themselves and the Isaacus effort—which was led by Sitra—were planning similar but separate service points. In the biobank integration report, this was acknowledged both as a possibility and as an unsolved issue:

> The importance of having a single contact point for accessing data has been noted. However, the connection between 'Biobank Finland's contact point' and the 'one-stop-shop' for all health and medical data has not been defined, although they may well be considered as being complementary. (Ministry of Social Affairs and Health 2016, 22)

At the same time, the Ministry of Social Affairs and Health was preparing a legislative proposal on the secondary use of social and health data, which would set the legal framework for the new centralized permit service and Digital Health Hub. Some people in leading biobank positions criticized this, as they thought that both Isaacus and the legislative framework were prepared hastily and without involving biobanks. The worry was that Isaacus would take over many tasks envisioned by biobanks to be their core activities, leaving biobanks as merely freezers for samples.

In contrast to the Biobank Act and the Genome Centre, the legislation for secondary uses of social and health data was being prepared rather quickly during the latter years of the 2010s. The Act on the Secondary Use of Health and Social Data was passed in parliament in 2019, and the new national permit authority, whose

working name Isaacus was replaced by Findata, started to operate at the beginning of 2020. Findata—a permit authority and data operator—has two main tasks: granting data permits to health and social data in a centralized manner when data are needed from several different keepers of registers, and compiling, combining, and pre-processing datasets for customers. In addition, Findata provides a secure analysis platform or operating system that can be used in analysing the data. If a client does not use Findata's Kapseli system, it must have its own analysis environment that meets certain technical and privacy criteria.

The data to which Findata grants access include medical records that are stored in the national Kanta services, population data, data about pensions and occupational illnesses, social security benefits and prescriptions, causes of death, matters related to social welfare and healthcare, and so on (Findata n.d.). In terms of genome data, Findata cannot currently grant access to them. Sitra envisioned the future of the new operator in 2019 as follows:

> The vision is that the new operator will be able collate data comprehensively from various public-sector registers including genomic, biological and clinical data registers. New genome laws and reforms of the biobank act are being prepared and these amendments may affect how the data from those sources can be accessed. (Sitra 2019, 23)

While Findata cannot provide access to genetic data, there is, however, another route to such material: the other one-stop shop envisioned by biobanks. The FINBB cooperative launched a Fingenious service which provides an access point to genome data returned to the public Finnish biobanks. This consists mainly of genotypes produced in the FinnGen project described above. So Fingenious offers access to biobanks' sample-related data, including both phenotype and genotype data, but data from Finnish national registries or hospital data lakes can be linked to this data set only by a separate application via Findata. So instead of a one-stop shop, Findata and Fingenious form a two-stop shop.

The Secondary Act made it possible to use health and social data not only in research and statistics but also in development and innovation activities, teaching, knowledge management, and supervision and steering in the social welfare and healthcare sector and in official planning tasks. This expansion in the interpretation of possible uses of secondary data was something that has interested other countries as well, since it expands considerably the possible uses of health and social data. Smooth and fast access to data was the main driver, as one representative from the Ministry of Social Affairs and Health explained in a press release:

> The data required for research can be obtained faster when the permit procedure becomes smoother and the researchers have access to ready-combined data. (Ministry of Social Affairs and Health press release, 2019)

However, as soon as Findata started to operate in 2020, its customers identified many problems with the legislation and especially with the new permit authority. Instead of offering smooth access to data, Findata became a bottleneck for research. Research groups complained about lengthy permit application processing times, and individual researchers argued that the pricing of the services hinders them from doing research. Even Findata and the Ministry of Social Affairs and Health acknowledge that Findata was under-resourced. Another problem was the secure operating system Kapseli, which also added to the costs. Some bigger organizations were developing their own secure analysis platforms, but most users do not have the resources and have to use Kapseli to access data. On the other hand, before Findata, researchers had to gain separate permits from each organization from which they wanted to collect data, which was not a quick and easy process in itself. Despite the many challenges faced after the setting up of Findata, the ethos has been to expedite data access for secondary purposes.

The story is not over yet, however (Figure 5). The possible re-emergence of the Genome Act and Genome Centre in some form—as well as new or upcoming national legislation dealing with

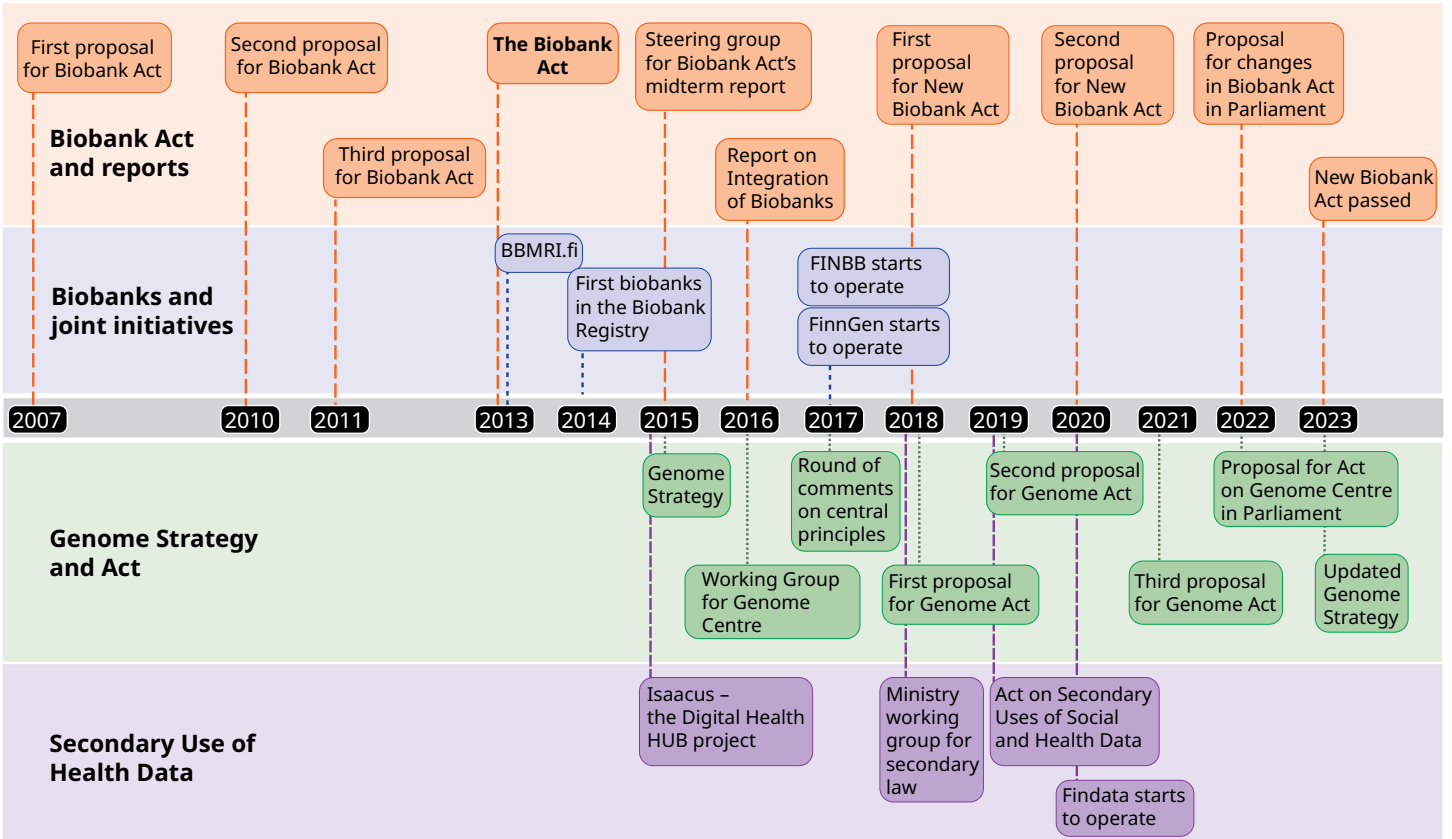

Figure 5: Timeline of important milestones of building up biobanking and other infrastructures.

customer and patient data—can change, for example, how genetic data are defined. The question is whether genetic data can be considered a form of patient register data or something different, what kind of protection they might require, where they will be stored, and who has the power to grant access to them. In addition, the planned European Health Data Space (EHDS)[6] initiative, which aims at sharing health, genetic, and biobank data across the European Union for both care and secondary uses, is expected to cause changes in legislation and practices (European Commission n.d.).

Conclusion

In this chapter we have discussed how the Finnish research environment developed from scattered sample collections to data repositories through several overlapping processes related to the initiation of biobank infrastructure and new authorities connected with genetic and health data and biological samples (see Figure 6). This has also meant continual drafting of new legislation and updating of existing regulation. Controversies among experts have arisen each time a new law or infrastructure has been proposed. The division of labour between different organizations—such as that between biobanks and the proposed Genome Centre in the mid-2010s—is often unclear.

In practice, then, the role of biobanks, as well as the organization and availability of genetic data, is being constantly rearranged and reinterpreted through new proposals for regulation. Rounds of comments and stakeholder resistance have sometimes influenced the ensuing proposals to such an extent that a complete U-turn has been taken. A big problem pointed out by many stakeholders is that the interrelated elements of legislation are presented one at a time, and therefore nobody can understand how the whole regulative architecture will look in practice.

The Finnish Biobank Act has been considered both as 'outdated as it was born' and as 'enabling regulation'. Although connecting the definition of biobanks to legislation may be an exceptional choice internationally, eventually it became identified as

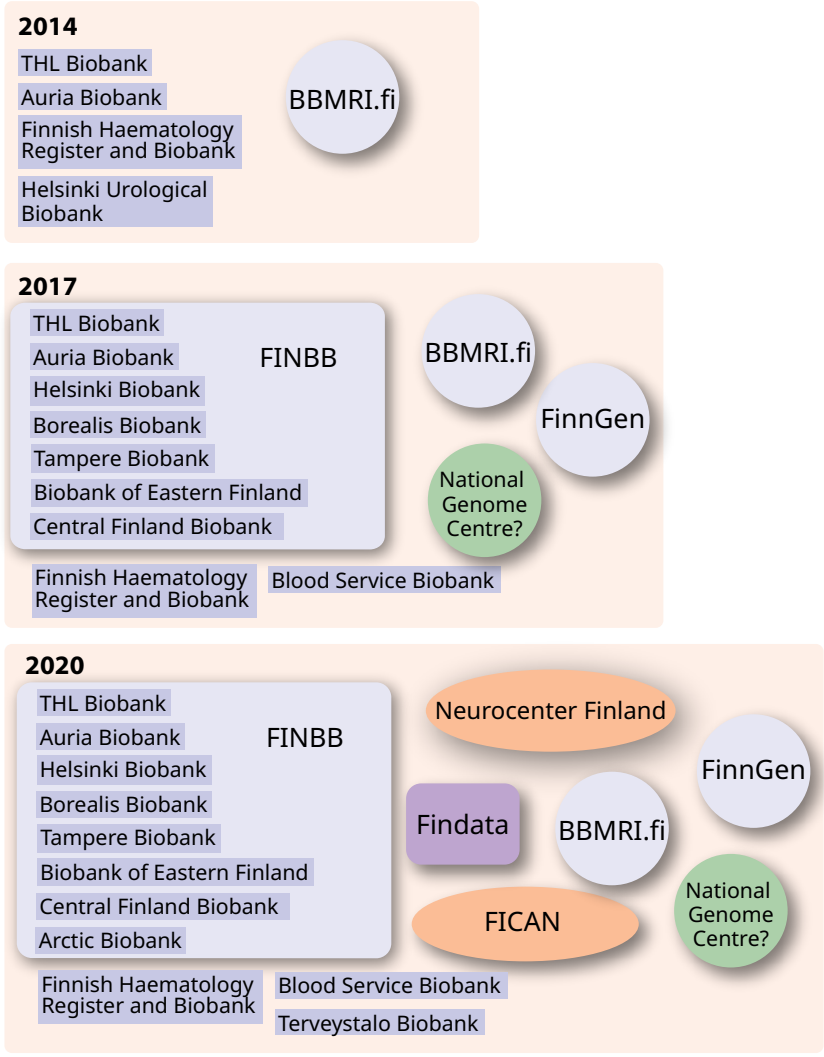

Figure 6: Biomedical infrastructures in Finland. The figure demonstrates how central biobanks were in Finland in the early 2010s. During the following years new, parallel, and competing infrastructures and projects emerged in the field as the political focus changed to personalised medicine and health data economy.

Note: BBMRI.fi is the Finnish node of the Biobanking and Biomolecular Resources Research Infrastructure; FINBB is the Finnish Biobank Cooperative; FinnGen is a public–private research consortium; Neurocenter Finland is the Finnish cooperation network for neurosciences and research; Findata is the national permit authority and data operator; FICAN is the Finnish national cancer center.

one of the special strengths of Finnish biobanking and the Finnish health sector (Finnish Government 2020). The law has been seen as a 'sustainable foundation that guarantees the international reliability of research' (Ministry of Economic Affairs and Employment 2014, 16). Moreover, the legislation and the interest in developing it were seen as a commitment from the government to support this area and its success. Still, the law required amendment immediately after it came into force. The latest renewal of the Biobank Act was discussed in 2022 and passed in the parliament in 2023. Interestingly, at this point, several biobank actors were defending the original Biobank Act, as they have adjusted their operations to it. Changes to it pose a threat to the stability and trust biobanks consider they have gained.

The example of biobanking shows that the field is constantly reorganizing and adjusting itself to the changing needs of research, genomics, legislation, and governance, as well as trying to identify and establish new services to attract the interest of collaborators and investors. Figures 5 and 6 summarise the often hasty and at times overlapping development of the field and its growing activity.

CHAPTER 4

A unique population: Registered, recorded, research friendly

Introduction

> Finland has exceptional conditions for a genetic research covering the whole population. Finland has an internationally unique Biobank Act that makes collections of hospitals and research institutes available for all researchers. Combined with the other strengths of Finland: comprehensive registers, electronic medical records and a research-friendly population, it enables extraordinary opportunities for new research and business. Moreover, thanks to the genetically unique Finns, genomics data is faster to analyse and the probability of findings is higher than in genetically heterogenic populations. (FinnGen n.d.)

The above excerpt has been taken from the website of FinnGen, Finland's largest and most ambitious genomics programme to date (discussed in Chapters 3 and 6). The text presents the benefits of genetic research in Finland. Not only does the consortium boast that 500,000 samples are available through the Finnish biobanks for this endeavour, but it also lists several of Finland's supporting features. These include the data already collected in public repositories, enabling regulation, and the research friendly population. On top of this, the efficacy and benefits for biomedical research are seen to result from the genetically homogeneous composition of the population.

In the previous chapters, we have described how Finland became the environment for biomedical research that it is today. This has taken place through building on the work that simultaneously established and utilized the unique genetics of the population from the 1960s and 1970s onwards, and through the availability of samples and data that were eventually opened for multiple use and made available in biobanks during the 2010s. These changes were followed by expanded secondary use of social and healthcare data in the 2020s. The first of the aforementioned developments resulted in wider knowledge about the genetic history of the population.

Today, the success of Finnish genetics is further reflected in the understanding that people in Finland are well informed about medical and genetic research, they value science and technology, and have a positive attitude towards scientific research (Snell and Tarkkala 2019). In the surveys conducted regularly by the European Commission, Finns consistently show high support for science and research and high levels of trust in society (e.g. European Commission 2021a; 2021b). This wider acceptance and support of research in society is considered crucial, since biomedical and medical research constantly requires participants whose involvement hinges on the trust they have in research and its organizers. It is a matter of whether the public sees research organizations and their research as important, justified, responsible, and accountable. For example, in societies where general trust in the public authorities and politicians is not particularly high, the willingness to participate in research tends to be low as well (European Commission 2010; Gaskell and Gottweis 2011; Gaskell et al. 2013).

The importance of positive public attitudes and 'research-friendliness' already suggests the significance of a wider, supportive, and enabling environment for conducting studies and doing research. There is a special section on the homepage of the Finnish pharmaceutical industry collective, Pharma Industry Finland (PIF), titled 'Engaged people and culture of trust', where they describe Finland as 'a research-friendly environment' whose legislation 'sustains the development of the pharmaceutical industry'

(PIF n.d.). Similarly, FINBB, the national biobank cooperative, claims on their homepage:

> Finns trust researchers and the Finnish healthcare system. Therefore, they are willing to participate in public screening programs and to donate their samples to biobanks. (FINBB n.d.)

Clearly, research cannot exist in a vacuum, and much more is needed than bodily substances such as blood, urine, or saliva for conducting research and producing meaningful results in science (see Tarkkala 2019). Doing research is a matter of infrastructure, data, regulations, participants, expertise, technologies, and methods, to name just a few requirements.

Thus, even though it is the unique genetics that is advertised as making Finland an exemplary environment in which to undertake studies and seek collaboration, genetics alone is not enough to attract and sustain interest and collaboration and frame Finland as the place to be. Indeed, the institutional and regulatory environment is also marketed as a characteristic that makes Finland a lucrative location for biomedical R&D (Tarkkala 2019). The acknowledgement that the wider context matters for genetics and research is not, as such, new; nor is it being used in the public presentation of the Finnish R&D environment for the first time. The availability of data and the positive attitudes of participants have long been among the crucial prerequisites. For example, in 1999, several prominent researchers wrote in their article:

> The example of Finland shows how successful research of genetic diseases has been based on well-recorded population histories, the efforts of skilful clinicians and high quality health care. These advantages have produced reliable diagnoses and excellent population and health care registers, but even more importantly a high level of basic trust by the population of genetic research and consequent high participation rates in genetic studies. (Peltonen et al. 1999, 1920)

In this chapter we examine the different layers of the narrative of Finland's exemplary research conditions—from the unique

population, through the national health data registers, to the popular trust that together make Finland an allegedly distinct environment and inviting test bed for biomedical research and innovation. First, we start by contextualizing the notion of a genetically unique Finnish population in relation to research populations in biomedicine more globally, and present how these populations can be viewed as being made, and as matters and outcomes of practices. We also discuss the complexity of both the populations and practices that created them. We then move on to present how Finnish society and its institutional history are inscribed in so-called Finnish samples and data, since many socio-historical and institutional factors are essential prerequisites for the latter's perceived content (Tarkkala 2019). After that we present the imaginary of the positive and engaged population, willing to participate in research, as one important factor that is seen to contribute to Finland's being a unique test bed, and critically discuss how this positive attitude is utilized in marketing. We point out that a positive attitude towards research, as such, does not necessarily mean actual willingness to participate, and therefore this cannot simply be assumed, and certainly cannot be used to justify abandoning consent procedures (Snell and Tarkkala 2019). Finally, we conclude by reflecting on the assumptions of the unique, willing, and engaged population attached to genomics in Finland.

Constructing unique populations

In everyday life, we tend to understand population as a group of all individuals living in a specific region, usually a nation state, city, county, or borough. However, the notion of population is much more fluid, flexible, and technological than it seems at first glance. In the social sciences, especially in science and technology studies, populations are highlighted as compounds of artefacts that are made or constructed within scientific, political, and social practices (M'charek 2005; Ruppert 2011; Tupasela and Snell 2015). In research, the construction takes place through the selection of individuals for studies that researchers consider representative of

the general population, or, in the case of Finland, Finns more generally. A population is, therefore—at least in terms of research—rather seen as a group of people pooled together for certain purposes, based on some shared characteristics defined according to the needs of the study. It is noteworthy that the notion of population emerged in a political context in the 18th and 19th centuries when nation states in Europe were under formation and central governments started to acquire numerical information about people living in their territories, which resulted in the alliance of state power governing people as a population and statistical knowledge production that conceived of populations in terms of quantity and measurement (Hacking 1990; Desrosières 1994; Porter 1998; Foucault 2007; Krieger 2012). This political embeddedness of the notion of population is not irrelevant for research populations and their malleability. Both researchers—demographers, and medical and social scientists—and public administration construct a population around shared characteristics that make people somehow 'the same': for example, men over 60 years of age, household members, or infants (see Biruk 2022). This kind of pooling and categorizing allows examination and metrics about populations, and comparison of populations, as well as the governing of a population or intervention in it by public authorities. As Cal Biruk (2022, 315) reminds us, a population 'is socially constructed, contingent, and malleable, reflective of the political, social, and historical context of those who create and use it'.

Biomedicine and genomics both require and establish populations in their work. First, a clearly defined and thus 'similar' group of people is considered research material from which one can expect to get meaningful results instead of mere noise due to the high variability in a random group of people. Second, by doing research on certain populations, the concept of that population becomes reinforced and strengthened as a population of some specific sort. Surprisingly, the populations of biomedicine are as much constructed and malleable as are, for example, the populations of demography, a malleability that leads to a more fundamental feature, namely, that related to the similarity and difference between

a population's members. British-Ghanaian philosopher Kwame Anthony Appiah (2005, 151) has noted, for example, that 'upholding differences among groups may entail imposing uniformity within them'. This observation points towards the active making and construction of similarity and difference both among members of a given population and between different populations.

According to Nancy Krieger (2012, 634), 'who and what defines and makes a population has everything to do with whether population means are meaningful or meaningless, with profound implications for work on population health and health inequalities'. Krieger argues that populations can be constructed as meaningful depending on what is needed for a particular research task. Therefore, how a population is sampled or studied (who is included and excluded) has a significant impact on what can and cannot be said about it. This observation can lead to two types of selection regarding biomedical research. The first relates to who is included in a study population, while the second type relates to which study populations are used for research. Although researchers may strive to collect and include people in a study population to make it as representative as possible, there are always different types of selection bias involved. In the UK Biobank, for example, several biases have been identified which have emerged because of the way population sampling was conducted (Fry et al. 2017). For instance,

> UK Biobank participants were more likely to be older, to be female, and to live in less socioeconomically deprived areas than nonparticipants. Compared with the general population, participants were less likely to be obese, to smoke, and to drink alcohol on a daily basis and had fewer self-reported health conditions. (Fry et al. 2017, 1026)

In large international comparative studies, then, biobank or population cohorts may be used based on the availability of data or the ease with which samples and data can be used and analysed. This may result in outcomes that provide only partial or incomplete information about the prevalence of disease around

the world, or results that do not offer generalizability to the wider population. Therefore, there are often negotiations over which populations are included in biomedical research collections, such as those held in biobanks, since inclusion reinforces the existence of a certain population, as more knowledge is gained from its members. On the other hand, inclusion makes it possible for this population to benefit from the possible results (see Epstein 2007).

We have already seen in Chapter 2 how understandings of broader population history can be entangled with genetics research. During the 20th century, for example, homogeneous nations and population isolates offered a clear route to studying monogenetic diseases; it was efficient to work with a population that shared similar characteristics, since a mutation associated with a single gene is easy and efficient to detect, even more so if you already know where to look. Therefore, the genetic homogeneity of the Finnish population was considered a treasure for scientists. Similar promises for biomedical research and development have been made about other populations as well, such as Old Order Amish, Hutterites, Jewish communities such as Ashkenazi Jews, and Sardinians (e.g. Mitchell et al. 2015; Thompson et al. 2010; Arcos-Burgos and Muenke 2002). These populations have also been used for research because of the extensive records they keep of family lineages or pedigrees, which have allowed researchers to make connections between disease occurrence and inheritance among community members.

The characterization of the Finnish population more generally as being 'genetically homogeneous' has been considered a major asset by the research community; however, since the rise of genome-wide association studies (GWAS), the notion of homogeneous population has no longer been seen as unequivocally beneficial, as we discussed in Chapter 2. GWAS rely on scanning a large number of different genetic markers across the genome and correlating these with diseases, a major approach since the mapping of the whole human genome. Consequently, the needs are different for GWAS, and the promise of unique genetics had to be realigned to match with the new era that was emerging in biomedical

research. The usability and portrayal of Finnish research materials in current research settings had to be convincingly shown to fit with other research populations as well.

This approach was an important change from the study of rare diseases localized and specific to Finland. It applied different methods to identify disease-causing genes, and indicated that the usability of the Finnish population to identify genetic loci for common diseases was relevant to international research—thus providing an increasingly important basis for developing comparative studies involving other Western research populations. As an article by Himanshu Chheda et al. (2017, 477), for example, states: 'We compared the genomic profiles of the 1463 Finns to a sample of 1463 British individuals that were sequenced in parallel as part of the UK10K Project.' The role of Finnish population collections was therefore becoming increasingly international in its significance, not just as a research population used to study itself, but also to make comparisons with other populations. The justification for using population isolates to study common diseases was important for the success of the subsequent internationalization of Finnish research, because if a population such as that found in Finland was considered too weird, too unique, or merely an outlier, it would not be perceived as producing generalizable findings more broadly. The point being made in this chapter, however, is that success in genetic research does not hinge solely on genetics itself; there is much more to say about research populations than attributions of nationality, genetics, or certainty of diagnosis (see also Tarkkala 2019).

Uniqueness is often, as we saw already in Chapter 2, portrayed in terms of the homogeneity or heterogeneity of a population. For example, the Finnish, like the Icelandic population, is advertised as a homogeneous isolate, whereas the Estonian population is promoted as heterogeneous (Fletcher 2004). Whether a population is considered homogeneous or heterogeneous has implications for the types of research for which it is deemed suitable, and the benefits it might offer them. Therefore, certain characteristics of a population are selected to be highlighted for biomedical R&D

to lure collaborations and investments. And because biomedical research is currently international and collaborative, it is important to portray the population as a good fit with wider projects and programmes, meaning that the population cannot be too odd or stand out from the bigger mass of samples from various origins. In practice, portraying a population as special and homogeneous, and portraying it as having a good fit with other populations and producing generalizable results among this bigger pool, might be at odds (Tarkkala and Tupasela 2018).

Conceptualizing homogeneity

Expert discussions of genetic differences and uniformity among people and nations have proliferated during the past thirty years (Lipphardt 2014). As the American anthropologist Nadia Abu El-Haj (2012, 22) has argued, genetic markers have been understood as '"mere" indexes of ancestry and origin'. From this perspective, genetics is considered a neutral representation and archive of human origin and ancestry. Some of the early genetic studies of various human populations, such as the HapMap project and the Human Genome Diversity Project, looked to provide scientific explanations of genetic variation between human populations. Their main shortcoming was that to do so they used a priori assumptions of what constitutes meaningful genetic difference in the first place (M'charek 2005). Therefore, to study the genetics of difference, researchers relied on political and cultural definitions of populations, which may not have been good markers of biological difference (Tupasela 2022a).

In the Finnish context, geneticists have used the notion of homogeneity as a feature that helps to distinguish the Finnish population from other research populations. Although the conceptual meaning and significance of homogeneity, and conversely heterogeneity, may appear self-evident, it is important that we briefly discuss their definitions, since they have an important bearing on international discussions of population isolates and, in Finland, of the Finnish population. As we saw in Chapter 2, nowadays the

origin story of the Finnish population connects with Finnish Disease Heritage (FDH) studies, and this has built and reinforced certain narratives about the cultural history of Finns more generally. The founder effect, or bottleneck theory, which geneticists and doctors have used to explain the enrichment of certain genetic traits, has in Finland also come to characterize and stand for the wider population history of the whole country.

Although the notion of homogeneity is commonly used to describe the internal similarity of population isolates, different meanings can be attached to the concept. Despite the complexity of the human genome, we share a staggering 99.9 per cent of our genetic material with other humans; the remaining 0.1 per cent gives rise to the vast variability (Lander 2011), and discussions of homogeneity and heterogeneity address this small fraction. As a Finnish researcher noted of homogeneity and its relation to genetic diversity,

> I would not use the term 'homogeneous'. I would say that in Finland there is little genetic diversity. I know that Sally used the term homogeneity to mean little diversity. They are not the same thing because homogeneity means that it is the same all over and that is not entirely true in Finland. (Interview with geneticist, 2012; see Tarkkala and Tupasela 2018)

This interview excerpt highlights some of the salient features of the Finnish discussion on homogeneity. First, the researcher makes a distinction between the latter and variability in relation to Finns, most notably pointing out that Finns are not homogeneous, that is to say, 'the same all over'. Second, the quote opens the discussion of the distinction that must be made in relation to the criteria of homogeneity in genetics. A useful distinction that some Finnish researchers have introduced concerns the difference between the Icelandic and the Finnish populations; although both are considered population isolates, the Finnish population is considered more homogeneous, since there is less internal variability (Helgason et al. 2000; Salmela et al. 2008). In other words, despite the relative isolation of both Finns and Icelanders, the founding

population in Iceland had a higher level of genetic diversity than that in Finland. Currently, Finland has a population of about 5.5 million and Iceland only 320,000; however, Iceland's founding population consisted of around 5,000 individuals while Finland's is estimated to have been around 1,000 (see Andersen et al. 2016; Wang et al. 2014). To complicate the discussion even further, the point can be made that even if Finland has less genetic diversity in its population, its internal genetic differences may be greater than in a population with greater genetic diversity because the differences between the variations may be quite significant. As data scientist Sini Kerminen and colleagues have noted:

> The main genetic division within Finland shows striking concordance with the 1323 borderline of the treaty of Nöteborg. In general, we detect genetic substructure throughout the country, which reflects stronger regional genetic differences in Finland compared to, for example, the UK, which in a similar analysis was dominated by a single unstructured population. (Kerminen et al. 2017, 3459)

Differences between political, cultural, or social definitions of belonging, and the biological demarcation of a specific population, bring additional complexity to the notion of a homogeneous Finnish population. As one researcher commented:

> The biological definition of a population does not work very well with humans. It is spoken of as a type of nation-state-ethnic group, [although] ethnic group is perhaps much closer to it. Our use of the term is also not always consistent, so if I started to think about Finnish populations then I would perhaps group Finnish-Swedes as some type of sub-population, as well as East and West Finns separately. But then we also use 'Finnish population' as an umbrella term under which there are all types of diversity. So, in human genetics I don't think there is a consistent definition which would have [firm] criteria, so it is used differently depending on the situation. (Interview with geneticist, 2011)

As mentioned, the criteria by which individuals are representative or part of the Finnish population from a political or cultural perspective may have little to do with biological similarity. Biological similarity can also be constructed depending on the inclusion and exclusion criteria whereby individuals are considered representative of a given population. An example of this challenge is the construction of the Danish reference genome (Maretty et al. 2017), which mapped 'the genomes of 150 healthy Danes selected to represent the normal citizens in order to examine which variations can be observed in the Danish genetic material' (Genome Denmark n.d.). The terms 'mapped' and 'normal Danes' are misleading in the sense that 'mapping' refers to a neutral process of description of a natural phenomenon, whereas 'normal' suggests that there exists some type of measure by which normality is established.

