

Mapping Jewish Identities: Migratory Histories and the Transnational Re-Framing of 'Ashkenazi BRCA Mutations' in the UK and Brazil

Jessica Mozersky and Sahra Gibbon

Much social science attention has been paid to the notion of 'genomic futures' and the implications of *medical* genetics for health, identity and how we conceptualize risk and prevention of disease for self and family (Finkler 2000, Hallowell 1999, Gibbon 2006, Mozersky 2013). More recently, others are directing us to the ways in which *population* genetics can reconfigure the past (El Haj 2007, Nelson 2008, Palmie 2007). Moreover, there is an increasingly complex intertwining of medical and population genetic research, with the former exploring what genetic variation may reveal about health, disease and drug response and the latter focusing on ancestral and migratory histories. It has also been suggested that genomic knowledge acts as a 'telescope' to the past (and future) in the ways that narratives of migration, colonization and origin have become part of the temporal logic of the medical, scientific and social discourse surrounding BRCA mutations (c.f. Adams et al 2009, El-Haj 2007, M'Charek 2013 Gibbon 2013, Schundler in this volume). This is particularly evident, as new technologies such as high-throughput sequencing are impacting the scope and reach of genomic interventions and as the marketing of direct-to-consumer (DTC) testing (particularly in the US) is combining a so-called 'recreational' interest in genetic genealogy with medical genetic testing and the promise of preventative health (Lee 2013, Koenig et al. 2008).

This chapter builds on studies at the emerging intersection of medical and population genetics by examining the different ways in which narratives of ancestry and migratory history have been, and are becoming, linked to mutations in the high-risk breast cancer genes BRCA1 and BRCA2. In particular, it examines how these historical narratives – initially associated with Ashkenazi Jewish communities and subsequently with diverse regions and populations – have become variously incorporated into different domains of transnational medical research and clinical practices. Drawing on ethnographic research in the UK and Brazil, this article compares the ways in which health professionals (research scientists and clinicians) communicate, understand and situate narratives of history and migration associated with Ashkenazi Jewish populations and the varied consequences this has for the way patients engage with and incorporate this knowledge into understandings of risk and identity.¹ In this way, we aim to highlight several contrasting dynamics surrounding the transnational re-framing of 'Ashkenazi BRCA mutations'.

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Before turning to our ethnographic data, we begin with a discussion of the ‘Ashkenazi’ BRCA founder mutations and their relationship to migratory and demographic history of both Jewish and, more recently, non-Jewish populations.

Founder Mutations as Links to the Past

The increased incidence of genetic disease among populations can be the result of processes known as the founder effect and genetic drift. A founder event occurs when a population has a small number of founding ancestors who are separated from the larger parent population and who goes on to found a new population (Stone et al. 2007). Alternatively, a founder event can be the result of an extreme reduction of a population, by over 50 per cent, due to famine, war, epidemic or some other event that causes a significant number of a population’s members to die. In both of these situations, the new founding population carries only a fraction of the original population’s genetic variation and is not representative of the entire diverse population from which it was derived (Stone et al. 2007). A founder effect occurs when one of the members of the new founding population carries a genetic mutation, which is then passed on to future generations, and the concept of genetic drift refers to this happening purely by chance. The mutation may substantially increase in frequency if the population remains reproductively closed – for example, due to geographic isolation or cultural/religious practices, such as endogamy (Janavicius 2010). Founder mutations can sometimes be traced to a single common ancestor or ‘founder’, and as such can create temporal links to a group’s ancestral past.²

The Ashkenazi BRCA Founder Mutations

The increased risk of BRCA breast cancer among Ashkenazi Jews is the most well-known and established population risk that has been identified to date, having emerged shortly after the cloning of the BRCA genes over 15 years ago. Ashkenazi Jews have the highest known population risk of carrying three specific founder mutations in the BRCA1/2 genes. The three ‘Ashkenazi mutations’, as they initially came to be known in medical and scientific literature, are illustrative examples of the ways in which founder mutations create temporal links to the past.³ Population geneticists have dated the Ashkenazi founder mutations and each has been correlated with a specific event and time point in Jewish history when the population was suddenly reduced and then re-expanded (Neuhausen et al. 1998, Risch et al. 2003, Slatkin 2004). For instance, the oldest mutation (185delAG on BRCA1) was originally estimated to be between 2,000 and 2,500 years old and believed to have originated in the Middle East prior to the first important founder event in Ashkenazi Jewish history when Jews left the Middle East and settled in Europe following the destruction of the Second Temple around 70 CE (Slatkin 2004). The second mutation (6174delT on BRCA2) was estimated to be about 700 years old and to have arisen in Eastern Europe, as most Jews settled there at this time. It is associated with a second important founder event – the severe persecution of the Jews during the Crusades and the Black Death, both of which led to an extreme population reduction (Slatkin 2004).⁴