Notions of the genetic homogeneity of a population and differences between populations are also important in terms of reference genomes. According to Alice Kaye and Wyeth Wasserman (2021, 1) reference genomes have two purposes: they provide 'a persistent structure against which findings can be reported' and reduce 'the computational costs and time required to process genomic data by creating a scaffold that can be relied upon by analysis software'. Although reference genomes are presented as merely technical characterizations of a given population, they also implicitly help to delineate forms of population inclusion and exclusion. As a result, these characteristics also play an important role in representations of nationhood through genetics (Tupasela 2021; de Souza and Santos 2014). Although this is not always explicitly stated, there have been many 'spillovers' of genetic representations of Finnishness into popular culture and the media, such as the use of genetics imagery in a stamp collection published by the postal service for the celebration of 100 years of Finnish independence (Pirinen and Kerminen 2017).

Figure 7 exemplifies how homogeneity is constructed in Finland. This image shows a map of Finland with three distinct population groups based on their genetic similarity. The homogeneity

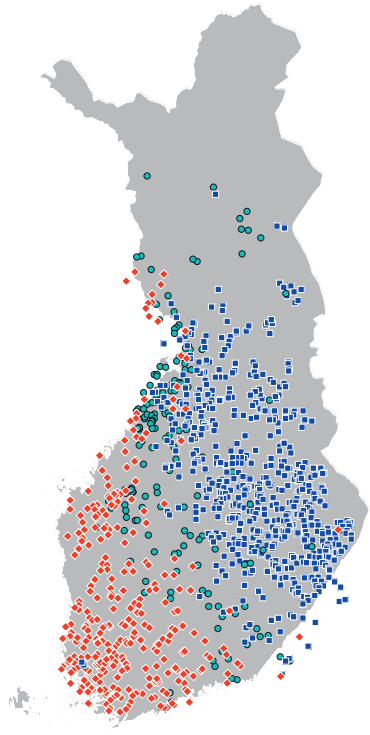

Figure 7: Genetic grouping of 1,042 Finns with their geographical location. Individuals that cluster in red diamonds and blue squares differ significantly from each other genetically, thus representing the so-called duality of Finnish genetic difference. Dots marked in turqoise circles do not cluster strongly with the other two groups. Both parents of the sampled individuals have been born in close geographic proximity to each other and the genetic clustering is said to represent genetic differences of Finns around the 1950s. (Pirinen and Kerminen 2017, released under the license CC BY 4.0).

can be seen internally in relation to those individuals within each group, while national heterogeneity is represented by the differences between the groups. In this sense the image shows both homogeneity and heterogeneity depending on what the reference point is.

Another significant part of the discussion about genetic homogeneity and heterogeneity has to do with the part of human

heredity that is being compared and studied, as there are multiple areas of human genome inheritance, and, depending on the areas of human biological heredity that are compared, the history and inheritance patterns might change. This, in turn, can have significant implications for the discussion, as a quote from a study of Finns expresses effectively:

> The Finnish population in Northern Europe has been a target of extensive genetic studies during the last decades. The population is considered as a homogeneous isolate, well suited for gene mapping studies because of its reduced diversity and homogeneity. However, several studies have shown substantial differences between the eastern and western parts of the country, especially in the male-mediated Y chromosome. This divergence is evident in non-neutral genetic variation also and it is usually explained to stem from founder effects occurring in the settlement of eastern Finland as late as in the 16th century. Here, we have reassessed this population historical scenario using Y-chromosomal, mitochondrial and autosomal markers and geographical sampling covering entire Finland. (Palo et al. 2009)

Considering these factors, any attempt to discuss homogeneity and heterogeneity as inherent characteristics of a given population is fraught with pitfalls and subject to interpretation and contextualization.

Making up a unique society

In the quest to portray and promote Finland as a unique environment in which to conduct biomedical R&D, the genetic composition of the population is only one characteristic highlighted by the spokespersons of Finnish medical genomics and biomedicine. Indeed, the claim of being unique can easily be attached to populations, and this association has frequently been made around the world. What is offered as unique might be a population that can pass as European, as is the case with Argentinians (Lakoff 2012) in Latin America, or Asian, as in Singaporean

medical genomics and biomedicine initiatives (Ong 2016), which would offer the possibility for wider generalization. The attribute of uniqueness can also refer to the ability to serve as an 'innocent' population—one that is not as widely medicated as the populations in many Western countries. This enables more efficient and accurate testing of the 'true' effects of medication instead of pondering whether the results are based on unknown combinations of past medicines consumed by members of the study group (Petryna 2009). Therefore, uniqueness can hinge on several factors, such as genetics, lifestyle, representativeness, or the availability of research subjects, samples, or data. In Finland, the availability of health-related data is often brought up as the most meaningful characteristic contributing to Finland's unique environment. This, as we discuss below, is entangled with the historical trajectories of Finnish society, the state, and other institutions of public authority, and the related collection of data from the population, patients, and clients of public services (Tarkkala 2019).

International state-of-the-art research requires large quantities of samples, pooled together from various sample collections, with a key requirement in terms of usability being their similarity. Only when the samples can be somehow considered 'the same' can analyses based on them be comparable and produce meaningful results, instead of outcomes that simply reflect and repeat variation between them. This underlines the need for the standardization of sample collection, processing, and storage: that is, good record-keeping of where the samples have come from and been stored, and how they have been taken care of (Tarkkala 2019). Simultaneously, it highlights how the suitability of the samples for the specific research setting at hand is what matters, and that suitability must be portrayed convincingly.

Furthermore, the ability to find the very samples that are needed from the vast sample collections requires availability of data about the samples and also the donors. It is crucial to know whether the sample is from a male or a female and has certain genetic markers, if the donor patient has received any kind of treatment or medication and the outcomes, and so on. The ability

to stratify the right kinds of samples and specific research populations produces a notion of uniqueness that does not hinge on genetic homogeneity. For example, even though Finnish biobanks claim to store samples from the 'Finnish population' and aim to 'capture all incomers' to the hospital clinics, in practice, the samples collected and stored for research are collected from all the residents in the country, which creates a population that is far more heterogeneous than so-called homogeneous Finns (Tarkkala 2019). In the context of actual research projects, sample requests sent to biobanks could be pooled together based on similar diagnoses, medications, treatments, disease outcomes, or disease-related markers, which implies that in practice the Finnishness of the samples simply refers to the context of their collection and the kind of data that accompanies them, not to genetically homogeneous Finns as such.

Although the notion of population is malleable and flexible, as we discussed earlier in this chapter, there are also limits on how and what makes a population and its related data. Often the populations that can be formed in the first place are dependent on the already existing data that is usually collected by the states (in the shape of health authorities, statistics offices, etc.) for the purposes of administration and governance. Therefore, the public authorities tend to be the ones that have the much-needed data to be reassembled for the needs of research (Hinterberger 2012; Ong 2016). Obviously, one result of this is that the data are often reorganized according to the logics of governance and administration, thereby highlighting the institutional and social histories of certain countries. For example, in Singapore, the populations that the country is able to offer for research are based on the national classification of the citizenry, which has its origins in the British colonial era and ideas of race at that time (Ong 2016, 13). Singapore thus serves as an example of how the old colonial practices of classifying people into certain groups are now utilized in the field of genomics in the populations on offer in such research settings (Ong 2016; see also Sun 2017).

In Finland, the population and patient data depositories and registers are said to facilitate the stratification of populations for research purposes, offering the means of finding exactly the right patients for a given study (Tarkkala 2019, 76). This acknowledges the exceptional character of Finnish health-related data as the strength of Finland and Finnish biomedical research, and, at the same time, the long history of systematic collection of population data and record-keeping by the state and other public authorities, dating back to the early 18th century. During the 20th century, the collection of population and health-related data became more systematic and routinized, with the decisive development of comprehensive, well-ordered national data reservoirs taking place, parallel with other Nordic countries, during the heyday of planning and building the welfare state, especially between the 1950s and 1980s (Tupasela et al. 2020). At that time, the Finnish public authorities routinely started to collect data about all aspects of people's lives, and repositories of that data were organized to serve the planning and maintenance of public welfare services like healthcare, social insurance and assistance, pensions, and education, as we discussed in Chapter 3. A specific feature of this welfare state data practice was the 'universal' personal identity number (PIN) that was introduced with the pension and health insurance reform in Finland in the early 1960s (Alastalo 2009; Alastalo and Helén 2022). The PIN identifies each individual citizen throughout the fabric of welfare services and administration and is used both to allow the allocation and provision of pensions, subsidies, and services, and to keep track of the individuals in different services and registers. It is also fundamental to research because, with the help of PINs, several types of information can be combined on each individual from a variety of registers, which, in the context of biobank research, allows such compiled datasets to be precisely connected with the samples. Welfare-state data sourcing thus provides an essential element of the usability of biobank samples and widens the purposes for which the samples, or only the so-called 'real-world data', can be used in biomedical R&D. In this, Finland resembles the other Nordic welfare states in how these countries

promote themselves to potential collaborators and investors (Nordforsk 2014; 2017; see also Hoeyer 2019; Tupasela et al. 2020).

Finland is by no means the first Nordic country to advertise the value of its health data. A biobank and health data repository were initiated in Iceland about a decade prior to Finland's, with strikingly similar reasoning behind why it would be the place to be for biomedical R&D (Tarkkala 2019). According to Pálsson and Hardardóttir (2002, 276), Iceland was portrayed through its 'small, relatively homogeneous, and comparatively isolated' population, accompanied by the national 'passion for keeping genealogical records', as well as a 'strong and centralized medical service' with 'extensive medical records'. So, as in Finland, it is the wider environment, data, and public records that became identified as important elements of Icelandic offerings. In terms of data, Icelandic census-taking commenced in 1703, and there are genealogical records dating back to the 1650s and even further. Thus, by the turn of the millennium in Iceland, it was already clear that this business is not merely about specimens from individuals, as such, but about the multiple and compiled sources of information about individuals (Rose 2001; 2003; Tarkkala 2019). The availability of data was seen as a special feature; indeed, the availability of medical records made it possible to explore

> new questions on the interaction among a number of variables apart from genetic makeup and genealogical connections, including variables pertaining to lifestyle, physical and social environments, the use of particular medicine, and degree and kind of hospitalization (Pálsson and Hardardóttir 2002, 276).

According to British sociologist Hilary Rose, Iceland was even seen as 'the perfect location' to do R&D because of 'the nation's small size, high quality universal health care, medical records dating back to 1915, purported genetic homogeneity, and large and well-documented tissue bank' (Rose 2003, 78). Similarly, the Finnish public institutions are seen to be able to provide comprehensive data covering practically the whole population, and many advocates claim that the corpus is exceptional regarding

socio-economic factors. Since everyone is entitled to public healthcare, specialized care in hospital clinics included, and it is provided to practically all inhabitants through the universal healthcare system, Finnish clinical data is particularly unbiased in socio-economic terms.

Many promoters of the Finnish biomedical R&D milieu emphasize that there are more advantages specific to Finland than just the bare existence of Finnish health-related data in well-ordered and easily accessible form. Their views of the additional exceptionality of data from Finns relies on assumptions that Finnish public institutions, society, and culture are somehow independently homogeneous, which is then reasoned to involve a certain inner standardization of the population that transcends genetics (Tarkkala 2019). An example of this kind of assumption is a statement in a piece on Finland as an environment for genomics research by Copia (2016), a private think tank. In addition to genetic data and accessible medical records, Copia notes that the

> common diets, easily obtained socio-economic data and the cultural environment also make Finland a unique population for studying and controlling for non-genetic factors for many health-related issues (Copia 2016).

Furthermore, another layer of standardization inscribed in the samples and data results from the allegedly uniform medical education provided in the handful of medical faculties at Finnish universities, which is seen to contribute to the unbiased quality of Finnish data. It is argued that the medical records across the country contain a similar understanding of medicine, since the patients in public healthcare have been treated in a similar manner and according to unified guidelines (Tarkkala 2019).

In addition to unified Finnish record-keeping and uniform practices in medical and educational institutions, some advocates of Finnish medical genetics and biomedicine assume a homogeneity of lifestyle and cultural customs among Finns—for example, in eating and diet—which may also be useful for research in medical genetics and genomics (Tarkkala 2019). In 1998, medical

geneticists Hannele Kangas and Leena Peltonen wrote on the subject that 'relatively similar lifestyles reduce the confounding effects of environmental factors in genetic analyses' (Kangas and Palotie 1998, 2). Thus, Finns sharing a similar lifestyle within Finland produce material that reflects what is studied rather than, for example, random environmental factors. Similarly, Leena Peltonen (1999, 73) stated in an article on the benefits of using genetic isolates in research that 'shared environmental and cultural homogeneity in many isolates might ultimately be more beneficial than actual or assumed genetic homogeneity'. Seen from this angle, the allegedly homogeneous Finnish population is portrayed as a population grown like lab mice in a controlled environment in which healthcare and even lifestyle are very similar for everyone (Tarkkala 2019, 84).[7]

Besides medical records, other kinds of data also make a difference. The registers maintained by the public authorities for administrative purposes can now be used as sources of information on a variety of characteristics that can be considered meaningful for research purposes. For example, the FinnGen consortium (see Chapters 3 and 6) deploys personal data from 11 national population and healthcare registers and 5 previous epidemiological and cohort studies conducted in public research institutions or universities, on top of biobank data. In addition to multiple types of health information like data on drug prescriptions and causes of death, FinnGen uses data about the place of birth and residence, social service use, social and health insurance benefits, and a variety of socio-economic variables, while the PIN enables the selection of such data from the national registers person by person. Thus, the systematic and routine collection and storage of health and other data about individual patients, clients, and citizens within the welfare-state services and administration have, as a by-product, created the potential to pool populations for contemporary biomedical research. Universal healthcare covers the whole population and works according to relatively similar guidelines, treatment protocols, and medical choices. Therefore, the institutionalized and well-ordered data regime of the Finnish wel-

fare state contributes to making data-intensive medical genomics possible, while providing welfare services to the whole population and every citizen (see also Rose 2003; Snell et al. 2021; Tarkkala 2019; Alastalo and Helén 2022).

During the 2010s, healthcare and, to some extent, social service data, became increasingly promoted as the unique characteristic of Finnish sample collections and contribution to global medical genomics (Tarkkala 2019). It was essentially the way healthcare and other public services are organized in the Finnish welfare state that came to be portrayed as an exceptional feature and the key factor in producing the unique biomedical R&D environment. By recording the patients, clients, and citizens, and collecting data for decades as part of the mundane tasks of service provision and administration, the Finnish welfare state created something that was now seen as a massive repository of health-related data. The institutional history of the state, the organization of recording and collection of data from its inhabitants, and the provision of specialized healthcare in hospitals as a universal right, has contributed to the formation of a unique population as something that Finland now may offer to biomedical research worldwide. Ultimately, the existence and availability of Finnish data from the Finnish population praised by the advocates of Finland as the 'best' biomedical R&D milieu are embedded in the welfare state and its institutions, even though the welfare state rarely gets mentioned (Tarkkala 2019).

An engaged and willing population

In 2005, *Discover Magazine* published an article on genetics research in Finland in which Finnish medical geneticist Leena Peltonen estimated that three out of four Finns will say yes if they are asked to participate in research. 'As research subjects, Finns are an agreeable lot', the article concluded (Wheelwright 2005). Willingness to participate is a factor that is also highlighted in more official sources promoting Finnish genetics or exploring its possibilities in global competition. For example, in a report concerning

the founding of an institute for molecular medicine, genetics, and epidemiology in 2005, it was stated that Finnish data and samples are of 'high quality' (see Tarkkala 2019), but 'the positive attitude of Finns, good organization of the health care system and the comprehensive registers of the country' have also contributed to Finland's good standing (Halme 2005, 35–36). The report concerning the establishment of official biobanks in Finland repeated the same message two years later:

> The homogeneity of Finnish genetic heritage, well organized health care and population registries as well as the citizens' positive attitude towards research are factors which have provided an important basis for our research (Ministry of Social Affairs and Health 2007, 13).

The specific mentions of the positive attitude reveal that populations are not only significant insofar as they are stored and packaged in data and as data. In addition, the support and positive attitudes of populations matter, and can therefore be framed as a unique feature and used in the promotion and branding of Finland (Tupasela 2021). The reasoning behind such underlining of positive attitudes is quite simple: if people were against the use of their data for biomedical research, and in general not willing to participate in research, the use of data and research itself would become impossible, a topic we discuss more thoroughly in the next chapter.

The trusting attitude and willingness of Finns to participate is often seen to originate in the early days of FDH when certain families with rare, hereditary, monogenetic diseases started to participate in genetics research, providing research materials and family genealogies for the researchers. The Finnish population's willingness to participate has thus figured specifically in relation to genetics at least since the late 1960s. In the aforementioned *Discovery Magazine* article, the CSO and co-founder of a bioinformatics company Geneos, Tarja Laitinen, said that the people who participated in their studies 'knew that the benefits would be a long time coming' (Wheelwright 2005).

> 'But', she continued, 'Finland is a good place for medical research because people feel positive about it. So as a scientist I value the environment of Finland more than the genes' (Wheelwright 2005).

Today, this idea remains a valued Finnish attribute, framed in terms of citizens' favourable attitudes, support, and engagement. For example, Sitra, a think tank involved also in fostering medical genomics and personalized medicine (see Chapter 6), has published a series of PowerPoint slides about Finland's strengths, with a slide presenting Finnish people as engaged. To them, such engagement (see Figure 8) comprises willingness to participate, positive attitudes, high levels of education, trust in the public authorities, and even being tech-savvy. This special mindset of people in terms of research is acknowledged in both research articles and promotion materials and has led to a characterization of the Finnish population as being willing to participate in and support research. The rhetoric has been connected to the idea that Finns are well educated, which is likely to lead them to support science and research. For example, Business Finland promotes Finland for investors by claiming that '[t]he Finns themselves are engaged and committed to the improvement of healthcare and are very much pro-science and tech-savvy' (Business Finland 2022, 68). Thus, the rhetoric of a willing population is simultaneously about imagining Finland as a technologically progressive country and as having a population that is skilled and knowledgeable. This assumption and approach tend, however, to decrease the space allowed for a critical discussion of proposed changes and their possible implications (see Chapter 5).

In many ways, the repetition of the rhetoric of willingness can be considered as a version of the manufactured consent described by American journalist and writer Walter Lippmann (1922; see also Herman and Chomsky 1988). In this perspective, communication is seen as a form of propaganda that utilizes various types of mass media to generate favourable public consent. In terms of public opinion on biobanking in Finland, there have been very similar

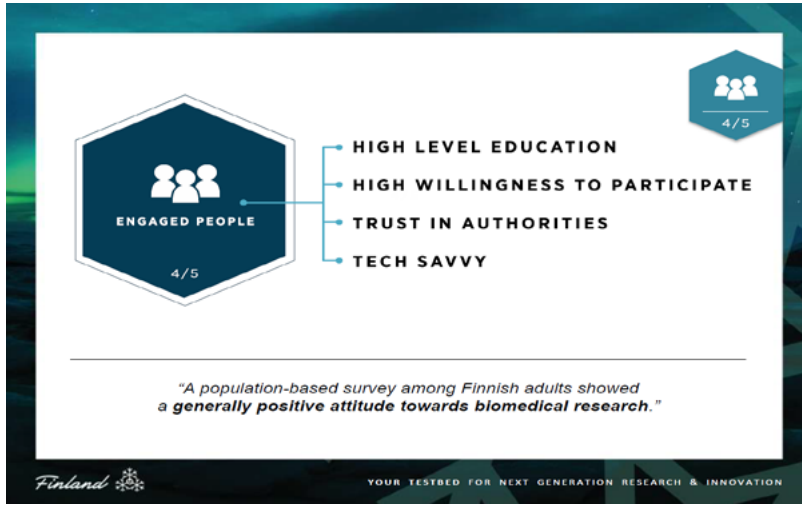

Figure 8: Strenghs of Finland and the Finnish population for R&D. The image above by the consulting company Medaffcon is an example of how Finnish strengths for R&D are often represented. The image below is a slide by Sitra highlighting the benefits of the Finnish population for research. Both images are examples of how the Finnish population is marketed internationally for biomedical research.

Source: Medaffcon 2021; Sitra 2015a. All rights reserved.

trends in the repetition of the claims about a willing population. In an interview with *Health Europa*, Finnish proponents mention 'public support and participation' as key factors in making the bold ambitions related to biobanks and personalized medicine a reality. Finns are described as people who accept biobanking generally and 'are very willing to contribute biobank samples and take part in different kinds of studies and research' (Health Europa, 2018). As a Finnish innovation officer Sampo Sammalisto continued, '[w]e are very innovation-friendly in Finland, so when you want to do scientific or clinical research, recruitment for studies happens much faster than in many other countries' (Health Europa 2018).

It is this supposed fast and efficient recruitment that Finland promises its customers in their promotional rhetoric. The recruitment, then, hinges on the vast data depositories that enable flexible stratification of research populations, as well as biobank legislation that allows the recontacting of people who have specifically consented to it. A similar rhetoric about the supporting and enabling population has also been used in Iceland. Hilary Rose (2003, 81) has noted that the documents of the Icelandic biobank and health sector database (HSD) 'speak of the Icelandic population as not only highly educated but also cooperative – by implication, with scientific and technological research'. Like Finns, Icelanders are described as enthusiastic about adopting new technologies, and as having the wealth to do so. As Rose (2003, 81) wrote, Icelanders' 'cultural enthusiasm for science and technology and its fruits is not shared by most other Europeans'. In a similar vein, Finns have tended to score as quite positive towards medical research in Eurobarometer surveys (European Commission 2021a; 2021b), although this positivity does not translate into unconditional support (Tupasela et al. 2010; Snell and Tarkkala 2019).

Populations on the market

The construction of the notion of a unique and willing population has a specific role to play in relation to the marketing of public resources (Tupasela 2021). Different populations have their own

offerings that can be highlighted for biomedical research markets. A very diseased population might provide information about the diseases it carries, a healthy population about the absence of diseases, thereby hinting at why people have remained healthy despite other factors that would implicate a disease; a so-called 'innocent' population might be beneficial to study, as it has partaken of fewer or no other medications, making it easy to spot the effects of the actual drug that is being tested. Thus, both healthy living and illness can be sources of wealth and innovation. This differs from how states have traditionally understood population health. Population health and well-being are, of course, continuous concerns for the nation states, and the burdens of disease and aging population are issues that are tackled by biomedical research and health-sector-related R&D. The rationale is that if people are healthier, they are also able to work, resulting in the well-being of society, to put it simplistically. Now, however, both health and disease are potential sources of wealth for the national economy, as data on the population (both healthy and sick) can be marketed for R&D purposes. For example, the older samples might be especially valuable, since they already come with information about disease outcomes and care pathways. Some of these samples might also contain information on disease that is hard to get now, perhaps because of more efficient contemporary treatments, which means that the disease no longer progresses as far—at least, not without certain, more recently developed, medical compounds being involved (Tarkkala 2019).

In Finland the population data that the biobanks store are not necessarily valuable because they come from what would be understood as 'genetically Finnish' subjects. Rather, a more important element of Finnishness in terms of R&D might be that one has simply lived in Finland and received specialized healthcare in hospitals. That makes a person potentially part of the Finnish population whose samples are stored (see also Tarkkala 2019). In fact, many biobank collections are for the large part based on the transfer of older sample collections into biobanks (Salokannel et al. 2019). These include, for example, diagnostic samples from

hospital clinics, made possible by biobank regulation that left open the possibility of transferring older collections into biobanks without formal consent by providing a public announcement instead. This was deemed enough, since it was estimated to be too costly and inefficient, and impossible to reach all the people whose samples were stored in pathological and clinical collections, as well as in the research cohorts of THL (see Chapter 3). Such sample transfer is simultaneously an example of the enabling regulatory environment that Finland aims to provide for biomedical R&D and health sector growth. The availability of samples and their uses has, at the same time, contributed to increasing knowledge about them. According to Sini Kerminen and colleagues (2021), this leads to sustained interest, as more and more detailed studies can be done, and therefore 'Finland will likely remain as one of the most accessible and best characterized populations for future research in human genetics' (Kerminen et al. 2021, 2).

A further demonstration of how a population becomes a product to sell, also discussed in the previous section, are the stratified populations to which a biobank has access; biobanks in Finland have, if the donor has consented, the right to recontact the participant and ask if they are willing to participate in a certain medical study: a vital element in how a certain stratified population can be sold as a product (Tarkkala 2019). For example, if a pharma company wants to study certain medical compounds in a specific population, they can ask the biobank to contact members of the population on their behalf, thus enabling the company to approach them about a clinical trial. By contacting these individuals about further study participation, biobanks play their part in facilitating access. After this, the interaction between the individual and the pharma company is direct and governed by the latter's own consent practices. These populations to which access has been granted constitute the product sold by a biobank and are again predicated on the ability to search and pool together certain groups based on existing data on their characteristics. These could be, for example, patients with a gene related to breast cancer or to Alzheimer's disease.

The populations on sale are thus multiple and malleable and cannot be understood in singular, ethnic, or population-based genetical terms (Tarkkala 2019); rather, they are usually based on Finnishness in the sense of the registries and records—the data—which allow the pooling and identification of suitable populations for specific research settings. Recently, Business Finland, an institution fostering innovation and Finnish international trade, specifically highlighted the electronic health records in their webpage:

> There is 100% penetration of electronic health records (EHR) and Finland has the only EHR in the world where clinical and social data are layered on top of each other. Furthermore, all Finns have 100% access to their EHR and [are] encouraged to contribute to it. This adds a very unique third layer of data to the EHR, which is real-time patient-reported outcomes. The fourth layer of the EHRs is e-prescription history and soon patient-drug interaction. (Business Finland n.d.)

This vast database makes it possible to stratify very specific groups of people with very specific characteristics, although the amount of data simultaneously means that expertise is needed to sort all of it into usable form. For example, on 30 April 2023 there were 3,335,185,471 files in the National Electronic Archive of Patient Records (Kanta) alone (Kanta 2023). As early as 2006, a report on the founding of biobanks concluded that attaching the widest possible health data to the samples created 'a possibility to sustain the business interest in Finnish sample collections' (Ministry of Social Affairs and Health 2006, 29). This understanding is still prevalent.

Consequently, during the 2010s, and by the 2020s, public communications portraying Finnish strengths as assets intensified side by side with innovation policy efforts in this field (Tarkkala et al. 2019). Reports and materials were drafted to this end, all delivering the same message aimed at an international audience of potential collaborators and investors (see also Tarkkala and Snell 2022). In 2015 Sitra put together a slideshow for stakeholders to use in

their public presentations, and Business Finland later published an updated version. In this more recent version, Finnish strengths and offerings are presented with the conviction that Finland is 'in a league of its own' (Business Finland 2022, 76). Finland is represented as offering smooth operation and backing to collaborators, especially when it comes to resources and materials, legislation, government, and public support, and a 'culture of trust', as well as highly educated researchers with whom to collaborate (the commercialization and branding of the national population is further discussed in Chapter 6).

Conclusion

In this chapter, we have discussed how the notion of a unique population within the Finnish context is premised on an active construction and representation of the population itself (Tupasela and Snell 2015; Tarkkala 2019). The purpose of this representation has in part related to the need to make Finnish samples and data of interest internationally for research, but increasingly for marketing purposes as well. The more recent push to generate marketing and promotional material reflects a strong interest in making existing resources valuable by appealing to the Finnish population as being interested and willing contributors of their samples and data to innovation. This type of market logic within the public sector has been a common thread over the past twenty years, an approach that has had significant impact on the regulation and governance of human tissue, as well as healthcare data markets in Finland.

In the context of branding and competition, the idea of an engaged and willing population continues to provide acceptance for the wide use of registers for secondary purposes, but also sustains the assumption of a positive attitude towards possible inquiries about further research participation—a necessity for pharmaceutical companies, for example. Simultaneously, the depiction of Finns as an engaged population is an imaginary not fully sustained by the facts. Although Finns tend to have positive

attitudes toward research and science, they do not have a great deal of knowledge about biobanks and biobanking (Snell and Tarkkala 2019). In addition, while people have expressed willingness in regard to having their samples stored in biobanks, they also think that it would be appropriate if they were asked first (Tupasela et al. 2010). This suggests that concurrence with sample collection and storage is not unconditional, but rather hinges on respect and transparency. In the context of the publicly funded, universal healthcare system (National Health Service) in the UK, which is increasingly envisioned as a platform for research, it has been pointed out that 'citizens wish to further the common good without being manipulated into doing it, while at the same time being safeguarded against various abuses' (Sterckx et al. 2016, 177).

Similarly, as most samples stored are the result of the transfer of older collections into biobanks, most biobank participants do not even know about their participation, which makes it difficult to describe these participants as willing or engaged (Salokannel et al. 2019). It is also becoming increasingly difficult to understand the complex ways whereby different kinds of data nowadays travel to be used for other than primary purposes—Finland has, for example, allowed the secondary uses of social and healthcare data and this also covers R&D purposes.

Population uniqueness, therefore, is not just a quality derived from genetic material, but, more significantly, encompasses a broader range of attributes assigned or ascribed to the nation. These qualities are not derived or negotiated through public deliberation within the Finnish context, but rather are elements of the narratives—a 'one voice' (Tarkkala and Snell 2022)—disseminated by various experts and policy development think tanks such as Sitra.

CHAPTER 5

Challenging informed consent

Introduction

This chapter traces Finnish debates and practices of informed consent in medicine and genomics and contextualizes them in international discussions. Like many other countries, Finland has struggled over the decades with finding a suitable solution that would address all the concerns and issues which surround the collection and use of tissue samples and genomic data. This challenge has been exacerbated not only by the changes connected with informed consent practices from the 1950s and 1960s to the present, but also by those associated with the changing use of samples and data.

The discussions on informed consent in relation to Finnish genetics and genomics research cast important light on the changing relations between patients, donors, physicians, researchers, and the state, and they also reflect transformations in the medical and economic expectations attached to genomics (Tupasela et al. 2015). They also highlight an ambiguity concerning the rights of individuals to decide whether and to what degree their information can be used without their knowledge. This has entailed extra challenges for the Nordic countries, since the state and its various organizations, such as the public health authorities, have traditionally collected substantial quantities of data on the population. The vicissitudes of informed consent reflected in the debates indicate the challenges associated with respecting individuals on the

one hand and the interests of the research community and the state on the other.

We begin by outlining the principles of informed consent in international medical research ethics and the impact of genomics and biobanks on the debate, then introduce the origins of the debate in Finland and the long tradition of public health research and register-keeping that have framed the approaches taken. We continue by exploring the 'biobank consent' model adopted in Finland and how it came to be regarded as a pioneering and responsible solution to sample collection, while in practice only being applied to a fraction of the samples utilized in biomedical research. In conclusion, this chapter offers an insight into the different consent practices and paths whereby individuals' biological and sometimes genetic information becomes part of biomedical research and development (R&D) infrastructures in Finland, and how consenting practices are still anything but clear, uniform, and transparent.

Informed consent and the origins of the debate

Medical, ethical, legal, and social science professionals have debated the question of informed consent in biomedical research for decades (Faden and Beauchamp 1986; Beauchamp and Childress 2001). Contemporary discussions are often guided by the principles outlined in international documents such as the Helsinki Declaration (World Medical Association 2013) concerning the autonomy of medical research subjects, and the bioethical Convention on Human Rights and Biomedicine (1996) issued by the Council of Europe. These documents define informed consent as the cornerstone of medical research ethics. The World Medical Association (WMA) developed the Helsinki Declaration in 1964, largely as a reaction to medical abuse during the Second World War. While physicians are its target audience, the WMA also encourages others who are involved in medical research on human subjects to adopt its principles. The Helsinki Declaration states that the goal of generating new medical knowledge should

never take precedence over the rights of individual research subjects, and that these rights and individual autonomy are to be respected with the help of informed consent. This means that the research subject has received adequate information about the study, participates voluntarily, and can refuse to participate or withdraw their consent at any time.

The Convention on Human Rights and Biomedicine (also often called the Oviedo Convention) provides a similar framework for the protection of human rights and human dignity by formulating fundamental principles applicable to medical research and genomics. Just like the Helsinki Declaration, it emphasizes individual rights and informed consent while still permitting medical research without informed consent when the latter is difficult to obtain, and the research is of great scientific importance. Another important document is the Universal Declaration on Bioethics and Human Rights by Unesco (2005), which also stresses human dignity, rights, and autonomy.

The Helsinki Declaration has been revised on numerous occasions, including its extension from research involving physical interventions to medical research 'on identifiable human material and data' that has taken place in the 2000s. This reflects the changing research environment at the turn of the millennium and the sequencing of the whole human genome, which challenged the traditional model of informed consent based on the notion of single research projects. Despite this, there remained strong international interest in respecting the rights of the individual. According to the WMA:

> For medical research using identifiable human material or data, such as research on material or data contained in biobanks or similar repositories, physicians must seek informed consent for its collection, storage and/or reuse. There may be exceptional situations where consent would be impossible or impracticable to obtain for such research. In such situations the research may be done only after consideration and approval of a research ethics committee. (World Medical Association 2013)

The consent debate, therefore, is more than just about the right of patients to have a say in what is done to them in a physical sense. As the research interests in medicine became more and more focused on biobanks and health databases, the WMA complemented the Declaration of Helsinki with the Declaration of Taipei in 2016 (World Medical Association 2016). The focus of this declaration was specifically on health databases and biobanks, as well as the collection, storage, and use of identifiable data and biological material beyond the individual care of patients. The Declaration of Taipei also stressed the importance of informed consent, but recognised the increasing interest of research to gain access to health data. The role of the Declaration of Taipei has been less apparent in Finland than in other countries, since the law on biobanking had already been established in 2013. Although amendments to the law have subsequently followed, Finland has seen itself as a leader in biobanking legislation (Soini 2013).

Since becoming embroiled in the world of biobanking, the informed consent debate has increasingly come to reflect the development of a type of data subjectivity or data relationship between the Nordic welfare state and its residents from which personal data (medical and social) can be extracted. It is within this context that discussions on biobanks and consent have been situated, yet often with a focus on samples more than data, despite the difference between the two becoming increasingly unclear in contemporary data-driven research (see Chapter 3). This has also been apparent in Finnish developments and discussion resulting from the developing legislation. Thus, key questions have remained the same over the past decades: Can consent be truly informed if future uses of samples and data are not yet known? Should a broader form of consent be applied to material stored in biobanks? Should consent be replaced with the possibility to opt out of biobanks and databases? The limits and boundaries of secondary use—that is, in addition to or instead of the original purpose—of tissue and DNA samples and the kind of consent model that should be applied to large tissue collections, especially biobanks, have become new hot discussion topics among international legal, ethical, and social aca-

demics (e.g. Hoeyer 2004; Caulfield 2007; O'Doherty et al. 2011; Steinsbekk and Solberg 2011).

Rethinking informed consent

In considering the legal landscape of data collection in general, it is important to note that within the European context, sample and data collection can be premised on either legal mandate or informed consent. In terms of legal mandate, data and tissue collection is governed by specific national legislation, which allows or creates the legal basis for a specific collection activity. The Nordic countries have a long tradition of this approach, particularly with regard to health and social welfare data, requiring, for example, that doctors report all cases of cancer diagnosis to the various Nordic cancer registries. Under specific legal mandates, individuals cannot choose to exclude themselves from such registries or databases.

The second approach to data and sample collection is based on informed consent, whereby individuals consent to donate or approve of the collection and use of their tissue samples or data. Examples of this include participation in medical research and the donation of samples and data to a biobank. This dichotomy has come to play an important role within the Finnish context, especially in relation to biobank samples and their secondary use.

For medical research, informed consent is obtained from individual participants in situations where the research involves any type of physical or psychological intervention or observation. According to the informed consent literature, this is considered to respect the autonomy of research subjects (Beauchamp 2011), yet from a historical perspective, the notion of formal informed consent is a rather contemporary practice (Hoeyer 2023) because, traditionally, medical authority was not questioned.

Biobanking, however, has been regarded as differing from traditional medical research in that collected samples have already been removed from the donor or patient and consequently do not include any further physical intervention. Informed consent

within biobanking only became of broader interest when researchers began to reuse and repurpose existing samples and data for secondary purposes. At this point, different viewpoints began to emerge, with some arguing that researchers should re-request consent every time samples are used for a new research purpose, and others stating that these additional consent processes are a waste of time and resources. The debate surrounding biobanking led to the development of new terminology and practices in the field of informed consent, with new terms—such as 'broad consent' (Sheehan 2011; Steinsbekk et al. 2013) and 'dynamic consent' (Kaye et al. 2015)—representing new perspectives on samples and data management. Broad consent refers to the process by which consent is sought for a set of research interests, such as the study of heart disease or diabetes, not just for a specific research project. Dynamic consent, on the other hand, means that individuals who have consented to the use of their samples and data can limit or broaden their consent over time depending on their interest, restricting their participation or the use of their samples and data to specific forms of research, while disallowing it for others. These terms also indicate the heterogeneity of approaches that were developing in relation to the management and governance of human tissue samples and data.

In addition to broad and dynamic consent, even more profound concepts such as 'open consent' began to circulate in the discussions and literature on consent. According to Lunshof et al. (2008)

> Open consent means that volunteers consent to unrestricted re-disclosure of data originating from a confidential relationship, namely their health records, and to unrestricted disclosure of information that emerges from any future research on their genotype–phenotype data set, the information content of which cannot be predicted. No promises of anonymity, privacy or confidentiality are made.

The notion of open consent emerged in relation to the developments in large genomic studies, such as those developed in the

UK Biobank, which sought to connect genomic data with healthcare information on thousands of people.

In addition to open, broad, and dynamic consent, other approaches to large national tissue collections were beginning to accommodate both respect for individuals and for the state and researchers' interests; for example, 'opt-out' and 'opt-in' solutions were explored to manage research participation and the secondary use of data. The idea behind these terms was that individuals could choose for themselves whether they wanted their samples to be used or not, although samples and data from individuals might be included in a biobank by default. Only by specifically choosing to opt out would samples and data not be allowed for research use. Newborn blood spot biobanks in Denmark, for example, took this approach. However, as Nordfalk and Hoeyer (2020) have shown, opting out has also had its problems in relation to individuals.

Another important debate related to informed consent has been the issue of returning research results and particularly incidental (or secondary) findings (Wolf et al. 2008). The debate focused on the contradiction between the ethical duty to return research results to individuals and the right not to know—the ethical principle stressing the rights of the individual to not be subjected to knowledge they have not asked for (Tupasela and Liede 2016). This debate around incidental findings led to the development of guidelines, for example in the UK Biobank, where medical imaging technology was in use and was producing many incidental findings (Gibson et al. 2017). The fear was that if not properly addressed, using new technology solely for research purposes could cause mistrust in the public if the public were to perceive that the research, in some way, disregarded the needs of biobank donors. In the USA in 2013, the American College of Medical Genetics (see Green et al. 2013) issued recommendations regarding reporting findings in clinical genome sequencing, advising laboratories to specifically analyse 56 genes. These genes were associated with severe diseases that are preventable if identified early. In Finland, however, these developments have not generated

a need to establish specific guidelines for how and when to report incidental findings from biobank studies.

Roots of the debate in Finland

In Finland, the consent discussion got started in the early 2000s with two significant documents. In 2002 a three-member expert group working under the remit of the Finnish National Advisory Board on Research Ethics (ETENE) and the National Committee on Medical Research Ethics (Tukija) published a memorandum on the use of DNA samples in epidemiological research (Aromaa et al. 2002). In many ways, the memorandum was an important milestone, marking the beginning of the contemporary consent debate in Finland. The purpose of the working group was to explore the possibilities and challenges related to reusing DNA samples in large, publicly funded tissue collections for new research questions and projects, and whether it should be necessary to regain informed consent. Leaving out the ethical questions related to commercial reuse, the working group sought to guide local ethical review boards in navigating a new terrain that was proving ethically, legally, and socially challenging. The opinion of the working group continued to be reflected in more recent legislative changes in Finland, including the Biobank Act of 2012 and the law on the secondary use of social and healthcare information that came into force in 2019.

The memorandum crystallized some of the important contemporary topics under discussion, contextualized them in Finland, and framed the paths of the Finnish consent debate. First, according to the memorandum, re-consent is usually not necessary for reuse of samples, since this does not require any further physical intervention. Second, the expert group aligned DNA and tissue samples with other social and healthcare data that researchers can use without having to gain consent from or notify individuals about data use. Thus, the memorandum did not support the idea of genetic exceptionalism—that genetic data are qualitatively different and more sensitive than other types of data—which was

a popular line of thought in the beginning of the 2000s and continues to be salient to this day (Garrison et al. 2019; Martani et al. 2019). Third, as the focus of the memorandum was on large epidemiological research projects that require extensive population data, the difficulties of recontacting people were emphasized. Eventually, this viewpoint came to be applied to other types of research as well. The memorandum stressed the impossibility of getting consent from seriously ill individuals and underlined the fact that only 50–60 per cent of the population tend to answer to re-consenting inquiries. This drop-out rate would be highly detrimental to conducting population research, and consequently recontact might not be a good option.

The discussions continued, but there seemed to be a consensus about the inapplicability of informed consent to storing genomic data and biological samples. As bioethicists Helena Siipi and Kaija Rossi (2003) noted, the condition of informed consent is not met in the most common model of biobanks, where samples are collected for unspecified further studies. In 2004, Finnish researchers were arguing that the existing legislation regarding informed consent was not adequate to address the needs of biomedical research. As one article noted,

> Genome research does not appear to fit the mould of existing legislation on patient data. The principle of informed consent is from the outset inappropriate for genomic research. Even broad consent assumes that one can describe clearly to the research subject the studies that their samples will be used in and that samples will be destroyed after they have been used. (Käpyaho et al. 2004, 8.)

The second important publication was produced by the Finnish parliament's Committee for the Future (2003), which published a technology assessment statement on the '[s]ocial and legal challenges of human genome and stem cell research' that emphasized that one of the key ethical, legal, and administrative issues is the question of informed consent for the use of an individual's genetic information. It introduced the idea of 'enlarged informed consent', which resembles what is typically called 'broad consent' in

the literature, as it extends informed consent from the research of some specific disease to the research of all diseases important for public health. The committee stated that the justification for enlarged informed consent should be examined, noting that

> The Committee for the Future is of the opinion that examination should be carried out and conclusions drawn in the near future on whether it is justified to use the so-called enlarged informed consent to collect genetic information. If necessary, legislation should be amended to enable enlarged consent. (Finnish parliament's Committee for the Future 2003, XVI)

The committee's technology assessment statement relied on a report by professor of anatomy Martti Parvinen, and futures researcher Osmo Kuusi, based on interviews with Finnish experts such as medical doctors and philosophers. In the report, they argued that the idea of enlarged informed consent could be very appropriate for Finland.

> There must be a very large number of people in Finland – possibly even the majority of the population – who are ready to give their consent to the free use of their genetic data, at least for purposes important to public health, even when well informed about the possible risks. This would be the case especially if they were informed equally about the expected benefits in addition to the risks. If the sympathy of the population could be demonstrated by a scientifically valid attitude survey based on sampling, the ideas expressed about the ethical dubiousness of enlarged informed consent would lose much of their edge. (Finnish parliament's Committee for the Future 2003, 23)

This quote effectively demonstrates the potential of a willing population, described in Chapter 4. The idea that Finns are supportive of research and willing to give their samples has continued to this day, and it also has a connection to the consent debate.

What is particular to the Finnish consent discussion is the balancing between legal mandate and informed consent as justification for sample and data collection. As a Nordic welfare state,

Finland, along with Sweden, Norway, Denmark, and Iceland, has collected extensive tissue collections over the decades as part of large public health campaigns and research projects, as described in Chapters 2 and 3. In many countries, large DNA databases and tissue collections were led by the pharmaceutical industry, but the collections in Nordic countries were mainly publicly funded. While many of the sample collections were gathered with specific consent for the original research, in some cases samples were gathered for diagnostic purposes without consent for research, but were made into a research collection by legal mandate. This is exemplified by the Finnish maternity cohort, which consists of serum samples drawn during the first and early second trimesters of pregnancy for the screening of venereal disease. The samples were gathered from almost all pregnant women from the 1980s onwards for diagnostic purposes, but were stored as a research collection based on the legal duty of the National Institute of Health and Welfare (THL). Only since 2001 has there been an obligation to ask for informed consent for research use of the samples (Salokannel et al. 2019).

While many of the samples have been gathered using informed consent, Finland and other Nordic countries have had a long history of collecting different types of register data from the population on the basis of legal mandate. Gathering information for population registers had its origins in the 18th century, but the centralized collection of systematic health and social welfare data and their use in research started in the mid-20th century. Gathering data for population registers is mandated by law and does not require the informed consent of citizens; nor does it enable opting out of the public registers. These registers have served an important function over the years in providing policymakers, politicians, and civil servants with important population data that can be used to develop governance and healthcare and welfare policies. What also sets the Nordic countries and their debates apart from most other Western countries is the fact that the Nordic countries use a personal identification number (PIN) that can be used to track and identify individuals across numerous different public

and private databases, including healthcare records (see Alastalo and Helén 2022). The PIN also enables different datasets to be combined relatively easily. In many countries, such as Germany, the idea of individuals having a PIN that can be used by different authorities to track individuals is considered ethically and legally problematic.

Before the Biobank Act—the medical use of human organs and tissue

As we described in Chapter 3, the Finnish Biobank Act came into effect in 2013 after five years of discussions and studies of public opinion (Sihvo et al. 2007), market analyses (Käpyaho et al. 2004), and expert consultations. Before the Biobank Act, however, legislation allowed researchers to conduct research on existing tissue samples, a practice governed by the Act on the Medical Use of Human Organs and Tissue (2001/101) also known as the Tissue Act. The 2001 revision replaced a previous version that dated back to 1985.

Up until the 2001 revision, the biomedical use of tissues was not specifically regulated. This did not mean that tissue samples were not used in biomedical research, but rather that such practices were regulated through other laws, such as the Law on Medical Research (986/1999) and the Law on Personal Data (523/1999). With the rise of interest in studying the human genome after the turn of the millennium, researchers felt that the use of human tissue was becoming such an important source of data that there needed to be a law to regulate such activities more clearly. This was seen as an important measure by many researchers to legitimate their research activities, which up until then had been regulated by a multitude of different laws that had not been written with tissue banking and its uses in mind.

In the proposal to the Finnish parliament on the law on organs and tissues (HE 93/2000), the same argument appeared that would be repeated a few years later in a memorandum on the use of DNA samples in epidemiological research (Aromaa et al. 2002)

in which health data and genetic data resembled each other in relation to the data that they contained (see also Tupasela 2008). According to the proposal,

> [t]issue samples can in certain ways be compared to patient records based on the information that they contain. The difference being that with tissue samples the information is in a biological form (HE 93/2000, 29, own translation).

At the turn of the millennium, therefore, the proposal was already beginning to grapple with the question of what type of information genetic information represents and how consent should be managed in relation to it. This also invoked the discussion on the scope of consent and the need for re-consent, issues which would later become cornerstones of the new biobank legislation that came into force in 2012 (see Soini 2013).

Ultimately, when the Tissue Act came into effect in 2001, it allowed research groups to apply for a permit to reuse tissue samples originally collected for other purposes. Mostly these were pathology samples taken and stored after a pathologist had made a diagnosis. The original authority tasked with providing these permits was the National Authority for Medicolegal Affairs (Terveydenhuollon oikeusturvakeskus [TEO]), and between 2001 and 2006, for example, the TEO gave permission to 136 different projects to reuse a total of over 262,000 tissue samples (Tupasela 2004; 2008). This trend has continued to this day, despite the introduction of the Biobank Act, suggesting that different sets of researchers obtain their samples using different legislative frameworks. FinnGen, for example, sources its samples though the various national biobanks, which allows it to gain access to large quantities of samples and data, whereas the Tissue Act tends to be used by researchers needing smaller numbers of samples drawn from those not yet transferred into biobanks, usually diagnostic samples in paraffin blocks that are stored by hospitals.

One of the major shortcomings of the Tissue Act was that it did not meet the requirement of genomics research and the reuse of existing large population cohort studies in Finland. In theory,

research groups could reuse these study samples and data for other research projects by applying to the TEO for a permit, but this would mean that a new permit would have to be sought for every new research project. Given that the large prospective cohorts that were maintained at national research institutes like THL were seen as a continued source of samples and data for numerous studies, a new governance mechanism was needed which would allow for this to happen. What was interesting and of note in the way of thinking at the time within state institutions was the strong belief in expert-led decision-making and the role of tissues in improving innovativeness (Snell 2009). For example, as a biobank working group report noted in 2007:

> Research sample collections collected with public funding, diagnostic sample collections and related information can be seen as being a part of the infrastructure that supports research and innovations, whose efficient utilization can be seen to benefit the whole society. … In biobank research, the interests of the researcher, the research participant and society are parallel. Biobank research produces significant new research findings. The translation of these findings into products and services that contribute to public health requires partnerships with the private sector also. Finland's prosperity is based on the generation of innovations, their uptake and the creation of new businesses. (Ministry of Social Affairs and Health 2007, 13, own translation)

This excerpt from the report highlights how the thinking at the time reflected a very utilitarian approach, wherein samples and data are seen as infrastructure for innovation and prosperity. More significantly, the statement observing that the interests of researchers, research subjects, and society are parallel reflects an almost complete lack of understanding of the origins and purpose of informed consent in the first place.

The perspective and way of thinking in Finland were very different from the approach that was in fashion elsewhere at the time. The World Health Organization (WHO), for example, argued

that individuals should always have a say in how their samples are used, no matter how they were obtained. As it notes,

> Body samples, and the information derived from them, represent two of the most intimate aspects of ourselves. Accordingly, we have a very strong claim to control these elements and their uses. Indeed, in ethical terms, that claim is akin to a property right, in that the primary control should always remain with the individuals who can stake a claim to samples, or the information generated from them. It should be irrelevant where, or how, these elements are gathered or stored. (WHO 2003, 3.1)

This line of argumentation was very much steeped in the notion of public engagement in setting up biobanks and genetic databases as a precondition for maintaining trust and legitimacy between the public and the medical research community. The WHO report, however, also indicates an ambiguity with regard to informed consent and genetic databases, as implied in the following:

> The value of databases derives from the collective nature of their data. Often, the prospect of direct individual benefit is minimal. Thus, the justification for a database is more likely to be grounded in communal value, and less on individual gain. And, while this is not to say that individual protection should be ignored, it leads to the question whether the individual can remain of paramount importance in this context. (WHO 2003, 2.3)

The tension between individual rights and collective interests becomes apparent when one compares these two sections of the same report. Finland, much like the WHO, was trying to come to terms with the relationship between common interests and potential harm to the individual with regard to biobanking and genetic databases. The WHO report does not define what constitutes 'communal values' or 'compelling reasons', allowing individual states to determine the extent to which they are willing to and interested in emphasizing one over the other.

Many of the international reports and guidelines, which were developed at the beginning of the millennium, also reflect this tension and uncertainty about the relation between individual rights and communal values. In Finland, the communal values and interests, which were highlighted and raised as goals, tended to relate to new innovations and the prosperity that would result from making samples and data more readily available by adopting a more lenient interpretation of informed consent. While the Tissue Act, which preceded the Biobank Act, made allowances for the secondary use of samples, it failed to address the needs and concerns of the emerging field of genomic research. This field required large numbers of samples to be used many times over, without continually having to apply for permission from a government authority. It is within this context that work on the Biobank Act began to take shape in Finland.

Implementing and circumventing biobank consent

The Finnish approach to informed consent in the new biomedical environment was formulated and actualized in the process of preparing the Biobank Act, whose drafting process took several years, as described in Chapter 3. One of the most debated issues was the implementation of informed consent. Some of the authors of this book were involved in mapping public opinion on biobanks and consenting practices and produced a survey, as well as interview and focus group research, in connection to the drafting process (Sihvo et al. 2007; Tupasela et al. 2010). Our research identified attitudes and expectations about biobanking that were in many ways positive, yet, along with trust in the Finnish authorities dealing with medicine, also exhibited a general preference for being asked for consent (Tupasela and Snell 2012; Hemminki et al. 2009). Ultimately, the results indicated an ambivalent situation where broad consent, specific informed consent, and an opt-out model were all regarded as having their strengths. In European comparison, Finns tend to align with people from other northern European countries

and support biobanking, and to be more willing to give a broad consent than the average European (Gaskell et al. 2013).

Concomitantly, the approach to consent in the Biobank Act was formulated as an ambiguous compromise between broad consent (or 'enlarged informed consent' as it was termed in the early Finnish documents) for new biobank samples and the possibility of transferring old collections to biobanks without consent if it were difficult to obtain. In practice, the process for broad consent—which became known as 'biobank consent'—meant that for each new sample collected, the donor would have the opportunity to consent to storage of the sample itself and its associated data for future research purposes. Biobanks must define the scope of the research they will accommodate when they apply to be registered in the official biobank registry. Most biobanks have formulated this in similar ways with very general goals, such as health promotion, identification of pathogenesis, prevention of diseases, and development of products and procedures that advance population health and well-being or medical care.

Internationally, the idea of broad consent was also gaining popularity. Asking for consent each time a new project commenced was regarded as placing too heavy a burden on biobanks. With the idea of broad consent, it was recognized that while the future uses of data and samples cannot be known, general principles concerning their use in research and how biobanks are governed could be foreseen. Thus, people could commit to the broad idea of biobanking and agree to professionals' deciding how their samples will be used, but this was not required for each separate project. Broad consent represented a compromise between offering individuals the right to choose and allowing medical science to proceed without too much bureaucracy. The principles of broad consent were justified by the public good that research might produce.

The Biobank Act also introduced the duty for biobanks to return research results for biobank donors. This part of the legislation was criticized on two accounts (Tupasela and Liede 2016). First, it was seen to place a burden on biobanks, as they don't have the capacity to validate the findings. Finnish biobanks noted that

they did not have the experts, particularly clinical genetic counsellors, who were qualified to discuss issues of risk with donors. Second, receiving results requires active action from biobanks to donors who need to request the information from the biobank. The biobanks could also be the initiators of sharing incidental findings, but only if the person had consented to this.

Legacy samples

It is important to note that Finnish biobank consent only applies to new samples and associated data gathered since the implementation of the Act, which provided for the transfer of existing sample collections—often called legacy samples—to biobanks after an ethical statement and personal notification to sample donors ('registered individuals' in the wording of the Act). This meant that

> an institute of higher education, a research institute, a health care unit or some other unit may transfer the samples collected and analysed in connection with a study initiated prior to this [A]ct's entry into force and the information related to them to a biobank (Biobank Act 2012).

The transfer must be approved by the Finnish Medicines Agency (Fimea) (before 2019, approval was done by the National Supervisory Authority for Welfare and Health [Valvira]). Before the transfer, the registered individual shall be notified in person of 'a change of purpose as concerns the samples and information associated with them'. Thus, the sample donor could object to the transfer.

However, the Act also states, that

> if due to the age or large number of the samples, or for some other similar reason, obtaining the contact information of a registered individual is not possible through reasonable effort, the notification … must be published in an official paper, in a public communication network and, as necessary, in one or more daily papers.

While this was framed as an exception, utilizing public instead of personal notification became the default practice for biobanks (Salokannel et al. 2019). By the end of 2018—five years into the establishment of the first biobanks—biobanks had collected just over 100,000 new samples with the biobank consent model, and over 10 million samples had been transferred to biobanks with public instead of personal notification (Salokannel et al. 2019).

At this point, what was meant by biobanking was rather vague to the public, as one of our surveys demonstrated (Snell 2017). Of 1,000 respondents, only 40 per cent had heard of biobanks, and of those respondents, over 70 per cent said that their knowledge of biobanking was insufficient or non-existent. It was likely that providing information about the transfer of samples in a newspaper or on a biobank's webpage did not reach all the people concerned, especially as most people were unaware that their samples could already be stored in biobanks. Auria Biobank calculated in 2014 that only 0.002 per cent of people whose samples were transferred into its keeping had executed their right to prohibit their use. Many people whose samples have been transferred to biobanks as legacy samples are thus not aware of their data and samples being utilized nor about their rights to know about what has been analysed from their data. This puts people in an unequal position in relation to the possible personal benefits.

The publication of newspaper announcements to fulfil the requirement of public notification has been a long-standing tradition in the Nordic countries with regard to the reuse of samples gathered for large epidemiological collections. As the collections often contain the samples and data from thousands of individuals, researchers have traditionally argued that recontacting all these individuals would be impractical, costly, and time-consuming. This argument also appeared in the much-cited memorandum from 2002 (Aromaa et al. 2002).

Figure 9 is drawn from *Helsingin Sanomat*, Finland's largest newspaper. The small advertisement in the corner of the page is the work of the Finnish Maternity Cohort, discussed earlier in this chapter, announcing the transfer of thousands of tissue

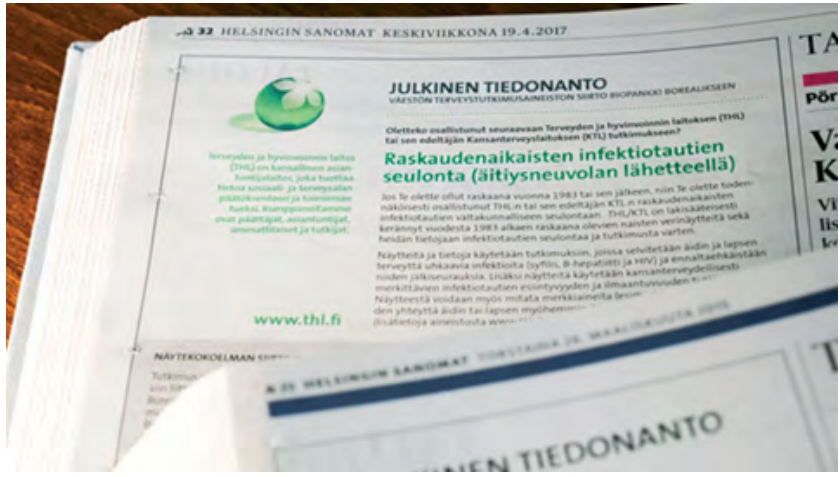

Figure 9: Public communication published in *Helsingin Sanomat* (19 April 2017). The communication informs the public that screening samples taken during pregnancy will be moved to a biobank (see also Tupasela 2021). The public communication states the following: 'Have you participated in the following THL or its preceding KTL study? Screening for infectious diseases during pregnancy (referral from maternity clinic). If you have been pregnant in 1983 or after it is highly likely that you have participated (...) in this national screening programme.

samples taken from pregnant mothers to a biobank. The chances that an individual affected by the transfer will notice the public announcement, however, are not high. What is more significant is that the likelihood of an individual even remembering whether a sample has been taken from her is even lower, which brings into question the significance of the public notice as a form of notification. In some countries, such as Norway, mothers are recontacted about four days after they have left the hospital to make sure that they remember the form that they have signed. This is to ensure that they are made aware of how samples and data from them and their children are collected and stored. Many studies have shown that individuals rarely remember or even read what they have

seen in an informed consent form (Pietrzykowski and Smilowska 2021; Crepeau et al. 2011).

In addition to the reasoning that it is too difficult to recontact such vast numbers of individuals, the research community has expressed concern that if re-consent is sought then there is a chance that individuals may choose to decline; furthermore, it is unclear what should be done with samples from individuals who do not reply at all. Asking researchers to remove a specific sample from their collection would be problematic, since it would decrease the validity and coverage of the collections themselves. Given that many of these large population studies are used to compare changes in public health over extended periods of time, such as twenty years, the degradation of the number of research partners has always been seen as a problem. In light of this, Finland, along with the other Nordic countries, has developed an interpretation of informed consent that has become common practice over the years. Although informed consent was an established practice of medical research, the traditions of register research and the large existing research and diagnostic sample collections in Finland provided working frameworks for conducting research without informed consent.

In an interview, a member of the Finnish National Advisory Board on Ethics described how the idea of transferring legacy samples was discussed in the working group that produced the memorandum:

> It was a brave and open-minded working group that wanted to provoke discussion about what was really worth protecting and tried to interpret as loosely as possible the existing laws on informed consent. … We wanted to challenge the existing notions by asking why one couldn't apply for a permit from the National Authority for Medicolegal Affairs for re-using samples originally taken for research, the same way one can do for samples originally taken for diagnostic or treatment purposes. The legislators and the Ministry for Social Affairs and Health have not yet reacted to this … in part due to the international legal obli-

> gations we have, which state that every time you develop a new purpose for the samples you should re-gain consent. (Member of National Advisory Board on Research Ethics interview 2004; quoted in Tupasela 2008)

After the enactment of the Biobank Act, many positive statements were made about its benefits, especially the so-called biobank consent. The possibility of transferring legacy samples into biobanks without asking for re-consent and also the legacy samples already stored in the biobanks were mainly left out of the public discussion. Many biobankers, researchers, and representatives of public administration and funding organizations identified the broad consent model and the position and rights of the sample donor as among the most advanced and positive aspects of the Act. The broad consent model was touted as the Finnish way in both national promotion materials and numerous public speeches at international biobanking conferences, while the absence of consent procedures in relation to the legacy samples and the huge number of them were issues that were not raised.