1 The two authors carried out their fieldwork separately in the UK and Brazil. In the UK, Jessica Mozersky worked in clinical research for BRCA carriers (2002–2010) and carried out participant observation of cancer genetic appointments followed by in-depth interviews with high-risk Ashkenazi women over a 24-month period. The ethnographic research carried out by Sahra Gibbon in Brazil was based on 18 months of research in three different cancer genetic clinics in urban centres in the southern region of Brazil. This research included participant observation in cancer genetic clinics as well as interviews with patients and their families, health practitioners and scientists working in the arena of cancer genetics.

2 The same founder mutation can arise independently in different populations and is therefore not always linked to a single common ancestor.

3 Although we do not use quotation marks for Ashkenazi for aesthetic reasons here, as this article suggests, we take this term to be contingent and fluid.

While other populations are also the subject of genetic research, Ashkenazi Jews are considered to be ideal genetic research subjects because they are descended from a small number of founding ancestors who were confined to one particular geographical area (primarily Eastern Europe) and who practised endogamy. Furthermore, the Ashkenazi Jewish population was often extremely reduced as a result of famine, war or epidemic. At the same time, Jewish history is fairly well documented over the past 4,000 years in terms of migration, location and population events, making them a particularly attractive study population. Without such historical knowledge, it would be much more difficult to make conclusive genetic statements. When individuals are stripped of all prior information about their ancestry, geographical origins or ethnic group and are then assigned to groups *a posteriori*, racial categories or geographical origins based on genetic data become much less reliable (Bamshad et al. 2004). Without external information such as texts, oral history or archaeology, it is difficult, if not impossible, to tell a coherent story about a population based on genetic information alone (Goldstein 2008). Genetic differentiation and mutations are therefore a reflection of the particular geographic, migratory, reproductive and socio-cultural histories of populations.

In addition to the reasons stated above, much is known about genetic diseases among the Ashkenazi Jewish population because they have historically been active participants in research and screening programmes, and they are over represented in genetic literature (Birenbaum Carmeli 2004).⁵ For example, the identification of the first Ashkenazi founder mutation on BRCA1 was derived from a set of approximately 700 samples of Tay-Sachs disease that had been stored since the 1970s.⁶ Tay-Sachs screening brought thousands of Ashkenazi Jews into clinics, where their blood samples were not only screened for Tay-Sachs, but often stored for future genetic research. The ‘unexpected research payoff’ (Kahn 2005: 8) was the availability of stored blood samples that were subsequently used for other research studies. One consequence of the prevalence of the Ashkenazi Jewish population in research is that, when researchers find a particular mutation or disease risk among this group, the results are often labelled as ‘Jewish’ (as in the case of ‘Ashkenazi BRCA mutations’), even though the existence of these mutations in other populations who are studied less is entirely possible (c.f. Azoulay 2003). Perhaps unsurprisingly, as researchers have broadened their pool of subjects, Ashkenazi and other founder mutations are increasingly being identified in populations who do not identify themselves as Ashkenazi Jewish (c.f. Javanacisus 2010, Hamel et al. 2011, Weitzel et al. 2013).