Biobank consent as a bottleneck

In practice, however, gathering and storing biobank consents caused practical difficulties for biobanks, and consequently for sample donors as well. One of the problems had to do with restricting the scope of biobank consent.[8] The Biobank Act states that all persons have the right, at any point, to impose restrictions on the use of their samples. Before the enforcement of the Act, parliament's Constitutional Law Committee had emphasized that those who give consent must also be able to restrict the content of the consent (PeVL 10/2012 vp). There was an open answer section in the first biobank consent forms of 2014–2015 where the sample donors could specify if they wanted to restrict the uses of their sample. Biobanks were faced with the problem of how to operationalize these freely written limitations in their consent databases and claimed that it proved to be impossible. Therefore,

samples that came with restricted consent were simply excluded from biobanks. Paradoxically, therefore, in addition to many people not knowing that their samples are stored in biobanks, there can be people who think they have donated to a biobank but have been excluded from them.

The formats of biobank consent have changed several times over the past decade; for example, in 2022, some of the biobanks, including Auria Biobank and Tampere Biobank, stated in their consent form that if someone wants to restrict the use of their sample, they must contact the biobank. No additional information is provided. Helsinki Biobank, however, has a form on its web pages that can be used if someone wants to restrict the use of their samples, which notes,

> Restriction of data and sample use requires you to indicate a reason for the restriction, which you may choose by ticking the appropriate box below (you may choose multiple reasons). After this, please describe how you wish to restrict the use of the information and samples.

Based on the General Data Protection Directive (GDPR), two reasons are offered for such a restriction: 'My information stored in the Helsinki Biobank registry is not correct and needs to be changed'; and, 'I believe that information on me has been used unlawfully'. In both cases the person should specify or explain why this is the case. Both approaches to restricting biobank consent—either contacting the biobank or filling in a separate form—are likely to appear anything but easy to the sample donor, requiring extra effort and skills. Thus, while restricting consent poses difficulties for biobanks, it is made even more difficult for the donors, a situation that effectively demonstrates the ongoing balancing act between respecting personal decision-making and supporting the functioning of biobank operations.

The different legal basis for processing data and samples—informed consent and legal basis for legacy samples—has also meant that if a person wanted to forbid their use, there were two different documents. The correct document had to be chosen for

the prohibition of the use of data or withdrawing consent to be legal, meaning that a person has to understand what legislation applies to their samples and data. In the web pages of Helsinki Biobank there are still separate documents for withdrawing consent and for objecting to processing of data:

> Withdraw your consent. This means that your sample and your data stored in Helsinki Biobank are no longer used for research.
>
> Objection to your data being processed. This means that after we receive your message, we do not process your samples or your information in the biobank. The notice applies to the processing of all such data that the biobank itself administers directly (the sample and data register, the consent register and the code register). If old samples are transferred to a biobank through a notification procedure, the samples and data are thus not covered by the transfer. (Helsinki Biobank 2023)

This is highly confusing for people, and most biobanks have tried to make it less complex, resulting in these biobanks now only having one document, 'biobank refusal', that covers both withdrawing consent and objecting to the processing of data.

Another problem, which was also openly discussed, was that many biobanks struggled to attract consenting donors (Snell and Tarkkala 2019). Some of the clinical biobanks were using a 'capture all incomers' approach, in which they aimed to collect new samples from every patient enrolled in the district's hospitals and healthcare units. Before a person attended a regular blood test or any other procedure involving biological samples, they would get the biobank consent and information forms to read and possibly sign. The return rate of the consent forms was, however, a disappointment for many biobanks. Auria Biobank, which was one of the first to start recruiting new sample donors and put considerable effort into public campaigning, reported that up to 80 per cent of people do not return the consent form or make a prohibition. They simply do not respond. The recruitment process, and the consent system supporting it, were regarded as time-and

resource-consuming—and taking resources from the biobank operations considered 'actual'. Gathering informed consents was therefore identified as impeding the success of Finnish biobanks. Indeed, a report discussing the future steps of Finnish biobanking identified consenting processes as a problem:

> An important current bottleneck is sample collection, which is hampered by the current informed consenting processes that have turned out to be less than optimally effective. Since a great majority of Finns is willing to provide biobank consent and samples, we need new ideas and resources to address this critical bottleneck. (Ministry of Social Affairs and Health 2016, 9)

At this time, international debate on consent in biobanking and health databases was concentrating on the implications of the upcoming European GDPR (e.g. Kaye et al. 2016), which came into effect in 2018, harmonizing many aspects of data protection across Europe. In principle, the GDPR emphasizes the rights of donors to know where their data are being processed, as well as the right to consent to data gathering and storage. However, the GDPR also allows for consent exemptions, and it was on this aspect that the Finnish legislative process focused. With difficulties in obtaining sufficient numbers of people consenting to be biobank donors, the Finnish debate was geared back towards legal mandates for storing and processing samples and data, and the benefits of considering all health data, including genetic data, as register data.

There were several attempts to reformulate the Biobank Act, especially consent procedures, before the implementation of the GDPR. The main arguments used for this were that the broad biobank consent model does not comply with the GDPR and consenting has become so difficult that it can actually hinder research. The debate among biobankers, researchers, clinicians, and representatives of the Ministry of Social Affairs and Health was predicated on the legal basis of processing data. Instead of consent, which was regarded as not being a valid base that would offer the

required protection for donors, the focus turned to legal mandate or public interest as possible grounds for processing data.

Although most stakeholders seemed to agree that consent cannot function as the legal basis for processing data, many still considered it an important ethical procedure for both donors and the perceived trustworthiness of biobanking, and wanted to preserve it. Consequently, between 2015 and 2022, many new combinations and models of consent practices were proposed by the ministry for comments. For example, in 2018 the solution presented was an opt-out model for old samples with broad consent applicable to new samples. The processing of data would be based on GDPR Article 6(1)(e), which states, 'processing is necessary for the performance of a task carried out in the public interest or in the exercise of official authority vested in the controller'.

Applying the notion of public interest to biobanking might seem intuitive at first—the research connected to biobanks aims at promoting national public health and the prevention of diseases. However, the Biobank Act pertains to both public and private biobanks, and many questioned whether all activities, even in publicly owned biobanks, could be regarded as being of public interest. Nonetheless, the ensuing proposals for renewing the Biobank Act in 2020 and 2022 followed the same logic of including some components of consent into a framework that is based on public interest as the grounds for legal processing. Along the way, the idea of an opt-out register for legacy samples presented in 2018 has been forgotten, and the scope of consent has become more limited as well. The most recent proposal, which reached the Finnish parliament in 2022 and was passed with small amendments in early 2023, proposed that consent would not be used as the basis for processing data anymore, and consent would only be required for taking the sample and storing it to the biobank. In practice, once the sample was taken, consent could not be withdrawn. It should be noted that restricting consent to the instance of physical intervention alienates the consent process from actual biobank operations. This latest proposal received negative feedback from many stakeholders and, most importantly, from the

biobanks themselves. In its official statement, the THL Biobank pointed to the fact that this new approach to consent could undermine the trust that biobanking has tried to secure for a decade. This was worded as follows:

> From an ethical point of view, THL considers the effects of withdrawing consent in the current proposal to be problematic, even harmful and damaging to trust. Securing trust is of the utmost importance so that activities based on volunteerism are not jeopardized. (THL 2023, own translation)

Even the draft presentation itself stated that changing the processing basis from consent to public interest weakens the individual's ability to prohibit the processing of their personal data, while at the same time it secures the processing of personal data in biobanking and research and development activities.

Adopting such an approach would place Finland in a somewhat critical light internationally. Finnish debates regularly appear to test the boundaries between individual rights and other policy interests, such as innovation and economic benefits, and the arguments favouring the latter often seem to correlate with nationalist notions of embracing a competitive edge over other countries.

In February 2023, a proposal for reforming the Biobank Act was passed in the Finnish parliament. However, the consent model was revised by the Ministry of Social Affairs and Health after discussions in the Constitutional Law Committee and the Social Affairs and Health Committee. Consent would be asked for sample-taking and it could be withdrawn until the sample was stored in a biobank. This timeframe is very short, however, as it can be a question of hours or days. Therefore, trying to provide greater autonomy to donors, the new version introduced a separate 'approval', which is asked for processing of personal data. Transferring samples and data related to the samples to the biobank thus requires both consent and approval. In connection to this, an opportunity to make a prohibition was introduced. The prohibition covers both withdrawing consent and prohibiting the processing of personal data at the biobank. While this enables

new biobank donors to drop out from the biobank in practice, this introduces yet another legal framework on top of the already existing biobank consent model and legacy samples. Therefore, depending on the time when a person has either given a consent or samples have been taken from the person, what kind of rights the person has varies. This complex layering of rights in relation to samples and data suggests that rather than simplifying practices and legislation, current trends are increasingly complicating the legal and ethical questions surrounding informed consent.

Multiple routes from samples to research and genomic data

As we have shown, there are numerous pieces of legislation and different consent practices whereby samples are turned into genetic data and individuals' genetic information becomes part of biomedical research and development infrastructure. Rather than being fixed, consent requirements, data-processing foundations, and the rights of those registered are under constant debate, while the legislative framework is evolving concurrently. As we wrote this book, the Finnish Biobank Act underwent revision in the Finnish parliament and new consent forms were issued by biobanks. However, there is not yet any evidence on how the new revision and consent forms have worked. Considering these factors, it is still difficult to say something definitive about the future direction and development of informed consent within the context of Finnish biobanking and tissue use.

What is clear, however, is that samples enter biomedical research use via multiple different routes. The consequences of this are somewhat unclear; each evolution of legislative change seems to increase the opacity of the status of the samples. Take, for example, the Tissue Act. In its latest legislative revision in 2019, the law transferred the issuing of permits to access the samples of deceased individuals from Valvira (previously TEO) to local research ethics committees. Under TEO, it had been possible to tabulate the numbers of samples being used from those deceased,

but with the revision in legislation this tabulation was stopped, as there was no provision made for the local committees to maintain the statistics on research permits. Consequently, given the dozens of such committees around Finland, the use of samples from deceased individuals has become impossible to monitor.

While the legislative changes in Finland have been claimed to streamline and simplify access to samples, the consequence for sample donors—including research participants, blood donors, and patients—have been the opposite. Understanding and accounting for how samples and data collected from donors are being used has become much more complicated and embedded in several overlapping regulations. Consent practices are anything but clear, uniform, and transparent. We demonstrate this through concrete examples.

The first one concerns the Finnish Maternity Cohort (FMC) discussed above. The cohort is a collection of two million serum samples that researchers gathered from 98 per cent of pregnant women in Finland between 1983 and 2016, mainly during the first trimester of pregnancy as a routine part of prenatal care. While collected for diagnostic purposes, they were stored as a research collection based on the legal duty of the Finnish Institute for Health and Welfare (THL). Before 2001, asking for separate consent for research use was not required. In 2017, the collection was transferred from THL to the Borealis Biobank in Oulu, meaning that many women probably do not even realize that their samples are located in a local biobank in an area in which they have never lived or received healthcare. The transfer was announced in newspapers (see Figure 9) and on the web pages of THL, when it was made possible for registered participants to opt out, and since the transfer, Borealis Biobank has started to ask for a biobank consent for each new serum sample. It is important to note that samples gathered with FMC consent are not used for genetic research.

Even this single case demonstrates the different ways in which people might exercise their autonomy. We would hesitate to call them donors; certainly, the pregnant women in the first decades of the testing cannot have known that by accepting a routine

medical procedure in prenatal care, they were providing samples for research or biobanking in the future. Thus, a woman who was pregnant in 1999, for example, might not know that her sample is in a biobank, and cannot, therefore, influence the use of her data and samples. On the other hand, those pregnant after 2001 received information about research use, and those since 2017 have been notified about biobanking.

There is a clear problem of autonomy for thousands of women caused by the lack of information about the storage and uses of samples; however, Borealis Biobank has made it relatively easy for women whose samples might be stored in the biobank to prohibit their use. While the legal terminology and process of forbidding the use of personal samples and data differ, Borealis Biobank is one of those with only one form for this—the biobank refusal form—regardless of the legal basis for processing samples and data. Therefore, a person does not have to remember whether or not they originally consented to having their samples taken; nor do they need to understand which legislation applies to their samples and data. Most biobanks have streamlined their consent withdrawal and prohibition of sample and data use formats into a single document, but imbalances remain between legacy samples and biobank consent samples, both on practical and rhetorical levels. It is misleading that biobanks are often depicted as only consisting of new biobank samples gathered with biobank consent, as the following example involving FinnGen demonstrates.

FinnGen is Finland's major national research project on human genomics, launched in 2017 (see Chapters 3 and 6) and advertising itself as an expedition to the frontier of genomics and medicine, claiming that every Finn can be a 'biomedical pioneer' and part of the FinnGen study by giving a biobank consent. However, around 200,000 of those used by FinnGen are legacy samples that have been transferred to biobanks through the Biobank Act (Salokannel et al. 2019), while the other 300,000 samples have been collected with informed consent. In order for an individual whose legacy samples are being used to opt out, they would first have to find out which biobank their samples have been transferred to

and then get in touch with the biobank itself in order to have the sample withdrawn from the FinnGen study. Although FinnGen has more public visibility than traditional research projects, it is highly likely that the majority of individuals do not know that their samples are being used. This type of heterogeneity of sample use has helped FinnGen to analyse a large number of samples and data from Finns.

Biobanks are not, however, the only route to accessing diagnostic samples for secondary use. Despite the continued interest in revising and fine-tuning the Finnish Biobank Act, the use of diagnostic samples has remained relatively steady over the decades. Most of the samples that were transferred to the various biobanks had been collected for specific large cohort studies by various institutions, such as THL and universities; hospitals, however, still had the majority of their diagnostic samples (mostly in paraffin blocks) stored in their cellars, and these have remained a mainstay for researching, for example, cancer in Finland. Since 2001 their use has been governed by the Tissue Act, and, with the exception of samples from deceased individuals, it requires a permit from Fimea (previously Valvira and TEO). As mentioned above, however, despite the attention that the Biobank Act has drawn to the practice, the secondary use of diagnostic samples has not decreased since the Act's introduction, which seems to indicate that the intended beneficiaries of the two Acts are different user groups within the biomedical research community.

Public, commercial, or donor interest?

A somewhat different contribution to the discussions on informed consent has been made by Jones and colleagues (2017), who have suggested that researchers have an ethical duty to utilize data that has already been collected to minimize waste. This approach would also help to maximize the potential of identifying treatments and cures. According to this perspective, once data or samples have been collected, a duty emerges to maximise the benefits accruing from these resources. An example of this approach

can be seen in early Finnish discussions, formulated as follows: 'As a counter question, one can ask whether it is justified from the perspective of Finnish taxpayers not to exploit the enormous commercial potential which Finnish biomedical research has produced during the past years' (Käpyaho et al. 2004, 10). This text, which was part of an early evaluation of the commercial possibilities of the Finnish tissue collection system, demonstrates how the framing of the possible rights and interests of individuals has been explored in relation to those of society more broadly. Obviously, within this context the discussion is heavily influenced by innovation policy discourse; nonetheless, it is an indicator of the ongoing tension between various interests.

Rather than attempting to define what constitutes a benefit (public vs private), this latter discussion has highlighted a utilitarian approach to solving the issue, an interpretation that can also be seen in policy texts when the first Biobank Act was being discussed. The framing of sample collections in this approach is constructed more in terms of an infrastructural or technical issue that benefits all of society, and nicely highlights the ways in which public interests are aligned with those of the research community without any reference to possible concerns or even disagreements. Legislative changes, therefore, have a very utilitarian approach within the Finnish context. It should also be noted that the infrastructure argument is something that has been highlighted within the BBMRI framework, where biobanks are seen more as material constructions than as entailing social relations.

The discussions surrounding informed consent in terms of biobanking and healthcare data reflect structural and practical changes relating to the secondary use of samples and data. While the Nordic welfare states have a long history of using state-collected data for research and population governance, the secondary use of samples and data has only recently taken on a broader significance, particularly from a commercial perspective. State-initiated data collection practices have also differed because biobank data and samples have traditionally been collected for a single, specific purpose. More recently, however, the type of consent obtained

from donors and research participants has become increasingly broad; thus, the data in the databases and tissue collections can also be collected and maintained for secondary purposes. This change in collection and consent practices reflects the new and altered approach to data collection and supply systems.

The discussions about consent in biobanking have increasingly migrated to include debates and discussions on big data ethics. As Jake Metcalf (2021), for example, has noted,

> [d]iscussions of human subject protections in big data research are necessarily discussions of how data ethics will relate to already established norms and institutions that have not yet grappled with the ways in which data research impacts human subjects.

Metcalf's formulation highlights that data use and reuse should take into consideration a much broader set of interests and potential impacts on the lives of data subjects. The context in which genomic research operates today is quite different from the research context that existed in the 1980s and 1990s. The capacity for analysis and data linkage is considerably greater than it was thirty years ago. Given that secondary use may entail a much broader set of applications and uses, which go far beyond the original purpose, it also seems appropriate to take this into consideration when thinking about informed consent. In relation to biobanking and health data, this discussion has centred on how private interests are managed in the reuse of publicly collected and maintained data.

Conclusion

In this chapter, we have explored the evolution of the informed consent discussion within the Finnish context from the turn of the millennium to the present. The discussion reflects many of the ethical, legal, and social concerns that have been explicated over the past two decades in Finland and globally, particularly interpretations of individual rights in relation to the needs and interests of researchers, companies, and the national innovation system. The

Finnish interpretation of consent has in many ways favoured the latter over the former. The idea of respecting physical integrity of people fits poorly in the context of genomics and big data analytics, and therefore ideas such as broad consent and informed consent as a bottleneck for research have become popular in Finland. The rights of the individual are said to be at the core of legislation, and the right to receive information about one's health from biobanks, for example, points to this direction. However, people are not always even aware that their samples are part of a biobank collection and have limited possibilities to influence or opt out of the collections.

Perhaps the most salient feature of these discussions and changes over the past two decades has remained the somewhat tenuous status of tissue samples and individual rights within the broader legal and technical interpretations of Finnish legislation on the biomedical collection and use of human tissue for research.

Another significant feature appears to be the paradox of the so-called 'streamlining' of the legal framework within the Finnish context. Rather than simplifying the system of tissue collection and use, new legislation appears to have increased the complexity and heterogeneity of the system, tending to follow a pattern whereby smaller and smaller interest groups are able to tailor national legislation to fit the needs of their research field. The Finnish Biobank Act, for example, appears to serve the interest of genomics research for the most part, whereas the Tissue Act continues to serve the needs of cancer researchers in Finland. This complicated layering of legislation and changing interpretations of informed consent are increasingly complicating the legal and ethical questions surrounding informed consent. Ultimately, the heterogeneity of the system problematizes the degree to which the interests and rights of individuals, patients, and donors are being sufficiently met.

CHAPTER 6

Good business?

Introduction

In the early 2000s, the National Public Health Institute (KTL) drafted a new research strategy (Eskola 2005) aimed at more effectively monitoring the role and impact of molecular medicine on the different functions and research streams undertaken at the institute. An outcome of the strategy was the more intense utilization of tissue samples and epidemiological data in KTL's repositories, including samples and data from the largest prospective cohort studies in the country and the epidemiological follow-up study FINRISKI. The KTL tissue collections and registers were a major national resource that had been identified as a possible source of innovations in several consultation studies (Technomedicum 2004; Käpyaho and Holthöfer 2003), and the more efficient use of these resources added a completely new element to the organization of KTL's medical research division, namely, commercialization.[9] This emphasis, along with the idea that business activities should be an essential component of the public medical research institution, broke new ground in Finland.

In this chapter we examine the development which led up to KTL's pioneering plan, that of pursuing medical genomics and biobanking both to boost biomedical business and as a business in itself in Finland. We have repeatedly referred to this topic in previous chapters, but here it receives close scrutiny. We start by describing the biotechnology boom in Finland as the context for concerted efforts to make commercial use of Finnish sample and

data repositories and expertise in medical genomics, then focus on early commercialization and its difficulties and successes. This leads into examination of how, in the wake of the 2010s, innovation policy became the main framing for addressing and defining the objectives of biobanking, biobank research, and, more generally, biomedical science. We discuss governmental strategies and implementation of the innovation policy presented in them, that of marketing and branding Finland as the best test bed for biomedical R&D business, as well as organizing the genome industry among domestic innovative companies. We then move on to the formation of the biobanking business model and the related merger of regional biobanks into a national one-stop shop (see Chapter 3). Finally, we examine the commercial aspects and business rationale of FinnGen, which took the flagship role in Finnish medical genomics into the 2020s. The chapter concludes by summarizing discussion about the assumptions, expectations, and actions of Finnish stakeholders in actualizing the commercial value potential of Finnish genomics and its uniqueness in the context of the global biomedical and health data markets.

Biotech boom

The KTL strategy and a few similar plans at the start of the 21st century reflected broader interest at the national level in making better use of resources, such as tissue sample collections, in collaboration with private industry (Tupasela 2006a). The wider frame for these embryonic business models was the innovation boom surrounding biotechnology at around the same time. With the rise and unprecedented success of Nokia in the global mobile phone market, self-awareness of Finland as an innovative high-tech nation and economy spread widely among politicians and the public in the late 1990s. In this somewhat hyperbolic mood, biotechnology emerged as the next major innovation domain in the future landscape of national economic and industrial policy; medical biotechnology was seen as a particularly promising area in which Finnish stakeholders might generate successful R&D

and innovation business. Sparked by this optimistic mood, several efforts were launched in the 1990s, including extensive programmes like the funding schemes of national innovation agency Tekes for boosting domestic drug inventions and development (Tuunainen 2011; Valtakari et al. 2013), and more focused projects like developing and patenting an infection-resistant GMO (genetically modified) potato at the University of Helsinki (Tuunainen 2005).

An early example of the success and expectations connected with medical biotechnology was the first Nordic transgenic animal, Huomen the cow (Figure 10), which was born in 1993 (Väliverronen 2007). Developed at the University of Kuopio in central Finland, the cow had the human red blood cell growth hormone erythropoietin (EPO) gene inserted into its genome. Dubbed within the media the 'medicine cow' and the 'golden cow', Huomen

Figure 10: The first Nordic transgenic animal, Huomen the cow, on display in Kuopio.
Photo courtesy of the University of Eastern Finland. All rights reserved.

was symbolic of the huge economic expectations that researchers and investors alike attached to the biotech boom. The head of the research group, professor of biotechnology Juhani Jänne, set up a company called FinnGene to commercialize the research developed in the group, which was later acquired by a Dutch pharmaceutical company Pharming. Like many other transgenic animals of that period, such as Dolly the sheep (Franklin 2007), Huomen the cow was so plagued by medical problems that the research team did not want her to reproduce. In the wake of the critical public debate in Europe surrounding transgenic animals and GMO plants, Huomen was put down and today can be found stuffed and standing in the lobby of the Bioteknia building on the Kuopio campus of the University of Eastern Finland.

The challenges and expectations associated with the case of Huomen the cow were indicative of some of the tensions that arose in the wake of the biotech boom in Finland and elsewhere in the world. These tensions were mostly related to and generated by new multitasking requirements, as research conducted in public research organizations, such as universities and KTL, were expected to produce innovations and spin-off companies, generate new revenue streams for institutions, and initiate industries connected to the technologies being developed.

The rise of Finnish medical genetics to the forefront of international biomedical science (see Chapter 2) occurred concurrently with the emergence of biotechnology as the locus of innovation policy investments. This 'planetary conjunction' resulted in the view of innovation policy shared by scientists, politicians, and governmental officials that research in medical genomics would be the most important spearhead of biotechnology innovation in Finland. Therefore, investment in a national research infrastructure serving mass-scale genome sequencing in the biomedical context was considered a priority not only in terms of science but also in terms of innovation business.

The vanguard of medical geneticists, especially at the University of Helsinki's Meilahti campus, was very active in giving voice to the economic and commercial potential of Finnish research

and its resources in Finnish sample and data repositories. Leena Peltonen gave interviews and wrote widely about the inspiring prospects of the new genomics era, Finnish excellence in the field, and the economic benefits and business opportunities the new biomedical science would unlock in the future. In 2004, in an article in *Duodecim*, she and her husband Aarno Palotie wrote about 'the great impact on the national economy' that utilization of Finnish sample collections and health data repositories would likely have (Palotie and Peltonen-Palotie 2004); in *Bioteknologiainfo*, the newsletter of the Finnish innovation agency Tekes, she presented the following vision:

> Wouldn't it be great if a Finnish company were able to construct a commercial IT program that, for example, an American physician could use in clinical work? The physician could pass information about the patient's lifestyle, past diseases and genes to the program, and receive a treatment recommendation based on epidemiological knowledge. (*Bioteknologiainfo* 4/2004, own translation)

The geneticists' hype also took a more programmatic form. In 2004, an article titled 'Suomalaiset geenit hyötykäytöön' (Finnish genes should be utilized) was published in the journal of the Federation of Finnish Learned Societies. In it, the authors—including Leena Peltonen and Markus Perola, a KTL geneticist and epidemiologist—declared:

> Both Finnish research and our bioindustry have a great opportunity to take advantage of and benefit from refining our genome information as far as possible, and fantastic chances will open to the Finnish IT industry to develop their capabilities with top-class bioinformatics applications. A common challenge but also a remarkable international opportunity for the Finnish bioindustry, researchers, public authorities and officials and legislators is to develop the utilization of the Finnish genome data into a domain of research and industry that is ethically credible, transparent in

> its activities and acceptable for all citizens in regard to its objectives. (Käpyaho et al. 2004, 10, own translation)

This message was supported by feasibility studies and reports which sought to elaborate the new economic and commercial role of research in medical genetics and genomics. More practical responses to this call to arms were initiatives like the KTL business plan for its new biomedicine research strategy (see above). Another proposal was a plan for the national Genome Information Centre (see Chapter 3).

The University of Helsinki's Genome Centre at Meilahti Biomedicum had been operational for some years, providing sequencing services for domestic researchers (see Chapter 2). Now a feasibility study (Technomedicum 2004; Käpyaho et al. 2004) presented the idea of a national centre that would not concentrate on sequencing services but instead would make the utilization of tissue sample and data resources easier, and thus facilitate collaboration and engagement with pharmaceutical companies, both big and small (Figure 11). The centre would work as a non-profit organization, merely providing a passage to new biomedical information for the commercial sector and its actors. The plan was not very clear about how Finnish samples, health data, and research would generate commercial value, except through new product development by Finnish and foreign companies, yet both the plan and the image exemplify the emergence of reasoning about sample collections and data repositories as a national resource for the commercialization of medical genomics. In fact, the model for commercialization is basically very similar to the one guiding FinnGen operations in the 2020s (see below). However, this plan of the Genome Information Centre did not take off, partly due to lukewarm response to the initiative in professional circles (see Chapter 3).

Underscoring the enthusiasm and initiatives in Finland was an influential and widespread international trend that encouraged or even directed academic scientists—in the natural and life sciences in particular—to develop scientific discoveries and findings into knowledge or applications that would be useful for society,

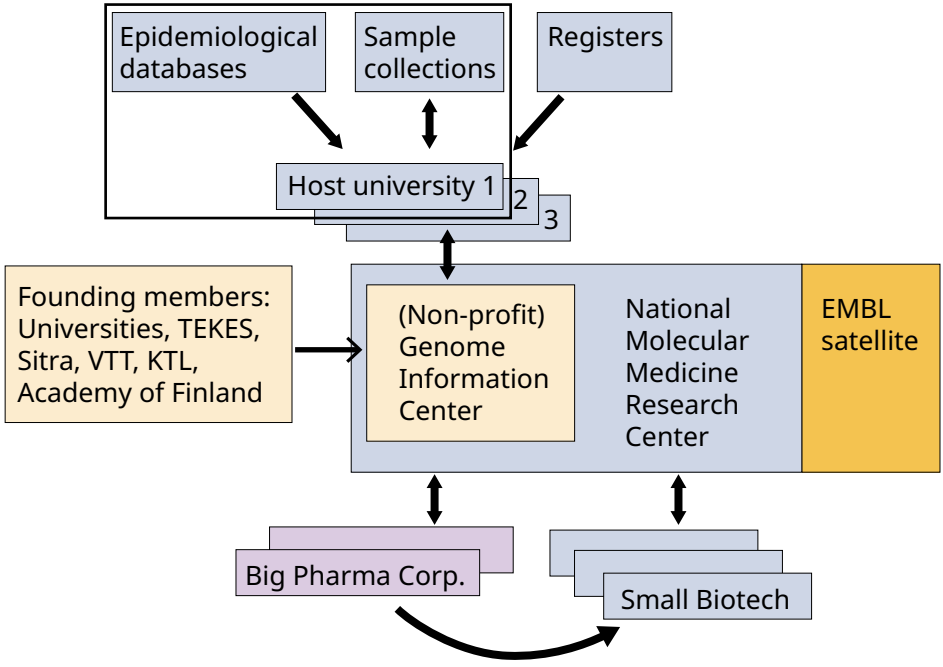

Figure 11: A blueprint for the Finnish Genomic Information Centre (modified from Technomedicum 2003). The main idea of the Genome Information Center was to serve as a buffer or an intermediary organization between public resources and commercial companies. This model was considered less problematic than direct public-private partnerships.

either through commercializing researchers' activities or facilitating their collaboration with private companies. The research literature quite unanimously agrees that this trend has brought a profound change in the organization, financing, and even objectives of science, and given rise to something called 'mode 2 science' and 'triple helix' knowledge production (Etzkowitz 2008), or 'academic capitalism' (Slaughter and Leslie 1999), and the 'commodification and privatization' of science (Mirowski 2011), all terms that designate the development whereby academic institutions and science have become profoundly entangled with private business and profit-seeking.

The lineage of this mode of science reaches back to the Cold War in the late 1950s when a considerable amount of the federal state research and development (R&D) funding for military and space technology in the USA was also directed at private high-tech companies and especially at the joint projects of public research institutions and private companies, with the idea of introducing and sustaining a wider market for new technologies (Mazzucato 2015; Mirowski 2011). In the late 1960s, the Organisation for Economic Co-operation and Development (OECD) and similar international organizations started to encourage the governments of wealthy industrialized countries to launch special policy programmes resembling those in the USA for the promotion of science and technological innovation, as part of their long-term economic and industrial policy (Godin 2015). The poor condition of national economies throughout the Western world in the 1970s, especially in the USA, increased the political resonance of this message. Since the late 1970s, the conviction that scientific research and expertise are the core elements of production and the most important resource for wealth in information societies has become the cornerstone of major policy guidelines throughout the post-industrialized world. At the same time, advocates of the knowledge-based economy have promoted the transformation of basic scientific research findings into technical innovations and their commercial appropriation, claiming this as the most powerful impulse for economic growth both nationally and globally.

In the late 1980s and early 1990s, on both sides of the Atlantic Ocean, an innovation policy action model was consolidated in which scientific endeavours were ideally embedded in public–private partnership. In this model, science was seen ultimately to result in products, methods, or solutions that would be practically useful and commercially profitable. Consequently, national governments and transnational organizations like the EU and OECD intensified their efforts to implement focused innovation programmes and establish funding institutions or instruments to speed up the utilization of new sciences and technologies. As a core element of the policy model, academic and public research

institutions were encouraged and even obliged to think about their activities in terms of business models, to seek business partners from private companies, and to initiate start-ups themselves (Etzkowitz 2002).

The trend directing cutting-edge science towards this mode of knowledge production was reinforced by a policy that loosened regulation of the investment activities of the big investment banks and pension funds, first in the USA in the 1980s and then globally. This meant that a lot of investment capital entered the global finance market, and innovative companies in the fields of emerging technologies like ICT and biotechnologies started to attract venture capital investors (Nicholas 2019). The conjuncture of the innovation policy and the growing interests of investment capital in emerging high-tech formed the context in which biomedicine likewise became quite extensively subject to marketization and commercialization efforts (Styhre 2015).

These international trends also influenced research funding and science policy in Finland. In the 1980s, the government's efforts to increase public and private investment in technological innovation and to establish a specific innovation policy were significantly reinforced. In addition, a great deal of public research funding was directed to innovative R&D through the national innovation agency Tekes, which was a public funding organization whose goal was to support innovation and industrial collaboration. Founded in 1983 and operating until 2018 when it was fused with Finpro (a provider of internationalization services for Finnish companies) and renamed Business Finland, Tekes played an important role in funding large research programmes, which often centred around developing areas like biotechnology and pharmaceutical research and development (Lemola 2020; Miettinen et al. 2006).

Universities and public research institutions were increasingly encouraged to orient themselves towards developing basic science into commercially profitable products and patents or engaging in collaboration with private companies, especially in the fields of engineering and the natural sciences. Telecommunications

was the spearhead technological domain, and, with the success of Nokia, this policy trend became more consolidated and vigorous in Finland in the 1990s. As already noted, in the latter half of the 1990s, innovation policy became increasingly focused on biotechnology and medical genetics, which were seen as a new horn of plenty for the Finnish economy and high-tech companies. Numerous reports and future forecasts were written by a variety of innovation policy offices, and Tekes introduced many programmes encouraging academic and private collaboration in commercializing research findings in the fields of drug development, diagnostics, and bioinformatics. The projects and experiments mentioned above were launched in this context, and in the following section we take a closer look at some efforts engendered by this trend.

Restructuring practices to meet business needs

The KTL strategy suggested that new intermediary organizations should be established to manage the commercial exploitation of KTL's research results and activities: one would be a state-owned company controlling the ownership of intellectual property rights (IPR), patents, and so on, while a second would oversee business collaboration with pharmaceutical corporations and upstream technology companies. The idea of intermediary organizations was derived from the USA university sector, where, following the Bayh–Dole Act of 1980, universities had set up patenting and licensing offices for the management of IPRs and the commercialization of university-derived inventions (e.g. Rafferty 2008). Following this trend, many European universities began to re-evaluate and redefine their relationships with the private sector. Public–private collaboration—something that had traditionally been frowned upon within the academic community—was now becoming the norm, reflecting a new perspective on the role of university research as an engine for innovation, business, and economic growth. Although many university-based research areas, such as medicine and engineering, had maintained close ties

with industry, universities had traditionally retained a somewhat ambiguous or even hostile attitude toward public—private partnerships. In Finland, universities and other public sector research organizations also started to change their response to private sector and business collaboration offers, whereupon KTL likewise felt a need to re-evaluate its role in relation to industry and reorganize its activities to meet the new demands of industrial and innovation policy.

To some extent, the idea of intermediary organizations reflected a political concern. Policymakers and legislators in Finland were wary of possible public backlash if public–private collaboration was not managed appropriately. Although there was an interest in improving the transfer of technology and innovations from the public to the private sector, there were also concerns that without proper buffers between the two, public research initiatives would be influenced and guided by private sector interests. Finnish policymakers were, of course, aware of the controversy that had emerged in Iceland when the Icelandic parliament gave deCODE Genetics monopoly rights over national resources, and neither they nor the politicians were interested in encouraging the development of monopolies; the goal was, rather, to generate conditions that would allow for competition among companies. This openness was seen to be a better way forward in developing clusters of innovation that would have sufficient critical mass for new industrial sectors in Finland. The increased interest in making public organizations like KTL more responsive to the needs and interests of private industry required that specific measures be put in place to serve as buffers between private and public actors.

KTL and other public research organizations were inclined to consider intermediary organizations as the way towards commercialization for another and more practical reason. In order to better meet the demands of innovation policy, they needed an interface that would allow them to transfer knowledge and innovations to the private sector smoothly. Given that none of the existing infrastructures in public research organizations was suited to meeting these new demands, a need emerged to develop

new types of intermediaries to facilitate these practices. Since the resources (tissue collections and population registers) could not be privatized or sold, they would naturally be maintained within the confines of the public institution that housed them. At the same time, however, it was evident that public–private partnerships were unique in the sense that private industry demanded high levels of privacy and confidentiality about contracts and agreements, as well as the content of the research they conducted. These demands and requirements fitted very poorly with the traditions and requirements of transparency and openness practised by public institutions. Consequently, organizations like KTL felt that the best option would be to set up private entities which they would own in order to meet the requirements of the private industry while at the same time representing the interests of the public institutions.

The idea of using an intermediary organization to manage the IPR and commercial aspects of KTL was based on similar intermediary models being discussed at the time in Finland by the main public organizations. The goal of the intermediaries was to serve as a conduit through which research findings and especially IPR could be channelled to private partners without jeopardizing the legitimacy of the public organization.

Figure 12 presents the main components of and relations between the KTL intermediary organization ('National Public Health Inc.') and the private sector ('Business Inc', 'Pharma', and 'Biotech'). The intermediary would serve as a buffer between KTL's epidemiological databases and tissue collections ('Epidemiological data sets, Tissue collections') and the national registers, as well as other important data sources, such as genetic analysis data. The Finnish state would also own a stake in the intermediary, and there would be a mechanism through which the interests of the population would be represented ('Representative of public interest'). The business side would be supported by other Finnish organizations, such as Sitra, Tekes (later Business Finland), universities, and companies.

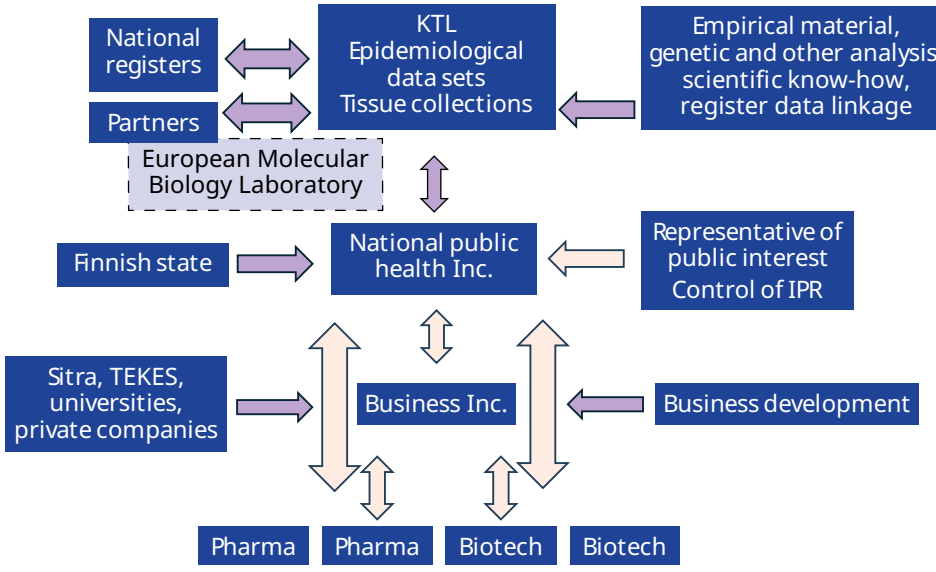

Figure 12: KTL intermediary model for commercialization of research results.

The restructuring of the KTL organization, the plan for the national Genome Information Centre, and the general reorientation of research focus were reflections of broader changes taking place in Finland with respect to the expectations associated with biotechnology. Yet, despite these expectations and the influx of funding, there were a lot of challenges along the way.

A bumpy start

As the patenting and licensing office boom took hold in the USA and increasingly in Finland as well, it became clear that universities and large public research organizations were not necessarily well suited to a business model based on the trend. Many universities in the USA gradually became aware of the risks and small returns associated with patenting and licensing university-based research results (see Baldini 2006); as Hsu and Bernstein (1997)

noted, most universities were not deriving large income revenues as a result. Except for a minority, universities and public research organizations were struggling with their new commercial expectations, often due to the long development cycle and risks related to developing products from patents. Institutional and personal resistance to the new commercial goals being set for the faculties also numbered among the challenges (Owen-Smith and Powell 2001).

The transition by universities and public research organizations towards a more commercially oriented research model likewise faced difficulties in Finland. The newly established patenting and licensing office at Finland's largest university, Helsinki University Licensing (HUL), had to undergo major changes in its patent portfolio only a few years after it began operations when it became clear that many of its patents were of no interest to industry and therefore could not be licensed (Tupasela 2000). Patenting and licensing inventions at public research organizations was not as straightforward as had been originally anticipated.

Conflicts arising from differences in principles also challenged the implementation of the new commercial orientation in some research groups. This is exemplified by the development and commercialization of GMO potatoes and the conflicts that arose within the university department concerning the establishment of a start-up (Tuunainen 2004; 2005). Much like the challenges confronted by universities in the USA, Finnish public research organizations were having to learn to manage their academic outputs and evaluate their market value. At the same time, it was not clear which metrics of scientific output had become the standards according to which researchers were being evaluated.

Despite many challenges associated with the commercialization of publicly funded research, there were also successes, which helped to highlight the possibilities. One of these was the KTL study of the genetics of hypolactasia, which reduces the digestive system's ability to process milk products (Tupasela 2006). Given the high level of dairy consumption in Finland, the study had a solid domestic market for testing and resulted in the development

of a genetic test that could be administered to diagnose the condition. Shortly after patenting the diagnostic test, KTL was able to license it to a large international company, Promethius, which began offering it in its catalogue. In general, however, early experiences related to patenting and licensing provided sobering reminders of the hollowness of some of the hype surrounding biotechnology.

Nonetheless, the belief in the potential of Finnish biomedical R&D for innovation and successful business did not die down among scientists and innovation policymakers in the years to come. On the contrary, innovation policy took a firm grip on cutting-edge biomedicine in Finland, and the drive for commercialization became more intense than before. Hopes were now invested in scientific and commercial collaboration with partners from abroad, and biobanks and healthcare data were given a key position in the new visions. Finnish policymakers wanted Finland to specialize in the refinement of data, not just consist of a source from which high-quality tissue samples and population data could be sourced. This interest in developing analytical capacity can be seen, for example, in Finland's choice to outsource most of its sequencing to large international organizations found at the Sanger Institute, Broad Institute, and Beijing Genomics Institute. Investment in expensive machines was seen as secondary to developing analytical expertise and know-how, as data analysis and refinement were seen to lead to a more profitable business approach than the bulk work of mechanical sequencing. Subsequent developments in the past few years, especially those surrounding the FinnGen consortium, have increasingly reinvigorated the commercial expectations connected with analytical capacity (see below).

Innovation policy takes over

The economic importance and commercial potential of sample collections and related data—which were starting to be discussed as biobanks—especially in the service of biomedical research

based on mass-scale genome sequencing, were widely acknowledged in Finland in the early 2000s (see Chapters 2 and 3). For almost a decade, however, 'commercialization'—as business-oriented activities were called—was not the main path to follow or the frame in which to think of biobanks in the service of biomedicine and medical genomics. From about 2005 to the early 2010s, the advocates of biobanking and biobank research—mainly scientists and managers from academic and public research institutions—focused on the development of national and cross-border infrastructures for data-intensive biomedical science and the pursuit of legal groundwork for biobanks in Finland (see Chapter 3). At that time, most of the people involved thought that building a stable, standardized, and smoothly operating biobank infrastructure would best allow medical genomics and other biomedicine to fulfil their grand promises of benefiting healthcare, public health, and also business prospects. Finnish researchers and biobankers participated actively in European networking efforts like the Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) to develop standards for biobanking that would facilitate sample queries and naming practices (see Chapter 2). One idea was that there would be a single search engine that researchers could use to locate samples and data across all participating countries (Holub et al. 2016).

In 2014, two governmental strategy papers based on broad consultation with relevant stakeholders—governmental and regional administrators, healthcare and social service managers, the rank and file in healthcare, social services, and ICT professionals, representatives of interest groups, industry, and business—were released, indicating a change in direction. The first one was *SoTe-tieto hyötykäytöön* (Making use of health and social services data) (Ministry of Social Affairs and Health 2014), which was jointly produced by the Ministry of Social Affairs and Health (MSH) and the Association of Finnish Municipalities. This 17-page report outlined a strategy, extending until 2020, to make the utilization of data in public population and patient data reservoirs more intense, unified, and integrated. The purpose of 'bringing health and social

services data into utility' was to serve efforts to make public healthcare more effective, improve public health, and enhance health promotion. Thus, the strategy approached the use of health data repositories from a rather conventional public health perspective. It is notable that the report considered the biobanks elements of health registers under public authority (see Chapter 5).

The other report was a landmark in framing biobanking and biobank research in terms of commercial innovation and business potential. The *Health Sector Growth Strategy* was released by the Ministry of Economic Affairs and Employment (MEE), and it presented a landscape in which national data reservoirs of health-related and population data, biobanks included, would be intensively utilized in the service of academic and commercial R&D pursuing personalized medicine. In this scenario, scientific objectives and potential benefits for healthcare and public health were greatly overshadowed by rationales and goal-setting defined in terms of innovation policy, the national economy, and business. The leading theme of the report resembled what had been presented 10 years earlier, for example in KTL's strategy:

> Finland is considered to be in an especially good position as a leading country in the so-called personalized healthcare research. Research and know-how are at a high level and have available globally unique comprehensive databases about national health. This potential should be utilised. (Ministry of Economic Affairs and Employment 2014, 3)

These two governmental strategy papers and the Genome Strategy which the Ministry of Social Affairs and Health released in 2015 (see Chapter 3) epitomized a change of heart among the advocates of high-tech biomedicine in Finland. Expectations about biobanking, medical genomics, and personalized medicine were now articulated more explicitly and concretely within innovation policy and commercial framing, and the promises of benefits to public health and healthcare figured as complementary justifications for the strategies. In the frame of innovation policy, plans to develop biobanks and high-tech biomedicine were aimed

at making the 'innovation ecosystem'[10] of personalized medicine in Finland more efficient and vibrant, especially in the commercial sense. Simultaneously, the prime context for defining biobanking and medical genomics—their potential, worth, and purposes, and how such themes are to be discussed—changed from the discourses and rationales of biomedicine and healthcare to the those of innovation policy (Tarkkala et al. 2019). This change was overt in the Genome Strategy.

With this, the top governmental officials, politicians, and programme directors and consultants of innovation agencies and think tanks took over the mandate to voice the objectives and value of the creation of a medical future, even though the essential knowledge and expertise are derived from practitioners of biomedical science and medical business. An excerpt from a speech by Olli Rehn, the Minister of Economic Affairs and Employment, at the Brain Diseases symposium in Helsinki in 2016, exemplifies the reasoning that became predominant:

> Finland has invested in health-related science, research and education, as well as in research infrastructures and an extensive public healthcare system for decades. Now these investments are starting to bear fruit not only in healthcare but also as a source for innovation, business opportunities, jobs and economic growth. … Boosting health sector growth is one of the key priorities in our overall growth policy. We are building on our strengths; thus, digital health and personalized medicine are at the core of the growth strategy. (Rehn 2016)

The powerbroker

The Finnish Innovation Fund Sitra has been one of the most energetic and influential matchmakers between innovation policy, biobanks, and genomics. Sitra is semi-public: it is financed by the parliament of Finland, but it has a mandate to act independently as an innovation fund. It was founded in 1967 to support and encourage the technological, industrial, and business innovations

of Finnish companies in order to facilitate the country's economic growth and competitiveness. In its early days, Sitra invested in private companies and their projects, but in the past three decades it has transformed into a think tank focused on designing and advocating projections of future technologies and society, and on the educational activities and pilot projects connected with them. Since the 1990s, Sitra has concentrated on outlining grand visions and anticipating megatrends in Finnish society and globally, on presenting pathways to these futures, and on conducting experimental projects for the betterment of Finnish society and the economy through technological progress and innovation.

Sitra's innovation concept is emphatically commercial, and the overall ethos of the think tank's activities is affirmative with regard to the market economy and the interests of private business. Throughout the 2000s, Sitra's visionary and project activities have become more closely linked to ministries and the government, and it and its experts have gained greater influence over policymaking in many fields, despite being criticized for their detachment from the realities of society and politics. Indeed, they have been identified with

> the class of think tank people who need hopeful, inspirational and energizing visions which enable cheerful gatherings and thinking of oneself as a member of an innovative vanguard iterating a model society that will provide good for everybody (Julkunen 2007, 79).

People at Sitra joined the bandwagon of the first Finnish genetics hype in the late 1990s and early 2000s. They eagerly advocated the 'revolutionary' blessings that mapping the complete human genome would bring to medicine and public health and promoted the idea of collecting the genetic information of the whole Finnish population in a national 'gene library' to be used for the benefit of Finland and its science, citizens, and economy (e.g. Kuusi 2004). This message was congruent with the promotional discourse of top medical geneticists, including Leena Peltonen, claiming that new medical genomics has enormous potential to bring health

and also wealth to Finland (see Chapter 2). This created an alliance between biomedical scientists and an influential lobbyist at the heart of Finnish innovation policy.

In the 2010s, as biobanks were preparing to start operations (see Chapter 3), Sitra continued to advocate a vision of future medicine based on the knowledge, diagnostics, and cures provided by advanced genomics, framed by the catchphrase 'personalized medicine' (on the latter, see Chapters 1 and 3). As part of its campaign, Sitra emphasized the translation of genomic technology and knowledge into clinical use, and the sharing of people's information about their personal genetic characteristics and risks. To this end, Sitra collaborated with GeneRisk, a Tekes-funded project in which researchers at the Institute of Molecular Medicine Finland (FIMM) studied genetic cardiovascular disease (CVD) risks in a research population of over 4,000 individuals aged 45–65 from the Kotka area of southeast Finland and experimented with delivering CVD risk scores, both genetic and conventional, to physicians and patients via the mobile application Kardiokompassi. Sitra had been involved with developing the application, which was embedded in its own Taltioni platform, among other digital healthcare applications.

Taltioni, launched in 2012, was a platform enabling healthcare and social service providers—public, private, and voluntary organizations—to offer digital self-care applications or other services to customers and citizens. It was also planned to function as a data deposit in which the individuals themselves could store personal data, medical data included, and share their data with service providers, including commercial partners.[11] Sitra boasted that Taltioni was a 'trailblazing' initiative, combining the digitalization and personalization of healthcare with an emphasis on health promotion (Sitra 2012), but it eventually faded into obscurity, as it was unable to attract sufficient public interest in sharing personal data on the platform (Riso et al. 2017).

Sitra was also instrumental in the making of the national Genome Strategy we discussed in Chapter 3. It put a lot of effort to bringing together different stakeholders—companies, researchers, hospital

regions, university hospitals, biobanks, the Finnish Institute for Health and Welfare (THL) and other state authorities, innovation policy officials, and representatives of patient organizations—and co-hosted, with the MSH, the forums in which the stakeholders and experts discussed the strategy. Although openness was emphasized by Sitra and the ministry, in practice it was expert-led policymaking, in which public and patient representation was quite stringently controlled and managed, that appeared to provide support for the venture. Such a style of policymaking is quite typical in Finland and predominates in the domain of biomedicine.

For over a decade, Sitra had embraced an idea, or rather a vision, that future medicine would inevitably be based on genomics and that clinical applications would be in routine use in the near future, bringing unprecedented benefits to the public in the form of targeted prevention of diseases and improved treatment; this, in turn, would result in diminished healthcare expenditure. This imaginary, or even ethos, is pronounced throughout the Genome Strategy. It was perhaps most clearly voiced by a leading governmental official, Liisa-Maria Voipio-Pulkki from the MSH, who claimed at many events that 'genome information will be widely utilized in everyday healthcare in Finland by 2020', the objective set by the strategy. This visionary optimism contrasted with other perspectives from the field, as general practitioners working at the coalface of healthcare have been critical about such high-tech visions of the future (Snell and Helén 2020).

More importantly, Sitra contributed substantially to synchronization of the Genome Strategy with the Health Sector Growth Strategy and its innovation policy rationale. Evaluating its importance when it was published in 2015, Antti Kivelä, a director at Sitra, observed:

> If the proposals of the Genome Strategy are realized, society may benefit from the slowing down of the rise of healthcare expenditure and better targeting of the resources. Moreover, actualization of the proposals would ensure that Finland will be [an] attractive milieu for research and business in genomics. (Sitra 2015b)

This kind of reasoning underlined the view that biobanks, biobank research, and the translation of medical genomics into clinical use are primarily about innovation and thus about economy and business. With the Genome Strategy, this framing acquired governmental support.

In parallel with shaping the Genome Strategy, Sitra started advocating more extensive and efficient secondary use of the health data stored in national registers and regional electronic health record (EHR) systems and other healthcare and social service databases. The term 'secondary use' refers to all kinds of health and social service data applications besides that of serving clinical or preventive healthcare. Sitra and many medical scientists, ICT experts, top governmental officials, politicians, and lobbyists expressed their concern that public data repositories are not sufficiently utilized and their content does not sufficiently match the needs of research, policy, and the economy in light of the possibilities provided by advanced ICT. Voicing these considerations, Hannu Hämäläinen, an MSH official and later a senior advisor at Sitra, stirred up action on Sitra's website, writing, 'Finnish welfare data are being hidden in a treasure chest. It is time to start refining data safely and in more agile ways than before' (Hämäläinen 2016).

Sitra and other advocates repeated their concerns and demands in numerous blog writings and PowerPoint presentations for political and professional publics. They wanted to change policy, legislation, and regulation with two objectives in mind: first, they proposed wider secondary use of data for administrative, management, educational, and innovation and commercial R&D purposes; second, they demanded easier access to public data repositories for a greater variety of potential users, including private companies and their research divisions. For the advocates, the main problem was that the data were scattered across many administrative 'silos' where they were stuck behind legal and regulatory firewalls. It was proposed that new policy and legislation would be 'enabling', removing or bypassing these obstacles and allowing more flexible—that is, less regulated—'interoperability',

which would enhance more intense data sourcing from public repositories.

This promotion of more extensive secondary use of the population and patient data in public data reservoirs was highly significant for biobanks and biobank research in Finland, as discussed in Chapter 3. Such enhancement, making biomedical data infrastructure more seamless, was vitally important because of developments in international cutting-edge biomedical research—medical genomics, in particular—in which the combination of sample data and the personal health data from multiple sources of tens of thousands of people had become the main way to go (Cambrosio et al. 2014; Leonelli 2014). The development of more flexible access to national health register data and smoother interoperability between the registers was considered necessary if Finnish medical genomics was to keep up with data-intensive international biomedical science. This line of reasoning and action on a national scale culminated in the FinnGen consortium (see Chapter 3 and below), for which Sitra's promotion of enabling regulation and legislation for the secondary use of public health-related data had prepared the soil.

Sitra's persistent advocacy of more extensive secondary use of health-related data in public registers was not just talk and lobbying. The think tank also fused its visions in practice by initiating two pilot projects in which models, devices, and infrastructure for grand-scale data sourcing and usage were developed and experimented with. In the period 2013–2016, Sitra joined forces with the MSH and some regional authorities in charge of the provision of public healthcare and social services to construct a model for compiling data from public service providers' databases—both patient or client and administrative data—and from national registers. The idea was to organize data and data analytics tools into 'packages' that would help in compiling service demand prognoses, welfare and performance indicators, and client analysis, for the use of top managers and regional policymakers. The purpose of the project was to build a demonstrative example of data-driven management of health and social services, and 'data management

systems that would provide standard data and indicators about performance, economy, quality and efficacy' (Sitra et al. 2016).

The other project had more relevance to medical genomics and biobanking. In 2015, with the launch of the Isaacus project, Sitra started to promote and experiment with extended and more intense secondary use of healthcare and social service data in public registers on the national scale. Tied closely to the lobbying for legislation and regulation reform of the secondary use of health data (see Chapter 3), Isaacus brought together major national data-management authorities like Statistics Finland, THL, and regional healthcare and social service authorities. The joint task was to develop a 'one-stop shop' that would manage access to all data in the Finnish public healthcare and social services data repositories, and the delivery of data to a variety of users. Sitra and its collaborators believed that such a 'service operator for welfare data' would be indispensable in expanding the secondary use of the register data (Figure 13).

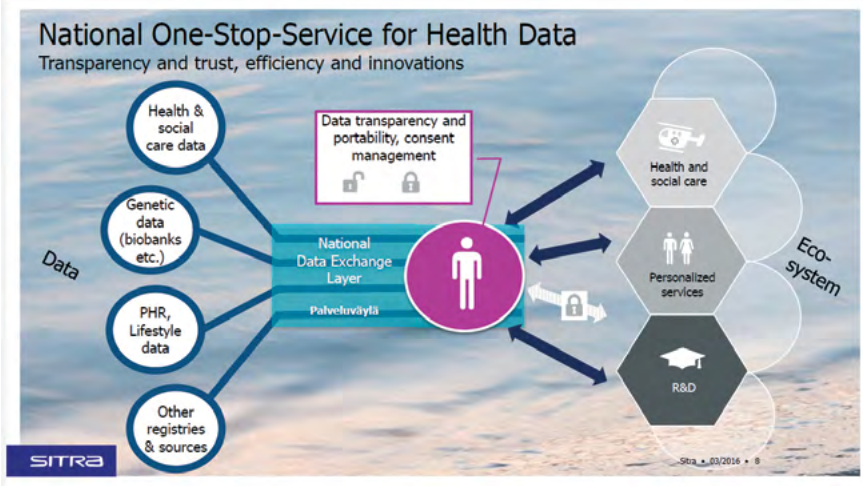

Figure 13: National one-stop-service for health data. Sitra's top executive Antti Kivelä used this graph (Sitra 2016) to illustrate the functioning of the centralized access point to the Finnish health data planned in the Isaacus project. All rights reserved.

Isaacus was carried out in a number of so-called pre-production projects in which infrastructure—such as technical solutions, data lake architecture, data models and standards, data protection requirements and solutions—were developed and tested. In a practical sense, Sitra's project was not unique, since a plethora of national and regional projects mapping and piloting new generation data management in healthcare and social services was being conducted simultaneously in Finland. Rather, its significance lies in its successful promotion of two ideas to a combined audience of the medical profession, government officials, and politicians.

The first was a prospect: it was claimed that wider secondary use of healthcare and social service data for administrative, management, educational, and innovation and commercial R&D purposes, and easier access to public data repositories for a greater variety of potential users, including private companies and their research facilities, would bring unprecedented benefits in the form of improved public health, savings in public expenditure, and new business opportunities connected to high-tech medicine.

The second was a concern: because of complicated and slow access to data dispersed in administrative silos behind legal and regulatory 'obstacles', they were neither utilized enough, nor in an appropriate manner, with regard to the needs of research, policy, and the economy, and given the possibilities provided by advanced ICT. According to Sitra and other advocates, introducing a new enabling policy that removed or bypassed legal and regulatory 'hurdles' would dissolve this bottleneck and allow smooth interoperability between public data repositories, which would boost more intense data sourcing for a variety of secondary uses:

> National data reservoirs and possibilities for their utilization can form an innovation platform that attracts both domestic and international researchers, developers and entrepreneurs. At its best, the Isaacus will enable utilization of different data repositories in an agile and reliable way. (Hämäläinen 2016) … A unique ecosystem will be opened for research and business collaboration,

> without jeopardizing data protection (Hämäläinen 2018, own translations).

Eager advocacy by Sitra and its collaborators was influential in many ways: an enabling law for secondary use of data was passed in 2019, following the establishment of Findata at THL as the central public gatekeeper and access service to public repositories. To a great extent, Findata's idea followed Isaacus' one-stop shop model (see Chapter 3). After a couple of years of operation, however, many stakeholders—especially Finnish researchers—started to complain about flaws in the law and functioning of Findata. The handling of access applications, the demand for an accredited data-protective ICT environment in Finland for data management, and the increased cost of data were regarded as making the situation worse than it had been before the new law. For example, in February 2021, during the Covid-19 pandemic, the leading experts in intensive care sent a letter to the leaders of the parliamentary groups in which they listed the negative impacts of the law for secondary use and Findata on their research activities. They wrote that 'as Findata operates according to the principle "one size fits all", it poses a threat to research conducted with a small number of patients and/or without substantial research funding'. They also stated that

> … because of the law for the secondary use and interpretations by Findata, we have to inform intensive care networks in other countries … that Finland will not be capable of participating in research project[s] based on register data and conducted in a rapid phase and without specific funding. The reason for this [is permission bureaucracy that] is too inflexible, too slow, and too expensive. (Own translation)

The Isaacus project, the making of the secondary-use legislation, and the founding of Findata were carried out concurrently with activities leading to the merger of Finnish biobanks under a nationwide service organization, which resulted in the foundation of the biobank cooperative FINBB in 2017. As discussed in

Chapter 3, those parallel efforts resulted in two access points to Finnish health-related data—one for tissue sample collections at FINBB and another for register data at THL's Findata—instead of a one-stop shop. Nevertheless, this pursuit of national centralization of access and delivery of health-related data in public repositories significantly paved the path along which medical genomics and personalized medicine were directed in Finland. The importance of those two lies in the fact that they constituted concrete plans and organizational arrangements to intensify and extend the actual use of health data. As such, they were crucial building blocks for consolidating the framing of innovation policy and commercialization in which medical genomics—especially biobanks and biobank research—were situated in the 2010s.