Beyond Ashkenazi Jews

Despite increasing discoveries of the Ashkenazi mutations in other populations, these are often situated and discussed in scientific or popular discourse in ways that suggest that the presence of these mutations are diagnostic of ‘hidden’ or ‘original’ Jewish identity. For instance, in 2005 a scientific study by Weitzel et al. to determine the frequency of BRCA mutations among Hispanic American women living in Los Angeles found a high number of one of the Ashkenazi founder mutations in the sample. This led the authors to conclude that these ‘apparently non-Jewish’ families likely descended from the Spanish Jews known as *conversos* who hid their identities by converting to Christianity during periods of anti-Semitism in Spain from the twelfth to sixteenth centuries (Weitzel et al. 2005: 1669). Building on this study, Laitman et al. (2013) led an international genetic study that found the presence of this same ‘Ashkenazi founder mutation’ among a population of Indians living in the state of Colorado who had immigrated from Mexico, which was considered evidence that this mutation originated among the same group of *conversos* who had immigrated from Spain. This research gained international interest not only in scientific publications, but also in popular media in the US and abroad. For example, the Israeli newspaper *Haaretz* ran the headline: ‘Group of Colorado Indians have genetic Jewish roots’

4 The third founder mutation is discussed in a later section

5 See Mozersky (2013) for a detailed discussion of the many reasons – genetic and otherwise – why Ashkenazi Jews have become such prominent subjects of research.

6 Tay-Sachs is a severe and lethal recessive disorder that usually leads to death before the age of 5.

(Even 2012). According to this article, the study 'proves' that this group of Indians has 'genetic roots going back to the expulsion of Jews from Spain' (Even 2012), showing how such information is not just diagnostic of risk, but in this case indicative of migratory history and previously unknown Jewish identity. According to another article in the *Smithsonian* magazine, when one US oncologist was alerted to the discovery of this mutation among Hispanic Catholics, she reportedly said, 'Those people are Jewish' (Wheelwright 2008). Rather than raising doubts about whether the mutations are specific to Ashkenazi Jews, the researchers' focus on the migratory history of Jewish populations and their conclusion about the mutations' origin have reinforced the notion of 'Jewish genes' and common ancestry to all of those who carry them.⁷

According to Neulander's (2006) research in New Mexico with non-Jewish individuals who were found to carry supposedly 'Jewish' genetic diseases or mutations, genetic disease may be used to infer 'secret' or 'crypto-Jewish' descent from ancestral Jews. These claims often serve a broader purpose of asserting an overvalued line of white ancestral descent and restoring 'prestige lineage'. Neulander (2006) argues that the use of the label 'Jewish' to determine who is at risk of genetic disease has paved the way for the popular use of heritable diseases to determine who is a Jew. This 'disease-based Judaism' (Neulander 2006: 389) is in fact a failure to differentiate between heritable characteristics acquired through DNA and cultural characteristics widely attributed to Jews, which are only acquired through learning. Neulander (2006) argues strongly against genetically labelling faith-based communities, and instead proposes that diseases be identified in terms of geographical origins, such as Eastern Europe, rather than Ashkenazi Jewish, since every heritable disease will have a founder who can be located in time and place. As our paper suggests, in the context of transnational research and medical practices associated with the Ashkenazi BRCA founder mutations, there are signs that processes of reframing and renaming the Ashkenazi founder mutations are taking place, while Jewish origins are simultaneously still being inferred for those who carry them.

This paper contributes to these recent developments by exploring the diverse social, cultural and scientific implications involved when researchers find Ashkenazi BRCA mutations in apparently non-Jewish populations. The existence of Ashkenazi founder mutations among self-identified Ashkenazi Jews, Hispanic Californians, Brazilians and Mexican Indians is thought to be evidence of a common ancestor among all of them. How might this knowledge of a supposedly shared Jewish ancestry affect the ways genetic risk information is understood by health professionals and/or communicated at the clinical interface? How do individuals (self-identified as Jewish or not) conceive of themselves and the groups to which they belong? Some social scientists suggest that renewed attention in scientific and medical research to genetic ancestry raises profound questions for the configuration of identity (El Haj 2008, Palmie 2007). The discovery of genetic mutations or markers, including those not related to disease has the potential to strengthen or threaten a group's communal narrative, religious, cultural and even ethnic identity (Davis 2004).⁸ As a result, examining how new knowledge about the origin of the BRCA mutations intersects with pre-existing narratives of identity and history is central to understanding these developments. A key issue is the potential role of newly geneticized historical narratives of migration and origin for the 'naturalization' of preexisting identities as well as their partial or unexpected transformation

7 It might be suggested that Israeli newspapers have a particular socio-political interest in claiming Jewish origins for other populations. However, it is also evident that these scientific discoveries are being picked up in other national contexts as well, suggesting that this research generates interest in both scientific and popular domains.