Marketing the promise

As noted, Sitra had been a key facilitator of the making of the national Genome Strategy (see Chapters 3 and 4) a couple of years before the launch of the Isaacus project. In this context, Sitra had compiled, in collaboration with key experts in medical genetics (mostly at the FIMM), a PowerPoint show of 48 slides as a sort of introductory tour to Finland as the wonderland of medical genomics and biobanking. The purpose of the slides was to provide a marketing package with a unified message and outlook that would help the promotion of Finnish research facilities and expertise in international biomedical science and business domains (Tupasela 2021). A wide range of spokespersons for Finnish medical genomics, personalized medicine, and biomedical innovation adopted slides from the package; consequently, the audiences of numerous workshops, seminars, and other events on biobanks, personalized medicine, or medical genomics at home and abroad became familiar with the hive-cell-shaped forms and background image of clouds and waves in shades of darkish blue in this Sitrayan scenery (Figure 14). Wide use and circulation made the image iconic, and it became a national landscape presenting 'Finland' as unique and the best 'test bed' for biomedical research.

Figure 14: The most advanced testbed in the world. A national landscape in a promotional slideshow by the Finnish Innovation Fund Sitra (2015a) presenting the five advantages of Finland as an environment for biomedical R&D. All rights reserved.

In terms of its content and style, Sitra's landscape image of Finland as the 'most advanced' milieu for conducting cutting-edge biomedical research and development was associated primarily with innovation policy and therefore with business potential and commercial expectations. The slides crystallized the message already being disseminated a decade-and-a-half earlier (see above): data in biobank repositories and registers of public healthcare and social services are an exceptional resource for biomedical R&D, and such data are therefore a national asset that provides Finland with a competitive advantage in biomedical research and business, as discussed in Chapter 4. The competitive advantage included speed of access in technical, ethical, and legal procedures, price, and quality. The idea was also consolidated in the domestic publics, and talks in seminars, workshops, and lectures on biobanks or personalized medicine, as well as texts about these topics, repeatedly referred to Finnish data repositories as a 'treasure chest' or 'pile of ore', or compared them to 'green gold', thus associating them with the importance of forests to the Finnish

wood processing industry. References to the mythical Sampo (see Chapter 2) also appeared. Similar metaphors have likewise been used consistently in other Nordic countries—in Denmark, for example, where national biobank collections and healthcare data are referred to as the new oil (Tupasela et al. 2020; Hoeyer 2019; 2023). With the consolidation of this view, the call to arms to actualize the innovation potential of Finnish health-related data by streamlining the access and interoperability of biobanks and public data repositories was widely heard and accepted by the medical profession and politicians alike.

The form in which the above message was presented in Sitra's slides framed Finnish biobanks, the health and social service infrastructure, and biobank research itself primarily as the foundations and servants of business activities with great commercial potential (Tarkkala 2019; Tarkkala et al. 2019). The outlook and style were deliberately designed for marketing purposes, which resulted in 'Finland'—and its characteristics as 'unique' and 'most advanced'—being presented as a brand. Thus, Sitra's landscape was a branding mechanism or, more precisely, had the goal of nation branding, which, since the 1990s, has become a common promotional activity among nation states and their regions when competing for foreign investments to vitalize domestic economies and business (Tupasela 2017; 2021). Sitra's landscape is emblematic of the development whereby tissue samples in biobanks and health-related data—or 'real-life data', as Finnish biobank experts called the latter (see Helén and Lehtimäki 2020)—were predominantly seen in terms of innovation, business, and commercial potential (Tarkkala et al. 2019; Tupasela et al. 2020). In addition to this framing, the Finnish population, data reservoirs, and research and healthcare infrastructure were regarded as forming a resource or an asset that was marketed internationally as a national brand (Tupasela 2021; 2017).

Sitra was not the only institution to brand Finland; top biomedical researchers, research organizations, and the governmental innovation and export agency Tekes (now Business Finland) also promoted Finland under the rubric of the 'most advanced testbed'

(Tupasela 2022b). This was meant to highlight Finnish genetic and health data resources, which were characterized as unique by virtue of the homogeneity of the population and the high quality of healthcare data reservoirs (Figure 15). While this branding of data and tissue samples for marketing purposes derived from the idea of the genetic uniqueness of the Finnish population, it eventually extended to the 'exceptional' reservoirs of population and personal health-related data in public registers, and the Finnish research milieu and public healthcare infrastructure (see Chapter 4). The goal of the marketing was to attract foreign investment and research collaborators, especially from Big Pharma corporations. FinnGen is an example of such research collaboration with industry (see below), while Findata exemplifies the public institutions and infrastructure that the government and other public authorities establish to facilitate the branding, marketing, and commercial collaboration connected with biomedical research.

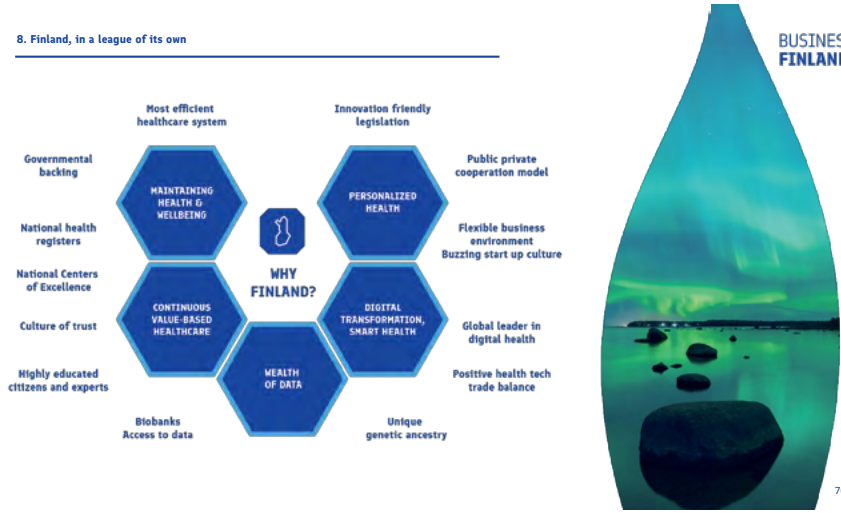

Figure 15: Finland, in a league of it's own. Many advocates of Genome Finland used visual imaginary similar to Sitra's national landscape in branding Finland as unique and the best testbed for biomedical R&D. This image is an excerpt from a Business Finland report (2022). All rights reserved.

As an agent of innovation policy and a facilitator of the development described above, Sitra can be seen as the *political* intermediary organization that mediated and shuttled between the government and ministries, the public innovation agency (Tekes/Business Finland), and the inner circle of biomedical scientists, as well as summoning the stakeholders together. These mediating activities impregnated medical genetics and biobanking in Finland with an imaginary and rationale of commercialization and directed the running of research and biobanks as if they were business. The latter required biobanks and organizations conducting biobank research also to think of their activities as business and conduct them according to a business model.

Sitra has continued the promotion of more extensive and intense secondary use into the 2020s. Its domestic effort concentrates on advocating more intensive utilization of health data reservoirs and advanced ICT, especially data mining and AI, in Finnish healthcare, both in management and everyday clinical practices (Sitra 2023). In addition, Sitra has become active in a European context; the Finnish think tank has actively participated in developmental work for the European Health Data Space (European Commission 2022) and a number of ancillary projects seeking to establish guidelines and standards for the sharing of health data across national borders in Europe. For example, Sitra directed the TEHDAS Joint Action that aims to create European principles for the secondary use of health data, with the goal of enabling individuals to control their own personal health data, including routinely collected and stored data (Tupasela 2022b). The adoption of the dual role as both major national policy mediator and developer of transnational sharing standards underlines Sitra's mission to contribute to the development of a global data assemblage. The changes surrounding the use of health data and tissue samples in Finland (infrastructure, law, and the ethos of innovation and commercialization) are all part of a broader endeavour to integrate Finland and the resources in Finnish public data repositories into global data infrastructures and economies.

Biobank business model

The shift in reasoning whereby biobanks and biobank research in Finland were predominantly seen through the looking glass of innovation policy and its economic and commercial presumptions took place at much the same time as preparatory work for the beginning of biobank operations around Finland. Consequently, the above imaginary—the national landscape of Finnish medical genomics—was widely shared and firmly consolidated among those involved in establishing biobanks and biobank research (Tupasela et al. 2015; Lehtimäki et al. 2019). In this context, plans, strategies, and practices for biobanking evolved in which the function of the biobanks was to facilitate biomedical science and R&D aimed at better diagnostics and treatments, interlaced with a business orientation.

Auria, a forerunner clinical biobank closely associated with the Turku University Hospital, suggested the outlines of such an amalgam business model early on (Lehtimäki et al. 2019; Helén and Lehtimäki 2020). Indeed, it can be seen as a paradigmatic example, since the main elements of the model have been widely adopted by Finnish biobanks and biobank research. Many biobanks in other countries also have a similar operative rationale (e.g. Timmons and Vezyridis 2017; Turner et al. 2013; Hauskeller and Beltrame 2016). Auria started operations vigorously, directly after the Biobank law came into force in 2013 (see Chapter 3). At that time, it defined itself as a public and academic institution with the primary purpose of advancing biomedical research, especially in Finland. Yet Auria's key actors reasoned that business activities and profitable collaboration with private companies were important, if not a necessity, in sustaining and developing biobanking operations. 'At least a half of the annual expenses of the biobank should be covered by our own funding received from business collaboration with private enterprises', said CEO Heli Salminen bluntly in an interview in 2016. Yet for Auria, biobanking business was explicitly *instrumental*. Commercial success, profits, and private investments were sought only to cover the maintenance of the

data repository and data management infrastructure considered crucial to biomedical research. Despite this emphasis, many of Auria's activities focused on dealing with commercial collaboration and marketing biobank services (Lehtimäki et al. 2019; Helén and Lehtimäki 2020).

According to Auria's key personnel and document material, the commercialization of both biobank data and expertise in data sourcing and management was to be carried out in a manner that enabled the biobank 'to survive in a business world' that has grown up around biomedical research (Auria 2016, 9, own translation), and this became a key task of biobanking (Lehtimäki et al. 2019; Helén and Lehtimäki 2020). From Auria's perspective, access to the market and commercialization primarily required the pursuit of profitable and longstanding collaboration with private enterprises, especially Big Pharma corporations. This was explained in the following way:

> Ongoing collaborative projects are clear indicators of [biobanks'] potential, and they provide income to a biobank. However, current activities are not enough to attract really big foreign investments in Finland. Auria is moving from collaboration projects to strategic partnerships … and negotiations about establishing a joint research centre at Auria are going on with the collaborative partners. To be a serious partner, we need a wider biobank frontier that is capable of providing an infrastructure for significant international investments. (Auria 2016, 29, own translation)

Auria—like biobanking experts widely in Finland—believed that initiation of collaboration with a significant pharmaceutical corporation or another medical company requires the biobank to have something unique in medical R&D that attracts potential partners. Although Auria's self-assessment was that it was unique compared with its competitors around the globe for several reasons (see Lehtimäki et al. 2019), one attraction was superior to the others: 'real-life data'. Expectations and experiences of collaboration with pharmaceutical companies had made Auria's people realize that clinical and other patient data were 'utterly interesting'

(IT expert, interview 2017), especially to their commercial clients, because such data are of great utility in targeting research in drug development, seeking a new diagnostic method or in feasibility studies:

> They are particularly interested in our phenotype data … it is precisely the clinical data of our hospital patients that allows deep phenotyping, so that we can find exactly the right patient for the right study. (Biobank CEO, interview 2016)

> If [the clients] need more data associated with the sample, then there are not many places where they can get similar data as we have. Elsewhere in the world, there are not clinical data collected from such a long period of time, and then we have PIN [national personal identity number] through which we can connect all the data [from different sources] with each other. And the law allows the biobank to acquire data from public registers, like the cause of death from Statistics Finland or information on drug reimbursements from the Social Insurance Institution of Finland. (Biobank project manager, interview 2017)

In Auria's business model, the main element facilitating business collaboration was not uniqueness of 'real-life data' per se, but its data management service. The biobank believed that pharmaceutical and other medical companies would be interested in its services for two reasons: first, it provided access to wide repositories of clinical and other patient data, and, second, it was capable of sourcing datasets from its own sample collections, patient record databases in hospitals and hospital districts (today well-being services counties), and national healthcare and population registers, and then customizing the data according to the customers' wishes (Lehtimäki et al. 2019; Helén and Lehtimäki 2020). Thus, the attraction of unique real-life data—considered the main catalyst for business collaboration—was embedded in the biobank's high-quality and flexible data management service, which would make Auria competitive and capable of surviving in the biomedical research market:

> We have invested in our service. We serve our customers so that they can get what they want, and we take care of all that needs to be done on behalf of our customer. And indeed, we have been thanked for being flexible, that it is easy to discuss with us, and that the projects proceed smoothly, and both partners are in dialogue all the time. (Biobank project manager 2017)

The Finnish biobank cooperative FINBB and the flagship consortium FinnGen (see Chapter 3 and below) follow a similar business rationale to Auria. They consider their activities to be primarily academic, with scientific objectives that serve the public and the common good in the domain of healthcare. They see their engagement with biomedical business as *instrumental* in the sense that the commercial dimension of biobanking or biobank research facilitates progress in biomedicine and, most importantly, provides the financial resources to sustain research. Yet both organizations tend to highlight the business dimension of their work and emphatically display their eagerness for partnership with private enterprises, preferably global pharmaceutical corporations.

Like Auria, FINBB and FinnGen also believe that their main value lies in their 'attraction' as a potential partner to pharmaceutical or other biomedical companies. The core of 'attraction' consists of their high-quality data, while the associated top-quality data management services provide their partners with access to the unique data of a unique population and expertise in customized data sourcing and analytics (Tarkkala 2019; see also Chapter 4). The biobank co-op FINBB described itself as 'a centralised gateway' to Finnish biobanks and continued,

> FINBB provides access to a network of data, unequalled in the Nordic region. Our service is designed to be utilised by both academic researchers and commercial developers of healthcare innovation and treatments. Researchers can discover populations of precisely the right subjects with the relevant data. Commercial organisations can find research partners with the correct specialty or level of expertise. One of our vital roles is to facilitate

> successful partnerships without either side getting bogged down in some of the heavy administration that goes with conducting a biobank study. (FINBB 2019)

FINBB is a cooperative of the six regional Finnish biobanks that can be characterized as clinical biobanks and two population biobanks; it was founded in 2017 and began operations in 2018. The co-op was the result of plans for a biobank merger which were initiated in 2016, and to a great extent it realized the objective—set by many key people in the Finnish biobanking scene and the MSH—to establish a nationwide biobank data repository and data sourcing service, a 'one-stop shop' (see Chapter 3). The merger was framed and justified by the business reasoning described above. Thus, biobanking and the utilization of biobank data through FINBB are nowadays predominantly discussed in terms of 'value creation', with a notable commercial connotation. This is exemplified by a graph of a biobanking 'profit cycle' that FINBB's CEO Marco Hautalahti presented in FinnGen's business ecosystem event in December 2019 (Figure 16).

The main driver of the plans and actions which produced FINBB was uncertainty among people involved in Finnish biobanking as to whether the data repositories in the Finnish

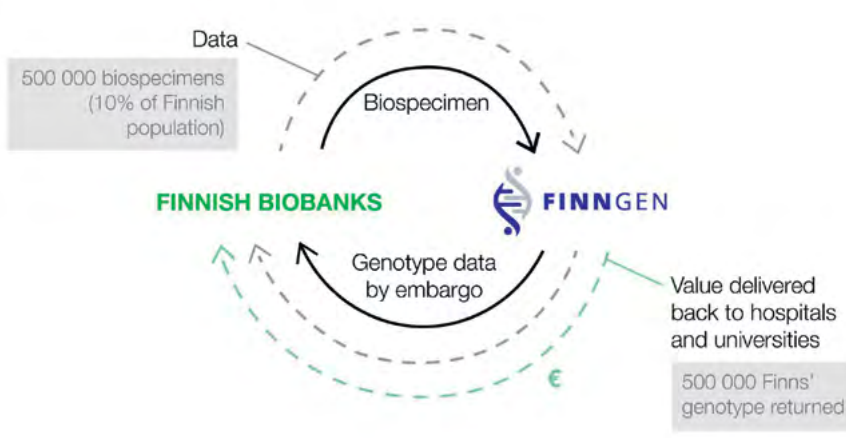

Figure 16: The biobanking profit cycle (Hautalahti 2019). All rights reserved.

biobanks were extensive enough to attract scientific and commercial collaborators from abroad, pharmaceutical companies in particular. Similar problem forecasts, concern over the risk of biobank data becoming useless and devalued, and issues connected with the financial sustainability of biobanks have been widely discussed internationally, engendering 'biobankonomics' as a special branch of research (e.g. Vaught et al. 2011; Tupasela and Stephens 2013; Simeon-Dubach and Henderson 2014; Chalmers et al. 2016; Kongsholm et al. 2018). Today, the mainstream view among the scholars in this field is that private funding cannot provide a stable basis for public biobanks, and that considerable public funding is required for sustainable data collection, storage, and service provision. In Finland, the latter view has not been loudly voiced; rather, Finnish biobanks have responded to sustainability concerns by acting to build a national service access point that makes larger repositories of biobank data more readily available for potential users. According to the above business model, the rationale of this effort was to ensure and increase the *attraction potential* of Finnish biobank data and data sourcing expertise (Helén and Lehtimäki 2023).

All in all, FINBB represents an important biobanking element of efforts to nationalize health-related data gathered by public authorities in their data repositories, a goal also pursued by Isaacus and Sitra's projects and advocacy, the enacting of enabling legislation for more extensive secondary use, and the founding of Findata. The move towards an effective national centralization of health data—although the end result was two access points instead of one (see Chapter 3)—fortified an innovation policy framing that emphasized the business and commercialization aspects of biobanks and, more widely, of all sourcing and utilization of health-related data. Within this framing, data in the repositories managed by the public authorities are a national asset because they attract collaborators from abroad to engage in high-tech biomedical R&D and business with Finnish research institutions and companies. The attractiveness, in turn, is derived from the data's value-creation potential in the biomedical innovation

business. This view of biobank data as an asset was also crucial in FINBB planning, as expressed in a report on biobanks: 'The biobanks should be seen as the guardian of national resources [that] should be harnessed as productively as possible' (Auria 2016, 5, own translation). The task of FINBB is to take care of the assets entrusted to it, and it sees itself doing so by serving as a go-between or matchmaker between the biobanks (as data management service providers) and their collaborators. This is assumed to increase the attraction of Finnish biobank data and thus commercial potential, which, in turn, would generate the financial resources to sustain a crucial biomedical infrastructure and open business opportunities to domestic innovation companies in the global genomics market.

The acceleration of domestic business was a key promise and objective of policy extending the secondary use of health-related data and facilitating health sector growth in general. Accordingly, the advocates and policymakers believe that the establishment of national 'one-stop shops' offering access to biobank and health register data substantially boosts the operations of innovative high-tech Finnish companies in the field. Toward the end of the 2010s, there was much enthusiasm about the growth potential of the field and the small Finnish companies in the business. Innovation policy experts highlighted the expansion of the global medical genomics market, the opportunities this expansion opens to small Finnish companies to capitalize on their expertise, and the support provided by Finnish biobanks and other data infrastructure to the commercial efforts of Finnish entrepreneurs. Quite often the same five companies—Abomics, BC Platforms, Medisapiens, Blueprint Genetics, and Genescoper—were presented as encouraging role models in terms of taking advantage of the business opportunities at hand. The launch in 2020 of a partnership to 'accelerate healthcare start-ups' by the Helsinki start-up event Slush and pharmaceutical company AstraZeneca exemplifies the hype.

In 2017, Healthtech Finland, an association of Finnish companies in the healthcare business, founded a special section for

the genomics industry: 'a community of Finnish companies involved in promoting production, utilization and enabling the use of genomic data and knowledge'.[12] In six years since then, the genomics industry has endured but not grown rapidly, and it would be misleading to speak about a tight innovation ecosystem or cluster. Rather, each company has found its own market niches and collaboration networks at home but mostly abroad, and some of them have been more successful than others. If successful or showing sufficient potential, the usual way for a small Finnish company to proceed is illustrated by the trajectory of Blueprint Genetics, a company specializing in clinical gene and genome testing for a variety of hereditary diseases: in 2020 it was sold to the US-based Quest Diagnostics, and the Finnish entrepreneurs and owners made millions of euros in profit. This is a typical path for successful start-ups and small innovative companies in biomedical high-tech around the globe. Notably such companies and their association are active in their efforts to make policy and the business environment at home more favourable to their operations. In this context, representatives of companies and the association complain about the difficulties of 'getting a seat at the tables where decisions are made', and some companies consider it complicated and difficult to start collaboration with public sector partners—healthcare or research institutions—because the public partner is often not very responsive to the expertise or technology the company has to offer to such a collaboration (Parkkinen et al. 2023).

As already noted, boosting domestic business in biomedicine has been considered of primary importance in national innovation policy, and there have been many initiatives and efforts to cultivate the growth of small, innovative companies in the field. Nonetheless, they have not been the main recipients of support in the actualization of the business prospects in medical genomics through initiatives and mechanisms such as the innovation policy rationale, tightening up the biomedical innovation ecosystem, promoting and branding the uniqueness of Finland as the biomedical R&D test bed, and efforts to centralize access to biobank and healthcare data nationally. Instead, activities and financial

support for the commercialization of biobanking, biobank research, and related data reservoirs have to a great extent been channelled towards a big flagship project, whose policy and trajectory affirm the goal of national centralization of the control of data sourcing and utilization. This flagship is FinnGen.

FinnGen—'an exciting public–private partnership'

As we briefly discussed in Chapter 3, when the second decade of the 21st century began, plans got underway for an unprecedentedly extensive biomedical genome mapping project at FIMM, with Professor Aarno Palotie as its director and frontman. The FinnGen consortium grew out of pilot projects, such as Sequencing Initiative Suomi (SISu), which genotyped 10,000 Finns, and GeneRISK, an experiment in the clinical use of CVD genome risk information. It was officially launched in 2017, a few months before the biobank co-op FINBB, with the objective 'to produce comprehensive genome variant data of 500,000 biobank participants, representing one of the largest studies of this type' (FinnGen 2019). Within a couple of years of setting that goal, it became the largest user of Finnish biobank data and their most important customer. Consequently, it also became the main route to the Finnish tissue sample and health data repositories for foreign researchers and companies.

FinnGen is essentially transnational. Its prime research activity is engagement with large-scale genome-wide association studies (GWAS) with massive datasets collected from all over the world, and its main scientific partner in global genomics is the Broad Institute of Massachusetts Institute of Technology (MIT) and Harvard University. Moreover, most of the project's funding comes from Big Pharma enterprises, while it uses Google's algorithm tools and cloud services for data analytics and data management.

From the beginning, FinnGen emphasized the importance of commercial collaboration and eagerly sought partnerships with pharmaceutical and other biomedical companies. This orientation is reflected in its applications for public financing from the national

innovation agency Business Finland and its predecessor Tekes, instead of conventional academic funding sources. The business aspect of the consortium is even more prominent in FinnGen's largely successful attempts to acquire financing from Big Pharma enterprises and high-tech biomedical companies through a form of club membership. In this arrangement, each company partner pays a 'fee' of 1.5 to 3 million euros, which allows it quite extensive access to Finnish biobank and health register data in collaboration with FinnGen; at the moment, there are 13 company members in the club, including corporations like Pfizer, Roche's Genentech, Merck, and AstraZeneca. In 2019, Aarno Palotie and colleagues wrote in *Duodecim*:

> International pharmaceutical industry is a core element of the FinnGen endeavour. The industry takes care of over 70 per cent of the financing, and the rest comes from Business Finland. Researchers with excellent scientific merits from the industrial partners are actively involved in planning and conducting the project as a community, across the company boundaries. The unique model of collaboration is possible because FinnGen is an endeavour focused purely on research. (Palotie et al. 2019, 990, own translation)

FinnGen presents their collaboration model as exceptional, and underlines that the project is primarily scientific and 'precompetitive' despite the major role of Big Pharma corporations. An all-encompassing range of good from FinnGen and its collaborators' cutting-edge biomedical science would then result in general benefits to medicine and benefits to

> the public, Finnish companies, biobanks, the healthcare system, and academic research'—some of the blessings FinnGen is touted to bring; on top of that, 'all breakthroughs that arise from the project will eventually benefit health care systems and patients globally (FinnGen 2023).

FinnGen is perhaps the clearest example of a duality that is quite typical of Finnish biobanking and biobank research.

The consortium presents a Janus-faced self-image as both an essentially academic-industrial partnership and a purely academic initiative with primarily scientific objectives serving the public and the common good. These two aspects are reconciled by the business reasoning discussed above. FinnGen considers that its engagement with biomedical business and its collaboration with major pharmaceutical companies are *instrumental* in the sense that the commercial dimension of research facilitates progress in biomedicine and, most importantly, provides financial resources to sustain research (Helén and Lehtimäki 2023). Since business activities with private companies are considered indispensable to the maintenance of FinnGen's extensive research agenda, instrumental activities dealing with commercial collaboration and marketing of services have become FinnGen's key or even dominant task. The consortium's spokespersons and documents express the commercial objectives and their priority clearly. On its website, FinnGen declares that one of its main objectives is to create a business ecosystem in Finland that will

> invite large international pharmaceutical companies and companies representing other industries to Finland. Especially international companies are hoped to increase their investments in Finland, financing of Finnish research and innovations and new companies generated by the ecosystem even after the end of the project. (FinnGen 2023)

Thus, FinnGen's pursuit of commerce is not restricted to its own interests, since it has raised expectations of boosting biomedical business on a national scale. The consortium claimed—or even promised—that an invitation to Big Pharma corporations as collaborators and financiers of FinnGen would

> strengthen innovation and business activities nationally, because it is expected to increase cooperation between Finnish companies, health care operators, researchers and/or companies and international researchers and/or companies. … The project will benefit companies in the form of new business opportunities e.g.

> in software design, IT solutions, genetic services, clinical testing, diagnostics and early-stage drug development. (FinnGen 2023)

Such a statement readily fits the promissory landscape of national innovation policy and health sector growth strategy; indeed, a Business Finland executive praised FinnGen's national importance:

> The FinnGen project is like a magnet that draws the interest of the global pharmaceutical industry to Finland and brings significant new players and investments to strengthen the ecosystem …. We expect remarkable growth in research and development investment over the next years. FinnGen has also worked extremely well in creating links between the international pharma and Finnish companies, which we hope will eventually generate more innovation, business and cooperation models. (Business Finland 2020)

FinnGen's business rationale is embedded in the national branding of Finnish biobanking (Tupasela 2021), and it tends to present itself at home and abroad as the flagship of the brand—quite successfully, so far. As a business, FinnGen focuses on providing attractive opportunities for collaborative R&D with great potential for commercial gain. For potential partners, the attractiveness of collaboration is based on the alleged uniqueness of Finland; it is collaboration with FinnGen—including financial investment—that provides foreign partners with the main entrance to the 'most advanced test bed in the world' and—most importantly—grants access to the unique data (in biobanks and population and healthcare registers) of a unique population, accompanied by top-quality data management services and expertise in biomedical R&D (see Chapter 4; Tarkkala et al. 2019; Tupasela et al. 2020). This model for the commercialization of biobank data, as well as its related performative and promissory rationale, are essentially the same as those of deCODE Genetics in Iceland (Rose 2001; Fortun 2008), and similar to collaboration models widely adopted in Denmark (Hoeyer 2019; Tupasela 2021).

The specific attraction of FinnGen is derived from 'real-life data'—clinical data (patient records, lab results, prescription records, etc.) and personal data from national health or population registers (see Chapters 3 and 4)—which, in combination with FinnGen's comprehensive genome variant data of 500,000 biobank participants, offers huge commercial potential to drug development, as the utilization of such data may considerably speed it up and cut its costs:

> The genome data is combined with health data originating from multiple national health registries. Data from these registries provide longitudinal, lifetime follow-up data from each Finnish resident. This unique data combination allows the FinnGen research team to identify correlations between genetic factors and health outcomes such as disease susceptibility or effectiveness of drug treatments in the Finnish founder population. The study has a huge potential to serve medicine initiatives and enrich drug discovery programs by enhancing drug target identification and prioritization. (FinnGen 2019)

With nearly 150 million euros of funding, FinnGen is an exceptionally grand project in Finnish biomedicine. It is noteworthy that it does not primarily rely on public science funding but on financial resources derived from the public innovation agency and private companies. As it is such a stronghold in terms of finance and research resources, FinnGen has considerable influence in Finnish biomedical science and biobanking. Research at FinnGen favours certain approaches like the utilization of knowledge on the 'penetration' of certain gene variations in Finnish populations, the study of 'loss of function' gene variations, the calculation of the polygenic risk scores (PRS) of common diseases, and the use of wide genome sequencing in defining potential target molecules for new drugs. These emphases may substantially direct resources and scientific interests in Finnish medical genomics more broadly. Perhaps more significantly, FinnGen has affected Finnish biobanks by uniting them as its tissue sample collecting subcontractors (see Chapter 3).

Thus, FinnGen has taken a leading role in the consolidation of commercialization as the master frame for reasoning about biobanking and biobank research in Finland. Its business model both exemplifies and reinforces a reverse version of its claims concerning the value of the benefits and utility of biobank data and research: while emphasizing its scientific nature and declaring that scientific discoveries and medical benefits are the primary and most valuable objectives of the project, striving for commercialization seems to dominate its activities (Helén and Lehtimäki 2023). FinnGen assumes that as a biobank research organization it must *first* pursue major biomedical and pharmaceutical enterprises for collaboration by performing prospects and making promises of commercial benefits. Extensive collaboration with its partners in the biomedical industry will provide finance and other resources for its scientific efforts, which may result in scientific or clinical discoveries that will prove beneficial to patients and healthcare more generally—and thereby increase opportunities for commercial collaboration.