8 For example, the Lemba are a South African tribe who claim to be descendants of one of the lost tribes of Israel. They have certain traditions, such as circumcision, dietary laws and rituals related to conversion, that are similar to those of Judaism. When researchers found that some of the Lemba carried the Cohen modal haplotype (a set of genetic markers found among some Jewish men who identify as Cohens), this was considered evidence that they were likely descended from Jews in ancient Israel, as their oral history suggests (Thomas et al. 2000). Cohen refers to a biblical priestly class of Jewish men who passed priesthood down from father to son. When another group in Ethiopia who also claim to be descended from Jews in ancient Israel were tested and found not to carry the haplotype, this was taken as proof that they were not Jewish (Azoulay 2003).

In the section below, we compare developments and scientific findings regarding the 'Ashkenazi BRCA mutations' in both the UK and Brazil, drawing on ethnographic fieldwork in each location. In the UK, research and clinical interventions regarding the Ashkenazi mutations have primarily been aimed at those who already identify themselves as Ashkenazi Jews. One result is that, in such a context, Ashkenazi mutations can have the effect of reiterating pre-existing or known Jewish identity and understandings of collective history. In Brazil, the discovery of Ashkenazi mutations among those who do not consider themselves to be Jewish can have diverse ramifications. For some, it might reveal previously unknown or hitherto hidden aspects of identity and history – the consequences of which can serve to both reaffirm pre-existing notions of being of 'European' origin and/or having mixed ancestry and as a result offering a confirmation of being 'Brazilian'. At the same time, an inferred Jewish origin can also be an uncomfortable scenario for Brazilian patients and practitioners, despite the positive symbolic value in Brazil of *mesticagem* – having mixed-race ancestry. These contrasting comparisons therefore reveal the multiple, context-specific ways in which genetic knowledge can affect notions of individual and group identity as well as the boundaries, moral frameworks and affective alignments through which genes, risk and identity are variously co-configured.

The UK Context: Reiterating Jewish Identity

In the UK, genetic testing is available free of charge through the National Health Service to any individual who meets the risk criteria set by the National Institute for Health and Clinical Excellence (NICE). In 2005, NICE published revised guidelines for the management of familial breast cancer, including the addition of Ashkenazi Jewish ancestry as an important risk factor to be taken into account by physicians when considering whether to refer a woman to genetic counselling services.

It is standard clinical practice to send women who are referred to genetic counselling a family history questionnaire in advance of their appointment. This questionnaire includes a question about Jewish ancestry for the purposes of determining an individual's risk. Women who self-identify as Ashkenazi and opt to undergo genetic testing are initially only tested for the three Ashkenazi BRCA founder mutations (as opposed to sequencing the entire large BRCA1 and 2 genes). Screening only for specific founder mutations is a cheaper, faster way to carry out genetic testing, as most Ashkenazi Jews who are tested will be found to carry one of the three founder mutations. Sequencing the entire BRCA1/2 genes (a more expensive and time-consuming process) only occurs if no mutation is found after this initial round of targeted testing. There is thus a very practical benefit for clinicians and scientists to be had from knowing whether particular founder mutations exist within a given population (Javanicius 2010).

In the UK, such clinical practices, alongside national guidelines and a large body of scientific research about this population, have had the effect of routinizing the clinical association of Ashkenazi ancestry with genetic breast cancer risk.⁹ In fact, genetic counsellors have reported a lower threshold for offering genetic testing to Ashkenazi patients (for example, one first-degree relative with breast/ovarian cancer) as opposed to the more stringent criteria used with other patients. This clinical practice reflects both the fact that testing for founder mutations is faster and more economical and the perception that being Ashkenazi Jewish is strongly associated with an increased risk of genetic breast cancer.

Mozersky (2013) has shown elsewhere how genomic knowledge about breast cancer risk can reiterate pre-existing ways of how Ashkenazi Jews understand their group belonging and collective history. For instance, the existence of Ashkenazi BRCA mutations is interpreted by some individuals as evidence of the interrelatedness of

⁹ It is worth noting that even this clinical association is contextual and not prevalent in all of Europe. For example, in France the universalist conception of citizenship does not favour focusing on ethnic issues in general or in relation to breast cancer. Thus, ethnicity and Ashkenazi Jewish origin are not significantly associated with breast cancer (Lowy and Gaudilliere 2008). In Germany, clinicians avoid discussion about disease risk and Ashkenazi or 'racial/ethnic' identity as a result of the contentious German past for Ashkenazi Jews during the Holocaust (see zur Nieden in this volume).

all Jews, their common ancestors or a collective history (including suffering) in the ghettos of Eastern Europe. The examples below illustrate this reiteration from the perspective of patient advocacy organizations, clinicians/researchers and patients.

Patient Advocacy

Jews Against Cancer of the Breast (JACOB) International is an international breast cancer advocacy organization dedicated to helping Ashkenazi Jewish women learn more about their increased risk of genetic breast cancer. Their goal is not only to educate Ashkenazi women, but to encourage all women (not just those at high risk) to seek genetic testing in order to fulfil their mission of breaking the cycle of hereditary breast and ovarian cancer. JACOB's website explains the Ashkenazi mutations as follows:

It is believed that these mutations can be traced back hundreds of years to their common ancestors, or founders. As the result of numerous intermarriages among Jews, all of today's Ashkenazi Jews are descended from a very small group of Jews who lived in Eastern Europe 500 years ago. These 'founding Ashkenazis' carried the particular BRCA1 and BRCA2 gene mutations which were subsequently passed on to their descendants.

(JACOB International 2013)

JACOB explicitly relates these mutations to the common history of Ashkenazi Jews in Eastern Europe. The US Jewish advocacy organization Sharsheret (Hebrew for 'chain') supports young Jewish women and their families who are facing breast cancer and acknowledges the 'unique concerns' of Jewish women, particularly by putting them in contact with one another and offering various support programmes. The cover of their education booklet about genetic breast cancer, entitled 'Your Jewish Genes', has a photo of four women of all different ages and generations and contains an explanation of '*what's Jewish* about hereditary breast and ovarian cancer' (Sharsheret website 2013, italics added). This further demonstrates how genetic breast cancer is framed as a Jewish disease caused by supposedly Jewish genes.

Donelle et al. (2005) have compared the portrayal of breast cancer risk in Jewish newspapers as opposed to newspapers intended for the general population and found that genetic breast cancer is especially associated with being a Jewish woman. They found a significantly higher proportion of articles that identified genetics as the main risk factor for breast cancer in Jewish newspapers as compared to non-Jewish newspapers (75 per cent of the articles in Jewish newspapers versus 12 per cent in general newspapers). Despite the fact that a very small proportion of breast cancer is genetic, even among Ashkenazi Jews, the Jewish newspaper articles 'insinuate that the identity of Jewish women, in part, entails the genetic risk of breast cancer' (Donelle et al. 2005: 191).

Clinical and Research Context

In the UK and several other national contexts, population-wide screening programmes are being piloted with the aim of screening all Ashkenazi Jews, regardless of family history, for the three BRCA founder mutations (Levy-Lahad et al. 2012, Metcalfe et al. 2009). In the UK, a pilot population screening programme began in October 2008. It aims to recruit 10,000 Ashkenazi Jews and 'to develop a strategy for prediction and prevention of genetic cancer. Over the long term it is hoped this will reduce cancers in the community.'¹⁰ The study is supported by a major Jewish charity in the UK, the liberal and reform Jewish movements, rabbis, other Jewish organizations and a major high street chemist. Similar to the UK, it was announced in May 2008 that Jewish women in Ontario, Canada, would be offered a free genetic test for BRCA mutations as part of a research study.

10 <http://www.instituteforwomenshealth.ucl.ac.uk/academic_research/gynaecologicalcancer/gcrc/gcapps> (accessed 20 January 2012).