For FinnGen, the instrumental search for commercial benefit in corporate collaboration is nevertheless primary in a temporal sense. In other words, this 'endeavour focused purely on research' (Palotie et al. 2019, 990, own translation) must first focus on ensuring the continuity of financing by attracting international company partners and financiers, before engaging in research activities aimed at scientific discovery and medical benefits. Given this order, commercial prospects are predominant in the valuing of biobank data, infrastructure, and the use of the data in research, as well as in the orientation of research practices, while scientific and R&D activities and achievements are subordinate to this business rationale.

FinnGen tends to highlight the national character of the endeavour. It presents itself as the flagship of the Finnish 'best test bed' brand and a 'honey jar' for Finnish research groups and high-tech biomedical companies seeking access to top-class international research and business ecosystems. Furthermore, FinnGen is keen to claim that its cutting-edge collaborative research has great potential to bring tremendous benefits to Finnish healthcare

and public health in the future. With the charm of this promise, it has managed to recruit and steer Finnish biobanks and FINBB into collecting tissue samples and patient data in order to reach the objective of 500,000 genotyped Finns with personal health data attached to the sample data (see Chapter 3).

Yet FinnGen is a large-scale transnational collaborative research consortium whose prime purpose is to seek collaboration with and finance from big pharmaceutical corporations and other medical and ICT companies by promising the partners commercial benefit and gains. As noted, these commercial pursuits are, in practice, primary and focal in FinnGen's operations, and both scientific activities and the 'national cause' are subordinate to them. Against this background, it seems that FinnGen has made Finnish biobanks its subcontractors in sample and data sourcing, and the flesh, blood, and data of Finnish people resources to be exploited. In these roles, they serve the business model in which the priority is the maintenance of the brand and competitive edge of both FinnGen and Finland in global biomedical research and business.

Capitalizing on 'Finland'

In the late 1990s, many Finnish scientists and research groups were acknowledged as belonging to the global vanguard of medical genetics (see Chapter 2). Almost instantly, thoughts and opinions that this scientific success and fame could also be utilized commercially emerged in Finland. This vision was associated with the idea that the excellence of Finnish medical genetics as science combined with extraordinary sample collections of large epidemiological, population cohort studies, stored mostly at KTL and in population and patient registers, formed a significant national asset: that is, a potential resource to be utilized in the medical genomics business that was taking off around the world. The domestic hype of Finland as a high-tech nation which was embedded in Nokia's success in the 1990s and 2000s, and the international biotechnology boom with its great commercial expectations, provided both landscape and support for the visions of the

tremendous commercial potential of Finnish medical genetics. Moreover, deCODE Genetics and its deal over the establishment of the Health Sector Database (HSD) in Iceland (see Chapters 2 and 3) provided an exemplary pursuit of the commercialization of a small nation's genome and data reserves. Although the effort failed commercially quite rapidly as deCODE faced bankruptcy in 2009, and the HSD was never actually established, at the turn of the century it was a lighthouse for scientists, innovation policy advocates, and entrepreneurs eager to begin a voyage on the high seas of the global biomedical business.

Since the early 2000s, there have been quite a few hopeful ideas, plans, and efforts to commercialize research findings in medical genetics and biotechnology in Finland, yet none of them have followed the Icelandic example. With trial and error and some success in, for example, founding university start-up companies or IPR portfolios, the start of commercialization was rather bumpy. Despite this, scientists and innovation policymakers did not lose faith in the potential of Finnish research in medical genomics and biomedicine for innovation and successful business. By the beginning of the 2010s, national innovation policy had taken a firm grip on cutting-edge medical genomics and biomedicine in Finland, and the drive for commercialization became more intense. Instead of focusing on medical products and patenting, hopes were now invested in scientific and commercial collaboration with academic and commercial partners from abroad, and the biobanks and healthcare data were given a key position in the new visions and 'roadmaps'.

Both innovation policymakers and advocates of biobanks and biomedical science wanted Finland to become specialized in the refinement of data, not just a source of tissue samples and health data. This interest in developing expertise in data management and analytics was considered a more profitable business approach than the bulk work of sequencing done by expensive machines. Yet a requisite for such an orientation is a business model for biobanks and biobank research in which the research organizations actively seek and engage in collaboration with multinational

pharmaceutical corporations and high-tech companies. In turn, success in collaborative pursuits requires the biobanks and research groups to become attractive to potential partners abroad. To enhance attractiveness, leading scientific advocates and innovation policy torchbearers joined forces and composed a national brand out of Finnish genomics, biomedicine, and their infrastructure. This branding presented Finland as 'the most advanced test bed' for biomedical R&D in the world because of its unique public data reservoirs (biobanks, population registers, and patient registers) of a population with a unique genetic profile, combined with extraordinary health data repositories and cutting-edge data management expertise (see Chapter 4). It also consolidated the frame whereby people working in biobanks and biobank research tend to think that the main attraction for company partners is their real-life data—a combination of tissue sample and personal data from population and health registers—and associated high-quality data management services.

Obviously, this branding of Finland was directed at audiences and potential partners in the international domains of biomedical science and business, although the spokespersons of academic research, biobanking, and innovation policy repeatedly promoted the brand in speech and images to professional, political, and lay audiences at home. This was an attempt to consolidate a unified frame of reasoning for the pursuit of medical genomics, and the use of the data infrastructure serving it, among the medical professionals involved and the general public—in other words, to create 'one voice' with which to discuss these matters in public. Domestic branding was also meant to justify the efforts being made to centralize biobank and health data management and the commercialization rationale.

The business model that focused on seeking collaboration with Big Pharma corporations, with 'real-life data' as the main attraction, was embedded in national branding; subsequently, the launch of FinnGen and then its takeover of large proportions of Finnish biobanking partly realized the desire for a 'one-stop shop' for the access and delivery of Finnish biobank and health data.

The consortium then managed to adopt a position as the flagship of the excellence and unique potential of Finland in biomedical R&D and business. This took place through two overlapping activities: successful engagement in worldwide academic networks and projects conducting large-scale GWAS-based studies, with large research populations and massive quantities of data, mapping the genetic risk profiles of a variety of diseases and the potential molecular targets of new drugs—and, in the case of a Finnish project, prolific acquisition of financing through collaboration deals with major pharmaceutical corporations and other medical companies.

The dominance of FinnGen has made certain tensions and contradictions in the instrumental business model of Finnish biobanking and biobank research more salient. Those tensions are not directly related to the 'academic–industrial' partnership, but rather to matching the national ethos with transnational corporate interests. FinnGen presents itself as a project of the whole nation, recruiting Finnish biobanks and healthcare organizations quite widely as subcontractors in sample and data sourcing and summoning every Finn to become a biobank donor. Moreover, the consortium highlights its potential to confer great benefits on Finnish healthcare and its patients, and to enhance the build-up and growth of domestic business ecosystems around personalized medicine. Yet FinnGen is essentially a transnational endeavour that focuses its activities on large-scale, multi-sited research projects and aims to assist its main financiers and collaborators in their efforts to make drug discovery more cost-effective. Thus, FinnGen primarily benefits—or may benefit—top-level research groups or networks in medical genomics, academic entrepreneurs and their innovative firms, and pharmaceutical corporations which operate around the globe and have their home base in the USA.

The above contradiction can be superficially reconciled by reasoning that, taken together, FinnGen, the centralization of data sourcing and access for biomedical R&D, and the intense branding of Finland as the best research milieu create advantage and assets for Finnish research groups and companies in terms

of international scientific and commercial competition. Consequently, the instrumental business model discussed above and the efforts to maintain a competitive edge are considered indispensable for the scientists, research institutions, and companies involved to keep up in the game. Yet it is quite reasonable to ask whether this business model and the assumptions in which it is embedded are justified, and to what extent. Do FinnGen and its model of cross-border, public–private partnership have the potential to generate good business? And if they do, how will the benefits of good business be distributed, and who will benefit most?

In the scale of Finnish science funding, FinnGen is quite exceptional because the amount of nearly 150 million euros vastly exceeds the normal public funding for a medical research consortium. The extraordinary character of FinnGen is underlined by the fact that it raised this funding from business and innovation financing sources, mostly from private companies. FinnGen's funds have also benefited many Finnish biobanks, as subcontracting to the consortium has provided them with the financial resources to start sample collection on a sound basis or extend its scale and to build up material and personnel infrastructure (see Chapter 3). Against this background, it is quite obvious that FinnGen and its business model have been a success.

Despite that, some critics have claimed that FinnGen can hardly deliver healthcare or economic benefits to the Finnish stakeholders it promises. They say that it is arguable whether the knowledge of genetic risks of common diseases FinnGen produces will make any difference regarding the prevention of common diseases and the improvement of public health (e.g. Knuuti and Kere 2020; Kere at al. 2020). Likewise, the critics see that if studies with FinnGen data contribute to commercially valuable findings in drug research or diagnostics, the great majority of revenues and profits will be capitalized on by US-based pharmaceutical corporations and biomedical companies, not by Finnish companies, patients, or public healthcare (e.g. Keski-Heikkilä 2020). Many critics argue that FinnGen's club model, in which for a couple of million euros the company partner can buy an almost

all-inclusive access to Finnish biobank and health register data by collaborating with a FinnGen project, may lead to the selling of Finnish health data very cheaply. In an interview in 2020, an experienced Finnish scientist and biomedical entrepreneur claimed that FinnGen considerably undercharges on its collaboration fee, and Big Pharma companies therefore pay 'just peanuts' for their extensive access to Finnish sample and real-life data; his reference point was the Ernst & Young market forecast report (2019) that estimated the NHS patient data reservoirs in the UK to be worth 5 billion euros per year. According to the critics, the same kind of financial discrepancy is also notable in regard to FinnGen's potential contribution to drug discovery. FinnGen highlights its potential for swift and precise detection, with the help of unique Finnish data, of target molecules for new medicines and rejection of non-effective targets. Such an advantage has considerable economic potential, since eliminating even one wrong candidate molecular target for a new drug with the help of genome analysis can save up to 100 million euros in drug development costs, a sum that greatly exceeds the FinnGen membership fee. All in all, the critics argue that, considering the commercial scale of the medical business in which FinnGen is involved, its business model can bring it comparatively minuscule economic benefits, while the revenue and profits that may derive from the use of Finnish data and expertise will be extracted by company partners abroad.

This criticism raises questions about the role of FinnGen and, more widely, Finnish biobanking and biobank research in global biomedical research and business. People involved in the research field in Finland assume that they have the expertise and capabilities to be involved in cutting-edge global R&D in medical genomics, drug development, and other domains of biomedicine. They also want to avoid a situation in which Finland would be just a provider of research material and data resources and a caretaker of some bulk data sourcing tasks in the global value chain of biomedical R&D. It is difficult to estimate if the assumptions of Finnish stakeholders are correct or not, and how high the ladders of Finnish research groups, innovative companies, or expertise in data

management can reach in the highly competitive global milieu of science and business. The characterization by Celia Young, medical director of the Nordic branch of cancer drug giant Celgene, provides one viewpoint on the FinnGen website: 'FinnGen is a unique data source of genetic information and health data. This constitutes a valuable resource to identify novel targets for drug development' (FinnGen 2022b).

This statement by a corporate research executive is congruent with highlights in the branding of Finland for potential foreign business partners and with what people in Finland assume to be their asset in international science and business competition: the unique data of a unique population, associated with top-class data management services. Efforts by FinnGen and others to facilitate and extend access and delivery of data by centralizing services on a national scale attempt to embrace this asset and maintain its attractiveness. Yet it is justifiable to ask whether health data from a population of some five-and-a-half million, or the genotype-phenotype sample of half-a-million of that population, will be attractive enough for potential corporate partners and whether domestic expertise in data analytics and management will reach the top level in the context of the expanding global health data economy. In the latter, transnational companies like 23andMe, which offer genetic tests direct to consumers, are collecting repositories of millions of DNA sequences and related personal data as a fringe benefit (e.g. Stoeklé et al. 2016), while companies like sequencing platform giant WuXi NextCODE are collecting massive sample data repositories worldwide (Jarvenpaa and Markus 2018). Moreover, cyberspace giants like Google, IBM, and Apple have shown growing interest in gaining access to academic and public health data repositories through partnerships with national governments and regional public healthcare organizations (Sharon 2016; Powles and Hodson 2017; Barber and Molteni 2019; Ngo 2020), and in cases like Italy's public healthcare or the NHS in the UK, patient data repositories are much bigger and more diverse than in Finland.

In 2016, when the global health data economy was beginning to emerge, we interviewed Auria biobank's CEO Heli Salminen, who considered the competitive niche of Finnish biobanks to be quite narrow:

> What pharmaceutical companies might be very interested in is our potential to identify patients for clinical drug studies. … I think that this stratification of research patients is almost the only [way] that we can compete [compared with pharmaceutical trials in India or China].

Since then, the global health data economy and market have expanded and consolidated, making certain questions even more acute today. Do the assumptions and business model on which the biobanking and biobank research ecosystem of a small country like Finland rely actually match with the expectations and assumptions of the pharmaceutical and ICT corporations dominating the global health data economy? Will national data repositories and data sourcing be extensive and attractive enough in the milieu in which evermore extensive data reservoirs are emerging? Will collaboration and 'strategic partnerships' with transnational pharmaceutical and/or ICT corporations create the benefits expected, distribute them to national and regional academic and healthcare partners and small companies, and bring benefits to Finnish healthcare and the society?

Conclusion

In this chapter, we have taken a closer look at the economic and commercial aspects of Genome Finland. Quite soon after some Finnish researchers and research groups gained considerable international merit in the late 1990s and early 2000s, top researchers like Leena Peltonen started to highlight the commercial potential of research made on the Finnish Disease Heritage, related unique genetic composition of the Finnish population, and data in public health and population registers and other health data repositories. This view, or even a conviction, that the Finnish population—

with an extraordinary genomic profile and extensive data depositories of illness and health of that population—are a national asset and bring competitive advantage to Finnish researchers and companies in global biomedical R&D business has been a significant undertone directing the formation of Genome Finland as an imaginary. It has also had a significant influence on concrete efforts and institutions.

During the 2000s, the biotech boom in Finland provided an encouraging milieu for proposals, plans, and projects to accelerate commercialization of academic research in medical genomics and more intense utilization of public data repositories in these pursuits; however, these early initiatives were mostly about trial and error in a terrain quite foreign to academic researchers and public research institutions, and they gave more learning experiences than commercial success to those involved. The 2010s brought major changes. With the national biobanks and Finnish scientists' active participation in international infrastructure building and large-scale GWAS projects, a permanent institutional groundwork for Genome Finland began to take shape. Simultaneously, the stakeholders' core understanding of what the purpose and value of Finnish medical genomics are changed significantly. Until the wake of the 21st century and even during the 2000s, the advocates and vanguard researchers underlined the scientific breakthroughs and benefits to medical care and public health. In the 2010s these expectations were surpassed in the Finnish discussion by the view that the primary value lies in the economic and commercial potential of genetically unique population and health data repositories. This redefinition was promoted by innovation policy institutions like Sitra, the Ministry of Economic Affairs and Employment, and Tekes (now Business Finland) and their officials, and by many leading biomedicine researchers and biobank managers. Innovation policy, with its emphatically commercial objectives, became the dominant frame of discussion about Finnish genomics. Thus, the ethos of Genome Finland shifted from science and health to wealth, which was perhaps the most profound turn in its history.

The FinnGen consortium, launched in 2017, became the epitome of this shift. Although FinnGen's spokespersons characterized the consortium as 'purely' scientific endeavour, in practice its main efforts concentrated on ensuring funding from the national innovation funding agency and, most importantly, through commercial partnerships with medical companies abroad, Big Pharma corporations included. With its overt orientation towards commercial collaboration with transnational medical and pharmaceutical corporations, FinnGen became an exceptionally well-funded biomedical project in the national scale and became the flagship of Genome Finland. In this position, the consortium consolidated a specific rationale and action model, often called a 'business model' in the Finnish discussion, as the core of Genome Finland. This model has two elements: first, even though much of the consortium's efforts concentrate on the pursuit of partnerships with private medical companies, preferably with Big Pharma corporations, such commercialization of genomic research is considered *instrumental*, since its purpose is to guarantee the continuity of the scientific efforts; second, access to unique Finnish genomic data and 'real-life data' in public healthcare repositories that FinnGen can offer to its partners is considered to be an asset that *attracts* potential commercial partners and facilitates the making of partnerships. Branding of Finland as the 'best test bed' for biomedical R&D, started by Sitra, ministries, and leading biomedical scientists in the mid-2010s, is affirmative to this business model and reinforces its assumptions of Finland's uniqueness as a test bed for biomedical R&D and promises of future wealth.

CHAPTER 7

Conclusions: Genome Finland –a fragmented unity

> The FinnGen research project takes 500,000 Finns on a discovery trip to genomic information and future health innovations. The FinnGen research project covering the whole of Finland is a joint study by all Finns, whose most significant findings can be found in anyone's sample and help people around the world. (FinnGen n.d., own translation)

With this characterization, presented only in Finnish and Swedish on the project website, the FinnGen research consortium presents domestic efforts to advance research in medical genomics as a national cause and FinnGen itself as the flagship of this endeavour. For the past quarter of a century, the advocates of Finnish medical genomics in academia, government, and business have created and consolidated similar images that highlight Finland as a competitive and internationally attractive milieu for biomedical R&D, thanks to the combination of a genetically unique and homogeneous population, extraordinary health data reservoirs and data sourcing infrastructure, and top-quality expertise in the related key fields of science and technology.

In this book, we have examined Genome Finland by mapping and disentangling the historical trajectories and political, social, economic, and technological configurations whereby medical genomics—as a national vision, performance, and concrete endeavour—has emerged, advanced, and been compiled and adjusted. In our analysis, we have also unpacked and

contextualized the notions of 'uniqueness', 'unity', and 'success' associated with Genome Finland in regard to the transformation of Finnish society and the welfare state, and the global developments in genomics and the health data economy, addressing themes that figure in contemporary social scientific discussions of these topics. In this chapter, we summarise what we have demonstrated in the previous chapters and present our main argument. We claim that although a shared vision of Genome Finland has been somewhat consolidated over the years, the view both historically and currently is kaleidoscopic: it is still dispersed, and a closer look reveals fragmentations, frictions, and contestations, not simply unity and 'one voice'; indeed, Genome Finland looks very different depending on the perspective of the stakeholder or observer. At the end of this chapter, we discuss what helps to maintain the appearance and the activities of Genome Finland. We also ask what the consequences of such maintenance work are, and whether the picture and promises of Genome Finland are still adequate.

Lineages

The pathbreaking groundwork for medical genomics in Finland today took place between the late 1960s and the early 1980s. The initiatives of pioneers such as Albert de la Chapelle were later taken up by paediatricians Reijo Norio, Jaakko Perheentupa, and many others who detected and classified rare hereditary diseases family by family, mostly in eastern and northern Finland, and grouped them as the Finnish Disease Heritage (FDH). Within international circles of academic medical genetics, FDH signified specific research targets that were enriched in Finland. This provided a springboard for the new generation of geneticists who conducted research on FDH and other Finnish samples and data and showed them to be particularly good for detecting associations between gene defects and certain diseases in, for example, linkage disequilibrium studies. Scholars like Leena Peltonen, Juha Kere, Aarno Palotie, and Jaakko Kaprio broadened the scope of

Finnish medical genetics, paving the way for research from rare diseases to common diseases, along with advancing the field and, most importantly, breaking through into the top international research domain of medical genetics in the mid-1990s.

These geneticists paid specific attention to the sample and data collections of extensive Finnish cohort and epidemiological studies, stored mostly in the National Public Health Institute (KTL) in the 1990s. Leena Peltonen in particular acknowledged the usability of those samples and data in medical genetics research, publicly praising the uniqueness and value of the collections and becoming actively involved in KTL biomedical research. A few reports on biomedical research infrastructure in Finland supported Peltonen's view and emphasized that the samples and data stored at KTL and some Finnish universities have huge value potential because they provide Finnish medical genetics with a competitive advantage in global scientific research and commercialization. This discussion, highlighting existing samples and data from previous cohort and epidemiological studies, set the stage for the self-image and promotion of medical genomics in Finland for almost three decades: the Finnish sample collections and health data reservoirs are unique in the world and, therefore, a precious resource for Finnish genomics and other biomedical research that should be made available for more intense use.

The above discussion in the early 2000s exemplifies an essential trajectory of the Finnish medical genetics we have highlighted throughout the book, namely, the entanglement of the development of medical genetics research in Finland with the building of the Finnish welfare state and its data sourcing, aligned with the rise of epidemiological research and specific public health concerns. When researchers and policymakers alike mentioned reports discussing Finnish health data collections as an underutilized resource for biomedical research, they often made specific reference to the KTL collections. These were produced by extensive epidemiological cohort and follow-up studies like the Mobile Clinic Study and FINRISKI, launched at the end of 1960s to support the planning and execution of health insurance and

public healthcare reforms between the mid-1960s and 1970s. Repurposing the data and samples collected in epidemiological and public health studies two decades after the initiation of those studies generated a concrete bond between the welfare state's data sourcing and domestic scientific endeavours in medical genomics on many levels. This connection was later repeated and consolidated in Finnish biobanking, the FinnGen research consortium, and efforts to accelerate the secondary use of healthcare and social service data. In these contexts, the connection provided the basis for the view that the unique genetic composition of the Finnish population, coupled with the public healthcare and associated, well-ordered data sourcing within the Finnish welfare state, form a resource providing an advantage for Finnish biomedical research in global competition, as we have demonstrated in Chapters 3, 4, and 6.

It is apparent that the supposed success story of Finnish medical genetics and its promotion into the 21st century were entangled with the worldwide genomics hype associated with the Human Genome Project (HGP) at the turn of the millennium. Enabled by technological development and growing public and private investment, the expansion of medical genomics created an environment of global scientific and commercial competition. Academic research institutions, private companies, and nation states vied for resources, fame, and commercial opportunities, turning high-quality samples and health data into an asset. Consequently, the national initiatives often used as exemplars in this book—those of Estonia, Iceland, the UK, Singapore, Denmark, and, of course, Finland—focused on building infrastructure, most notably biobanks, for sample and data sourcing, management, and distribution that would also facilitate the formation of a national 'ecosystem' for the research and commercialization of medical genomics. In these countries, the groundwork for such an ecosystem could be constructed by connecting public healthcare, repositories of population and patient data, and academic institutions, and then bringing—or attracting—private companies and investors on board. The situation in these countries was very

different from the USA, which is hegemonic in global biomedical science and business. There, however, healthcare is predominantly private, and universities and research institutions are business-minded, which does not allow ecosystem formation based on public institutions nor the utilization of public healthcare and other public systems in data collecting.

Scientific and technological development has a wider context of politics and the economy into which Western governments and transnational organizations like the OECD and the EU put their hope, investing in the 'knowledge economy' as the source of wealth and social progress. The term refers to the idea that technological innovations are the most significant source of economic growth in advanced industrialized countries, and the natural, life, and engineering sciences have great potential to accelerate innovations, which should be deployed by governments and private companies to boost the economy and business. The economic crises of the 1970s across the Western world provided an environment for the above ideas to influence national policymaking, and in the following decades the view that investment in scientific research and technological innovation is the main means to increase and maintain economic growth became a self-evident element of policy reasoning all over the world. The activities—mostly political—that aimed at building up knowledge economies have significantly reshaped academia since the 1980s, as universities and public research institutions have been encouraged and obliged to become business-oriented and managerial in order to direct research toward technological innovation in partnership with private companies or investors. The general development of an academic capitalism with the ethos of entrepreneurial science (Etzkowitz 2002) fundamentally modified emerging or rapidly advancing sciences and technologies.

Genomics was one such novel and promising field of science and innovation, and, not surprisingly, the HGP and medical genomics more broadly were also associated with the tremendous economic and commercial hype at the end of 1990s and early 2000s. Consequently, plenty of public and private investment

was made in medical genomics with expectations of new revenue sources. Genomics also became a prime domain of venture capital investment at that time. Within a milieu framed by HGP hype and grand commercial expectations, the national and regional genome initiatives mentioned above and discussed in this book were launched, also, or even predominantly, with future economic and commercial gains in sight. Finland was one among many countries—including the UK, Estonia, Iceland, and Singapore—where a national medical genomics endeavour initially started in order to advance biomedical science for the benefit of medicine but swiftly geared up to encompass the objectives of promoting biotechnology and related economic expectations. Eventually, the latter came to dominate the landscape and roadmaps of research in the field (Tarkkala et al. 2019).

The principle of legality

At the beginning of the 21st century, enthusiasm for the mapping of the human genome resulted in the launch of extensive national and regional genome projects and biobank infrastructure to meet an increasing need for samples and research data in genomics-driven biomedical research. In Finland, advocates of novel genomics in academia, the state administration, and politics acknowledged and emphasized that existing research and clinical sample collections and associated health-related data from a 'genetically unique' population constitute a resource with extraordinary international potential for research and commercialization. They argued that this may open doors for Finnish researchers into the first-class section of the post-genomics train that was moving at full speed around the globe.

The Finnish stakeholders took a legal route to facilitate the utilization of sample collections and health data reservoirs and to intensify the collection of new samples and associated data from the population. Leading academics, government officials, and politicians shared the view that a specific law and regulatory regime should be established first, because a solid and clear legal

framework would allow swift, efficient, and appropriate collection, management, and utilization of genome data, at home and in cross-border collaboration (see Chapters 3, 4 and 5). A milestone in the actualization of this reasoning was the start of preparations for the 'best' biobank law in 2005–2006.

Finland was not the only country that passed specific legislation on biobanking or the collection and use of genetic knowledge. However, it is rather extraordinary that the rule of law has been considered primary and even sufficient grounding for extensive sample and data sourcing in the Finnish discussion. In most Western countries, intense and multidimensional academic, professional, and public discussion on informed consent and other ethical issues pertaining to the new kind of data sourcing and usage was a landmark of the post-genomics era into the 21st century (Lauss et al. 2011; Reardon 2017). Consequently, it was seen as vitally important that data management in biobanks and genome projects should be embedded in explicit and accurate ethical guidelines and procedures that protect the privacy and personal integrity of the donors, patients, and citizens (see Lauss et al. 2011). In places like the UK, extensive public consultations were undertaken before establishing large biobanks, including the UK Biobank. In Finland, discussion on ethics was rather marginal, lame, and meagre, while the academic and political stakeholders primarily focused on questions of legislation and official regulation that would both enable more extensive collection and use of sample and health data in genomics research and development (R&D) and protect the donors and research subjects.

'If something is legal and approved by the regulating public authority, then it is ethical'—this kind of idea seems to have been the ethos of the advocacy of genomics and the required data infrastructure in Finland. The Finnish stakeholders have been rather consensual in assuming that the laws and the orders and instructions of the regulating public authority (Valvira until 2019, now Fimea) provide an ethical safeguard and justification for biobanking and other data collection and use in the service of both academic and commercial genomics. Such an opinion implies that

public regulation releases researchers, research institutions, and private companies from bothering too much about ethical reflection on their data collection and use. Appeal to the rule of law has also been used as justification for straightforward lines of action like transferring so-called legacy samples and associated data to biobanks without the donors' consent, and suggestions to bypass informed consent procedures in the research use of samples and patient data collected by public healthcare and social services (see Chapters 4 and 5).

However, academic, political, and business advocates and stakeholders of medical genomics in Finland have had an ambiguous attitude to the law and associated regulation. On the one hand, they have highlighted that specific legal regulation makes the rules of biobanking and other data sourcing transparent and unequivocal, which provides Finland, once again, with an advantage in international competition. On the other hand, many of them have complained that the laws and regulations are too rigid and have fallen behind scientific and technological development, thus introducing unnecessary obstacles for contemporary research. In the same vein, the Finnish biomedical research community has extolled the advanced level of the Finnish research environment on an international scale, while at the same time raising alarm bells that Finland is falling behind and needs to reorganize its research environment to be more flexible.

Partly because of this tension, creating a legislative and regulatory framework for data collection and usage has taken a considerable length of time; furthermore, it seems to be constantly under construction and becoming increasingly complicated as the process endures. It took over five years to get the first biobank law passed in 2012, and people involved in biobanking soon started to complain about its inadequacies. Such dissatisfaction initiated an administrative process to reform the law, which took a further decade. The reformed Biobank Act was finally accepted in parliament in February 2023. During the 2010s, preparation for several new laws on data sourcing and use related to biomedical R&D were launched by the Finnish government, which generated

diverse and ambivalent responses and confusion among the stakeholders and professionals in the field instead of increasing the clarity and transparency of the rules and limitations on action. As we demonstrate in Chapters 3 and 5, frequent revision and endless preparation of legislation have resulted in fuzziness in terms of building biobanks and other data management infrastructure projects.

With one voice?

The efforts to develop a consensual and unified vision—speaking with 'one voice'—of the essence and the strengths of Finnish medical genomics, and its prospects and future direction, have been exceptionally persistent in Finland. As we have shown throughout this book, government officials, leading academics in biomedicine and genomics, and the national innovation agency and semi-public innovation think tank Sitra (see Chapter 6) have joined forces to portray Finnish biobanks and data infrastructures, healthcare and research facilities, and genomics research in medicine as forming an efficient unity like no other in the world. Furthermore, they have urged regional healthcare organizations and biobanks, research groups and institutions, the keepers of public data repositories, regulating authorities, and legislators to follow this lead and execute national strategies and roadmaps whose goal is to consolidate this ecosystem and speak with one voice about the prospects and benefits of Genome Finland. These efforts have resulted in a national genome vision in which scientific and commercial objectives are entangled. The main elements of this ideology are the so-called success story that brings the past and the future of Finnish medical genetics together, an assumption of the competitive edge provided by Finland's unique population and exceptional data repositories, and a pronounced shared ambition to advance Finnish post-genomics. This ideology and an accompanying sense of urgency to take swift and rapid measures to accelerate Finnish genomics given global competition have impregnated almost the entire reasoning, discussion,

and practices in the field portraying itself as Finnish biomedicine (Tarkkala et al. 2019; Tarkkala and Snell 2022).

Despite the performance of national unity and the genuine pursuit of it, efforts to advance Finnish medical genomics as science, innovative R&D, and commerce have engendered multiple overlapping strategies and projects with diverse interests and objectives. This commotion has resulted in tensions and contests that have mostly remained implicit and eclipsed by the discourse of 'one voice' and 'shared ambition' among the stakeholders (Tarkkala and Snell 2022). At the level of national policy, the proposal for the Genome Strategy by the Ministry of Social Affairs and Health (2015) was made to match the general innovation and economic objectives of the Health Sector Growth Strategy by the Ministry of Economic Affairs and Employment (2014). However, the two national strategies and the related roadmaps were not fully coordinated with each other, nor with the endeavours to enhance and widen the scope of the secondary use of health and social service data repositories (see Chapters 3 and 6).

Diversity of action was apparent at ground level, exemplified by biobanking. From the very beginning, biobanking in Finland took several routes: cancer researchers in Helsinki pioneered the development of their own arrangements and piloted the concept of disease-focused translational biobanks, while researchers at Turku University Hospital moved swiftly to establish a regional clinical biobank. Simultaneously, the start was much slower for the KTL epidemiological sample and data collections and focused on lobbying for the biobank law. Investigations of biobank mergers also occurred in parallel. Initially, three regional biobanks—Auria, Tampere Biobank, and Borealis in Oulu—began to investigate this possibility in the mid-2010s. The Ministry of Social Affairs and Health took over the initiative, extended it into a national project, and established a task force to plan a national-level merger, which eventually resulted in the establishment of the national biobank cooperative, FINBB, in 2017, which the majority—but not all—of Finnish biobanks joined as members. Meanwhile, concurrently with the governmental activity, Aarno Palotie

and other major research directors at the Institute for Molecular Medicine Finland (FIMM) were preparing the FinnGen initiative, which was launched a couple of months earlier than FINBB. Funded by both the public innovation agency Business Finland and pharmaceutical corporations, FinnGen had become the principal customer and a significant funder of Finnish biobanks by the 2020s. At the time of writing this book, FinnGen is the national genome initiative in Finland, and, to a large degree, it defines the objectives and pace of biobank operations, a situation to which FINBB has also adjusted its Fingenious data management service (see Chapter 3). It is noteworthy that from the beginning of the 2000s persistent national efforts were made to coordinate and steer biobanks, other data infrastructures, and the genomics research, development, and innovation ecosystem under a public authority, yet today such coordination of the national genome project seems to be—in practice—executed by FinnGen outside the public framework.

The overlapping efforts directed towards the national centralization of biobanking in Finland were entangled with the unfinished and multiplying law-making (see Chapter 3), the preparation of enabling legislation to extend the secondary use of health and social service data, and the associated construction of a national health data hub (eventually Findata). Sitra in particular, which had already piloted a 'one-stop shop' from 2016 to 2019 in the Isaacus project, actively advocated the latter. Yet, at the same time, many prominent scholars, especially in cancer research, were expressing concern and doubt about the benefits of biobanking for their work, while a number of research groups in biomedicine did not want to transfer their samples and data, often collected over decades, to regional biobanks. Throughout the period from the mid-2010s to the present day, experts and other stakeholders in the field of Finnish genomics apparently have not had a clear picture of what actions will eventually be taken or the outcomes of all the strategies, plans, and projects, even those they have been involved in drafting and advocating. They have been wondering about the role of regional biobanks, how Findata as the national health data

hub will relate to the biobank fusions and FINBB, what exactly the planned genome centre will entail, and whether it deserves support. A sense of confusion has been discernible, for example, in several commentaries directed at ministries concerning the drafts of laws that have been circulated for comments (see Chapters 3 and 5).

Discussion of these questions reflects differences of opinion and even disagreements, but signs of genuine confusion about the actual nature, objectives, and benefits are also prominent. This is fuelled by a sense of urgency and rush in strategy papers, plans, and initiatives that is justified by appealing to the need to take rapid advantage of opportunities for research and commercial collaboration between Finnish stakeholders and international partners and investors. The proposal for the Genome Strategy (Ministry of Social Affairs and Health 2015a), for example, exemplifies this imperative to take fast action—one that has been repeated in Finnish discussions, even using the same metaphor, for over two decades (Tarkkala et al. 2019; Tarkkala and Snell 2022)—stating: 'The window of opportunity for exploiting Finland's strengths will be open for a few years at best.' Ultimately, despite the performance of unity and 'speaking with one voice', Genome Finland consists of commotion and plenty of erratic and somewhat disoriented tinkering.

Nonetheless, the national genome vision has been widely shared in Finland. As a result, there has not been much deliberation and open discussion about the line of action or the projects that should be undertaken to advance genomics research in Finnish biomedicine, let alone their reasonableness. Views diverging from the mainstream promotional discourse have been ignored or repressed, wide public discussion of the impact of genomics and related biotechnology in society has not been encouraged, most critical professional and public voices have been silenced, and dissident arguments have usually been ignored or overlooked by the strong scientific and political advocacy of biobanking and personalized medicine initiatives. Throughout the first two decades of the 21st century, the professional and public hegemony was

almost fully consolidated, allowing Genome Finland to proceed with minimal public or professional reflection or critical deliberation. It was not until the 2020s that critical discussion began to get a foothold in professional forums, nationwide media, and public discussion (see Chapter 6).

The persistent pursuit of consensus and ideological unity in Finnish medical genomics has been accompanied by a kind of professional and medical paternalism. This has been characteristic of the way academic advocates, innovation policymakers, and governmental officials think of and relate to sample and data donors, patients, and citizens in the context of Genome Finland. The prominent promoters of biobanks and FinnGen have repeatedly presented themselves and their projects as benefactors of the nation and the Finnish people, justifying their goals by claiming that the advance of genomic research will bring benefits to all. The projects and their promoters appear to be committed to continuing their efforts in the best interests of 'all Finns', to the extent of summoning everybody on 'a discovery trip to genomic information and future health innovations' (FinnGen n.d.; see also Tupasela 2021). Arguably, however, the claim of improved well-being for all is reiterated with such frequency in order to ensure that any dissenting voices, opinions, concerns, or aspirations of patients, clients, and citizens do not disturb the march of progress, a top-down attitude that has been most apparent in relation to issues of sample donation and health data sourcing. As discussed in Chapters 3 and 5, biobanking of so-called legacy samples from existing epidemiological and clinical collections without personal informed consent is a telling example of this, as are the proposals in the mid-2010s that biobanks should rethink the need for consent (see Chapter 5) for sample collection—the latter was a reaction to the concern that the biobanks could not catch 'all incomers' and would not be able to collect enough new samples (see Snell and Tarkkala 2019).

Seemingly, the idea that Finns should have their say as stakeholders, co-producers, and citizens in the business of genomics and biomedicine has been—and still is today—a challenging one

for the principal academic promoters and policymakers. The general attitude seems to be that Finns—each and every one of them—are providers of resource material for biomedical research, development, and innovation, and willing donors with favourable, positive attitudes toward research. However, there are some examples, albeit very few, of more participatory approaches and attempts to listen to participants and patients: the FHRB Biobank, for example, collaborates with a cancer patient organization participating in the steering group and planning of operations. In addition, before establishing its biobank, the Finnish Blood Service conducted a thorough and cautious deliberation about the blood donors' possible reactions and attitudes to biobank sample donation. During recent decades, surveys and group discussions have also been conducted to gauge the attitudes and understandings of citizens in terms of biobanks and the use of genomic data more generally. Yet it is not always clear how these have been taken into account in the further development of legislation and organizational structures or ethical review processes in the field.

From health to wealth

In the early years of the 2000s, a new element emerged in the discussion when assessment reports of biomedical research and writings by a few frontline geneticists highlighted the underutilized potential of Finnish sample collections and health data repositories. In combination with top-class Finnish genetics research, these repositories could provide a competitive edge for Finnish researchers in global science domains and especially for the biomedical and pharmaceutical business developing around genomics R&D after the Human Genome Project. Genomics as the spearhead field of biomedicine was seen as bringing economic benefits to Finland vis-á-vis the potential for facilitating progress in medical care. As we have discussed throughout this book and especially in Chapter 6, this rationale and the pursuit of 'commercialization' permeated the reasoning and efforts by which medical genomics were embraced and successfully advanced in Finland.

By the 2010s, innovation policy, with its commercialization, national branding, and business ecosystem rhetoric, had become the master frame for biomedical research in genomics and post-genomics; meanwhile—as promotors of national innovation policy—politicians, governmental officials, and Sitra have been at the helm of Genome Finland ever since. It is notable that this innovation policy frame has been applied to the consolidation of biobanks, conducting the national biobank merger with FINBB and its Fingenious search portal service, pursuing the extension of the secondary use of health and social service data and the establishment of Findata, and development of the FinnGen consortium. As a result, medical genomics has become closely associated with national strategies to enhance business and imports of healthcare technology, and with the commercial prospects of personalized medicine.

At the core of the business model for Genome Finland is the idea, or even the imperative demand, to transform Finland's specific competitive advantages—unique population, biobanks and other health-related data repositories, the public healthcare system, and top expertise in biomedical research and data management (see Chapter 4)—into a test bed for biomedical research and development. This would make Finland a top-class R&D milieu that would almost irresistibly attract academics and—possibly more importantly—pharmaceutical and other medical companies from abroad as collaborators and investors (see Chapters 4 and 6). The stakeholders widely believe that by being attractive in this way Genome Finland will bring economic benefits to Finnish researchers, high-tech companies, and the whole of Finland. As we discussed in Chapters 4 and 6, biomedical science in this national landscape serves R&D and its primary purpose to accelerate biomedical and pharmaceutical business and facilitate the production of wealth by companies and the nation. In this scenario, the whole of Finland—its population, data infrastructure, public healthcare, and people, sick or healthy—is reduced to the mere provider of raw material for biomedical R&D, which

facilitates efforts to keep Genome Finland competitive in the business and science domains of global genomics and biomedicine.

It is noteworthy that frontline academics in biomedicine and genomics have actively highlighted the economic potential of Finnish genomics and related data reservoirs and promoted their commercialization. In the late 1990s, Leena Peltonen started to attain the position of torchbearer for Finnish molecular genetics in medicine and became its most visible public representative. In the early 2000s, in articles published in professional journals and numerous interviews in a variety of professional and popular magazines and newspapers, she repeatedly endorsed more intense and systematic utilization of existing sample collections and public healthcare data repositories in Finnish genomics research, in collaboration with academic and corporate partners from abroad. She envisioned that such collaborative research would provide remarkable benefits for the Finnish national economy by improving public health and boosting domestic biomedical business. She argued that Finland already has a unique asset in epidemiological and cohort studies sample collections and patient and population data repositories in public healthcare, which provides Finland—and Finnish researchers—with a considerable advantage in international scientific and commercial competition. In addition, Peltonen promoted the plan for a national genome centre that would facilitate access to Finnish data and collaboration between Finnish research institutions and private company partners at home and abroad (see Chapters 3 and 6). She also urged Finnish stakeholders to take rapid action to catch up with competitors like Iceland and the UK.

When looking at the trajectory from Leena Peltonen's call to arms in 2003–2005 to the present-day situation in which Meilahti Campus-based FinnGen, an 'industrial–academic partnership', has achieved flagship status for Genome Finland, two traits are clearly observable. First, the ideas and basic elements upon which to build the future success of Finnish medical genomics existed and were already being touted at the dawn of the new millennium, and, second, the rationale and line of action have remained

essentially the same for two decades. The modus operandi common to FinnGen, biobanks, and FINBB is based on the pursuit of public–private partnerships with pharmaceutical and other medical company partners and investors from abroad, and such collaboration is assumed and hoped to radiate throughout the domestic ecosystem and boost high-tech biomedical business at home. A national genome centre does not exist, but FinnGen takes care of many of the functions mentioned in the original plan as the facilitator of access to Finnish data and coordinator of collaboration between foreign company partners and domestic data depositories and experts in biomedicine and data management. However, FinnGen's situation is uncertain, because it is not an institution or an official authority, but rather a project that private pharmaceutical companies and Business Finland have agreed to fund from 2017 to the end of 2027. At the time this book is being written, the fate of the project and the collected samples and data once the project ends is unclear. This dilemma has been at the forefront of many public presentations by FinnGen researchers, since the lack of institutionalization poses challenges for the future of both the data and the samples.

Another trait is also evident. The line of action adopted by geneticists at Meilahti and FIMM at the turn of the millennium and the route they have taken since then have left profound marks on the formation of Genome Finland. The mission and efforts originating with FIMM have affected and shaped the strategies, roadmaps, objectives, and ethos whereby medical genomics have been advanced in Finland up to the turn of the 2020s. People from FIMM have been among the most eager advocates, especially Professor Aarno Palotie, who has been the most eminent lobbyist in the efforts leading to FinnGen and the consolidation of Genome Finland since the death of his wife, Leena Peltonen, in 2009. Palotie has promoted similar economic justifications, commercialization rationales, and demands for the removal of legislative and regulatory hurdles as the health sector innovation policy advocates at Sitra and the ministries. Genome Finland has by no means been built and promoted solely by and for FinnGen, yet FIMM

initiatives leading to FinnGen have been quite successful. This is indicated by the fact that currently Finnish biobanking is to a great extent dependent on the consortium, and FinnGen's public–private business model also dominates post-genomics research in Finland in terms of funding and cross-border R&D collaboration. Nevertheless, there still seems to be continuous worry about whether the right things are being prepared for and achieved in Finland and how Finland should develop in order to adapt to the needs of potential collaborators and customers.

Finland is not the only country where grand-scale genome initiative and personalized medicine programmes have been advocated and carried out with economic benefits in mind. After HGP, research in biomedicine and other fields of biotechnology has predominantly followed the track of 'entrepreneurial science' (Johnston and Edwards 1987) all over the world. Policymakers and influential academics together have consolidated the rationale that new genomics could be a tremendous source of medical innovation and wealth and that the benefits promised by genomics can be delivered to people only through commercialization and markets. In small countries like Finland, the pursuit of success in the global biomedical market through new medical innovations and collaboration with dominant academic and company partners has been intertwined with the ethos that advancing the genome initiative and personalized medicine is a common national effort. In countries close to Finland, like Iceland, Denmark, and Estonia, post-genomics and related biomedical research infrastructures have been similarly promoted through an ethos that combines the orientation of biomedical science towards global commercial markets and an emphasis on the national cause and gain—in other words, as a quest for a 'new Nokia'.

A parallel ethos can be found in many countries and regions far from Finland and Europe. For example, the Biopolis initiative in Singapore (Ong 2016; Aarden 2017) was built up as an emphatically national project seeking to enhance and embrace the competitiveness of domestic research institutions and companies, a rationale essentially similar to that of Genome Finland. However,

the Singaporean initiative focused on the 'Asian' genome as an asset, and thus it placed more explicit emphasis on the ethnic and even racial aspect of the project than those in Finland, where balancing between uniqueness and more general appeal is under constant negotiation. Moreover, the efforts to develop the business aspect of Biopolis were clearly articulated as if directed against commercialization in the sense that they fight the dominance of US-based Big Pharma companies and their efforts to commercially colonize South Asian populations, data, and scientific expertise. Such a post-colonial view and rationale for commercialization are absent from Finnish strategies and discussion. On the contrary, Genome Finland is open and welcoming to collaborative and investor partners from abroad, with the expectation that the resulting science will be benevolent and beneficial to all. Consequently, Genome Finland attempts to facilitate their partners' access to a 'unique' population and its data, which implies that the Finnish population and its data are both conceived of and constructed as a global resource to be commercially and scientifically exploited, meanwhile being considered a national 'treasure'. Such an ethos can also be found in the genome and personalized medicine projects of the other Nordic countries.

At a crossroads

In the chapters of this book, we have de- and reconstructed the story of medical genetics in Finnish society, beginning with early research aimed at working with and helping families whose members either suffered from, or were likely to fall ill with, rare hereditary diseases, and following developments through to more recent national efforts to build and maintain a biomedical test bed nation attractive to scientific and commercial collaborators from abroad. One feature of this evolution is particularly evident: coordinating and building up Genome Finland has been predominantly a top-down initiative—through legislation, under the coordination of the ministries, and in the form of cross-sectoral strategies and big campaigns for branding and marketing Finland as the 'best' test

bed. Thus, all the ideas and work to build and maintain Genome Finland can be seen as efforts of a small nation's biomedical science to adapt into and keep up with the demands of global medical technoscience and related market environment; this endeavour and its various aspects we have portrayed in this book.

Although there seemed to be a way and apparently the will to push Finnish genomics and personalized medicine towards the top of the global science and business world, the progress of building and streamlining data sourcing and the ecosystem was felt to be too slow. Biobanks, researchers, companies, and other stakeholders in the field often became impatient and started projects of their own. This has resulted in the building of competing infrastructures and data hubs, which has undermined national coordination efforts. Assembling diverse operations, pilot projects, routes of samples and data, and their multiple uses into a coherent legal and operational framework has become very challenging. In many cases, the creation of a unified Finnish approach has proved difficult in practice, as the issues raised by informed consent procedures testify (see Chapter 5). Thus, despite a unified vision and roadmaps, Genome Finland has remained fragile. National political and innovation strategies focus on combining different sorts of health data, enabling easy access to that data, and promoting the whole system of public healthcare as an asset for biomedical research and business—a rationale which actually has little to do with what is happening in the fields of medical and healthcare innovation. Certainly, there are many efforts, projects, and innovations in Finnish biomedicine and health technology that can be considered successful; however, these success stories have mostly evolved independently of the visions of national centralization and related coordination efforts.

As we prepare this book for publication in 2024, it appears that Genome Finland is coming to a crossroads. The consolidation of a legislative and regulatory framework continues to be an unfinished and complicated business; the revised Biobank Act was passed only after a decade-long process and the genome law has been in preparation since 2016. During the past few years, more

vociferous professional and public criticism has emerged, targeting the establishment of a national Genome Centre and related privacy and data protection issues, the proposal to adopt an opt-out practice for biobank sample and data sourcing, failures in the reform of the secondary use of health and social service data, and even the GWAS paradigm in genomics. Simultaneously, implementation of the national genome strategy has proceeded in small steps, and the updated strategy papers list practically the same objectives, point out the same development tasks, and draw identical roadmaps based on the same idea and rhetoric of Finland as a test bed as nine years ago. Furthermore, debates and discussions of the national genome strategy, published in early 2023 (Finnish Government 2023), were significantly limited to experts and supportive patient organizations, and there has been little space for dissenting voices to raise critical issues.

The idea of biobanking has been tied to legislation, but also aligned to the issue of the relationship between the Genome Centre and biobanking in recent years (see Chapter 3). Simultaneously, there is growing concern about the continuity of funding for the regional biobanks, because the great healthcare and social services reform (MSH n.d.) shifted ownership of these biobanks to the new well-being services counties (HVA) in charge of all regional healthcare and social services. Already funding for the HVAs seems to be so scarce that prospects of maintaining and developing regional biobank services are not very encouraging.

Expectations are still sustained, and faith in Genome Finland is firm, according to what is shown to the public and potential collaborators and customers in promotional materials and strategies. However, the playground of Genome Finland is currently increasingly defined and dictated by the global markets of biomedical and pharmaceutical R&D and the emerging global health data economy, and it is unclear whether the scientific, technical, and economic assumptions about the attractiveness of the Finnish biomedical R&D test bed are accurate in this context. Big transnational companies rule the global health data economy, and companies like 23andMe are able to collect repositories of millions of

DNA sequences and related personal data from all those using their services to trace their ancestry and genetic health projections (Stoeklé et al. 2016). Similarly, ICT giants like Google, IBM, and Apple have shown growing interest in gaining access to academic and public health data repositories through partnerships with academic institutions, national governments, and regional public healthcare organizations (Sharon 2016; Powles and Hodson 2017; Barber and Molteni 2019; Ngo 2020). Facing this situation, the advocates and stakeholders of Genome Finland will inevitably have to reflect upon certain questions: Will national data repositories and data sourcing be attractive enough to academic and corporate partners from abroad when there are ever-more extensive reservoirs of big data for utilization in biomedical R&D? Will being a test bed and the instrumental business model based on collaboration and 'strategic partnerships' with transnational pharmaceutical and ICT corporations create scientific, medical, and economic value as expected?

For over two decades, Finnish biomedical research policy and strategy have emphasized the significant role that Finnish tissue sample collections and health data repositories can play in bolstering international competitiveness and wealth creation. The so-called uniqueness of the Finnish population has been a major selling point over the decades and continues to be so. There is no doubt as to the high quality and excellence of Finnish research and the significant strides that have been made in the reorganization of national resources to make them more available. At the same time, however, it might be prudent to pause for a moment to ask what 'competitiveness' and a 'competitive edge' actually mean in today's global arena of biomedical research and innovation. This question can further be extended to ask who gets to define the criteria of success in relation to the history and future of genetic and genomic research in Finland.

From a public health perspective, it is difficult to discern any significant impact and benefit that has accrued through high-tech genomic medicine. Even the early studies on rare diseases were, by definition, limited to a small number of individuals and families.

Undoubtedly, the study of FDH and the concomitant care and counselling that individuals and families have received has been significant and important, yet the application of the findings of contemporary genomic studies—using polygenic risk scores in disease prevention, for example—tends to be far more modest.

From a commercial perspective, it is clear that FinnGen is by far the most successful public–private partnership in medical research that Finland has seen to date. It is unclear, however, how such success translates into broader sharing of the promised benefits within the Finnish context or whether such expectations are even well grounded. Certainly, when compared with some of the early expectations and hype connected with genomic research in Finland, it would not be presumptuous to expect a more robust diagnosis and dissection of the benefits of its public–private partnerships. The notion of 'competitive edge' appears to be a rather loose and fuzzy buzzword that is continually deployed to create a sense of urgency. It remains unclear, however, whom or what Finns are competing against or with, and what would happen if a redefined strategy *not* based on such unclear claims of urgency were to be followed and implemented.

Another observation we find interesting in our work has to do with the significant impact that a relatively small number of researchers in the capital region of Finland have had on regulation and research policy on biomedicine in Finland. The FinnGen project has been a major undertaking in relation to Finnish research capacity, yet it reflects a systemic problem in Finland, namely, the lack of long-term planning and science policy at the national level. The fact that FinnGen is a project that has an end date begs the question of what will happen to all the data and information generated as it runs its course, a question with which the spokespersons of the FinnGen project are also grappling. Because of the lack of consistent, long-term funding, there is little clear vision or direction in biomedical research in Finland. Consequently, science policy and funding tend to be very cyclical and dependent on the whims and trends of innovation and industrial policy, introducing an element of arbitrariness that is responsible for wasting

resources, while not providing a clear picture for the public of the vision and plans that public policymakers and scientists may have for all the data collected and analysed. This shortcoming is also problematic in that the visions that contributed to the establishment of the FinnGen project were not necessarily shared by the biomedical research community in Finland more broadly, but rather were generated by a small group of individuals in Helsinki. It is prudent to ask to what degree contemporary genomic medicine in Finland can help drive the commercial expectations of the Finnish health sector in general.

Finally, we would like to highlight aspirations to make Finland a globally attractive research milieu or test bed. The recent and unprecedented harnessing of tissue samples and health data for secondary purposes also has significant repercussions for citizens, patients, healthcare, and science in general. A discussion in the Finnish newspaper *Helsingin Sanomat* regarding the sharing of health data through the national Kanta healthcare portal reflects some of this ambiguity. An op-ed piece describing the shock felt by an individual upon discovering that all her healthcare and prescription information was available to her optometrist highlights the challenges people have in understanding how personal data of this nature flows between different organizations and companies. In a follow-up commentary, a public authority tried to clarify the matter, but the response seemed to create even greater confusion, since it was clear that there are a very large number of actors with whom private health data could be shared and numerous situations in which this can happen. Although there can be clear benefits in sharing a patient's health data between healthcare professionals, the discussion has highlighted how unaware individuals are of the extent to which this takes place, and how complicated its management can be in the current climate of data sharing.

The increased interest in facilitating data sharing also creates questions regarding who benefits from it and in what ways. As it has become increasingly difficult to understand and manage personal health data in online platforms, the possibility of transparency has also become increasingly suspect. If data policy continues

to be expert-led and lack public involvement, the legitimacy of data sourcing and sharing in public healthcare services may come under question. Our work over the decades has highlighted some of the important transformations that have been taking place within the Finnish context in relation to genetic, genomic, and healthcare data and data policy. Given the trajectory that we have outlined in this book, we should ask whether we need to take stock of the driving forces behind these changes and whether they need to be reassessed or even rethought. Even more importantly, we need to ask how we should go about reforming the Finnish biomedical research vision and which stakeholders ought to be invited into the process of renegotiation.

Notes

1 Leena Peltonen married her colleague Aarno Palotie in 1981, and she became known as Leena Palotie and subsequently Leena Peltonen-Palotie in international forums of molecular genetics in the 1980s and 1990s. In the 2000s, she returned to using her birthname Peltonen, and we therefore use that name throughout the book.

2 Although the title of our book is *Genome Finland*, our focus stretches back to the early 1960s when the term 'genetics' was more commonly used. Only after the mapping and publication of the human genome do the terms genomics, medical genomics, and post-genomics begin to take centre stage in our story. It should be noted, however, that we do use genetics and genomics, human genetics, medical genetics, and medical genomics interchangeably throughout the book, often to refer to the same phenomena, since these terms are also used interchangeably in the literature and by our informants. We use those terms to refer to genetics as applied to humans and medical issues, leaving out plant and animal genetics.

3 We have studied molecular genetics and genomics, biobanking, and personalized medicine in the following projects and assignments: *Rights and responsibilities in biotechnology* (2002–2007, funded by Tekes); *Molecular medicine and public health* (2007–2010, University of Helsinki); *Privacy regimes in variation and transformation: The emerging field of post-genomics* (2009–2012, Academy of Finland/ELSA GEN); *Constituting difference through genetics – From historical to naturalistic explanations of population variation* (2010–2012, Academy of Finland); *Patients, business and the state – Translating health information into sustainable benefits* (2012–2013, Tekes); *Global genes, local concerns* (2014–2017, University of Copenhagen's Excellence Programme for Interdisciplinary Research); SHOK SalWe *Personalized diagnostics and care* (2014–2018, Tekes); *Good(s) for health* (2015–2019, Academy of Finland); *Data-driven society in the making* (2018–2022, Academy of Finland); *Genes with meaning – Constructing national reference genomes for personalized medicine* (2019–2021, Kone Foundation), *Policy, practice*

and patient experience in the age of intensified data sourcing (2016–2021, European Research Council), and *Data literacy and responsible decision making* (2020–2024, Academy of Finland/Strategic Research Council).

4 Over the years in the above-mentioned projects, we have released over 40 publications on the topics addressed in this book, including two doctoral dissertations (Tupasela 2008; Tarkkala 2019). The variety of topics is wide: we have analysed and written about biobanking in general and in Finland, informed consent, privacy, ethical and political regulation, legal aspects, issues regarding the uses of health data, innovation policy, economic aspects and commercialization, biobank promotion campaigns and population branding, and the views, attitudes, and willingness to donate of the public and laypersons. Our publications form the groundwork of this book, and we have utilized the analysis and arguments presented in them when writing the chapters.

5 Perhaps the most famous human samples are, however, the HeLa cells that were collected in the USA in the 1950s from the particularly aggressive cancer of a woman called Henrietta Lacks, an African–American treated in Johns Hopkins Hospital in Baltimore. With this sample, it was possible for the first time to keep cells alive outside the human body in a cell culture. These 'immortal' cells have since became the most widely used human cell line in medical research. It became possible to distribute them from laboratory to laboratory, making them a limitless resource for researchers around the world (e.g. Skloot 2010; Landry et al. 2013). HeLa cells have even travelled to the moon and been involved in several medical breakthroughs. Although, in comparison with biobank samples, the HeLa cell line is a special case, as it is more of a tool used in the laboratories than a sample from a biobank donor in a freezer, both the biobank samples and the HeLa cell line nevertheless share the potential of human samples to become something relevant for science in the hands of researchers—either technically or based on the results. For the HeLa cell line, this has already become reality and quite exceptionally so. For a single biobank sample, it is more likely to be relevant and important as part of a bigger pool of samples analysed together and in relation to each other.

6 We have not discussed the European Health Data Space (EHDS) here in detail, as it is still in the making. The EHDS, as well as the 1+ Million Genomes initiative, has emerged in Finland simultaneously with already ongoing attempts to reorganize data uses. As both openings are relatively recent in the big picture of Genome Finland, we have not focused on these developments in this book.

7 There are arguments also for differences in lifestyles within the country, opposing the vision of shared lifestyle. For example, during the latter part of 20th century, epidemiologists were working in North-Karelia in

Finland, specifically introducing lifestyle changes to the whole population in the area to prevent coronary heart disease and cardiovascular morbidity that were more prevalent in eastern Finland than in western Finland and were seen to be associated with differences in lifestyles and especially in eating habits. On this topic, see, for example, Jauho (2021) and Kananen (2018).

8 Another question that has been discussed much internationally, but not as much in Finland, is about the rights and consent procedures related to children. During the first years of biobanking, sample collection concerned only the adult population. In 2016, some biobanks started to collect samples from children. While the issue has been discussed in Finland, the collection of biobank consents and samples from children started without large debate (for an analysis of a biobank campaign for recruiting children, see Snell and Tarkkala 2021).

9 In addition to reorienting the focus of KTL strategy to optimize its commercial activities, the strategy recommended the discontinuation of many existing activities, such as vaccine development. According to the proponents of the KTL strategy, this was necessary because private industry had developed more advanced methods to produce vaccines. The reorientation sought to streamline the operations and research focus of KTL.

10 Policymakers, governmental officials, and researchers involved in strategy work and related discussions adopted the term 'ecosystem', which became a catchword in Western innovation policy discourse and in management and business organization studies in the late 2000s (e.g. Oh et al. 2016). It refers to many kinds of relationships and arrangements between research institutions, private companies, public authorities, and other stakeholders that facilitate their collaboration in technological innovation, 'value creation', and business. During the past thirty years, interrelated concepts like 'cluster', 'innovation system', and 'innovation network' have been used in policy and scientific studies to refer to the collaborative and systemic character and dynamics of business related to technological innovations (e.g. Adner 2006; Oh et al. 2016; Lemola 2020, 115–122).

11 Taltioni was formed as a cooperative of 47 member organizations. Among them were IT firms like Fujitsu and Nokia, and major domestic private healthcare companies Mehiläinen, Terveystalo, and Diacor, as well as insurance company Pohjola. The public sector was involved through their 'development companies' Tiera and Medi-IT, the technology research institution VTT, and FIMM. The members of the cooperative wanted to develop electronic services for their customers through Taltioni. The idea was that individuals with personal data stored in Taltioni—a copy of personal patient data files, for example, or meas-

urements from well-being applications—could decide for themselves with whom they wanted to share the data and from whose services they wanted to benefit. It was also thought that eventually it would be possible to store personal genomic data and to personally decide on its uses.

12 See: https://healthtech.teknologiateollisuus.fi/fi/genomiteollisuus

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