Within ten days after a national Canadian newspaper began advertising the study, over 2,100 women volunteered, illustrating 'considerable interest for genetic testing among Jewish women' (Metcalf et al. 2009: 1). This study found that 45 per cent of the mutations identified were in women who did not meet the local genetic testing criteria, leading the study authors to conclude that genetic testing should be extended to women who do not meet the current criteria (Metcalf et al. 2009). One female Canadian rabbi claimed that BRCA mutation screening will one day be offered in a similar way to Tay-Sachs screening because the 'rate of this gene is so high' (Priest 2008). In Israel, a general population screening study found mutations in 63 per cent of families that would not have otherwise been considered high risk due to minimal or no family history of breast or ovarian cancer, leading to the suggestion that population-wide screening of the Ashkenazi population is feasible, cost-effective and justified (Levy-Lahad et al. 2012).

These studies demonstrate how researchers stratify samples based on notions of a discrete Ashkenazi population with a unique risk of BRCA mutations, which in turn justifies population-wide screening. At the same time, self-identified Ashkenazi Jews actively participate in research, which has the effect of reinforcing notions of specific 'Ashkenazi' mutations and genetic diseases.

BRCA mutations are also portrayed as indicative of a collective Jewish history and identity because they have been dated and correlated to particular historical events. In addition to the two founder mutations described earlier, the third mutation (5382insC on BRCA1) is estimated to be approximately 1,800 years old but to have entered the Ashkenazi population about 400–500 years ago (Hamel et al. 2011). According to Hamel et al., the mutation may have spread due to the rapid expansion of Jews in Poland beginning in the sixteenth century, which 'significantly improved the odds of admixture' (2011: 305) between Jews and non-Jews, even for an otherwise relatively genetically isolated group (admixture refers to reproduction across groups). In contrast, the origins of this mutation have also been associated with another Jewish founder event: the Cossack massacres in 1648, which resulted in the 'total destruction of many Jewish communities' and the death of at least 25 per cent of the population (Risch et al. 1995: 157). According to one British Jewish breast surgeon, this mutation may have spread as a result of rapes during pogroms, leading him to describe it as 'another tragic event in Jewish history' and a 'component in the repertoire of humiliations experienced in the Jewish ghettos of the Pale of Settlement between the 13th and 19th centuries' (report of the Anglo-Israeli Workshop on the Genetic Risk of Breast Cancer 2006: 4). Of course, rape is not the only mechanism by which genetic mutations can be transferred across groups, as Hamel et al.'s (2011) more neutral suggestion of admixture suggests. The surgeon's comment rather demonstrates how specific aspects of the historical suffering of Ashkenazi Jews are invoked to explain hereditary disease-causing mutations.

Inferring Jewish origins from genetic BRCA mutations has also been reflected in clinical practice, as when one genetic counsellor has reported that occasionally an 'Ashkenazi mutation' is found in families who do not identify as Jewish. When asked whether this actually indicates that a family is really Jewish, given that some of these mutations are present in non-Jewish Eastern European individuals, she explained that it just shows how 'somewhere back there genes got mixed in' and that she usually has chosen not to inform the family that the mutation was specifically a Jewish one, presumably to avoid altering how this family/individual already conceives of themselves.

Women

Although most high-risk Ashkenazi women in the study were not aware of the specific Ashkenazi founder mutations and the endeavour of population geneticists to date each of them, they did attribute the origins of genetic disease and mutations to the collective endogamous history, and suffering, of Ashkenazi Jews in the shtetls of Eastern Europe. For instance, Jennifer was a British woman in her forties who had a very strong family history of breast cancer. She was awaiting her genetics appointment and explained her understanding of the increased risk as follows:

I know the history, you know, was that we were in tiny shtetls so I guess it makes sense, um, and it's just one of those things... we've all seen *Fiddler On The Roof*, so I guess you know it does make sense, they don't marry that far out really do they? It's different today I suppose there's a lot more, well for a start we're not living in tiny little shtetls. We live in big cities and we're just sort of out there much more, and everything has changed beyond measure what our great grandparents would have imagined, um, so I suppose it's probably reducing as well now, maybe... even if you were still marrying a Jewish person, it wouldn't necessarily be from your village, it could be from Birmingham or you know whatever, so that the kind of risk, that kind of intermarriage, is probably reducing now.

For Jennifer, life in the shtetls of Eastern Europe and the intermarriage between Jews helps explain the risk of genetic breast cancer. While this history 'makes sense' and is consistent with her understanding of Jewish history, she relegates such practices to the past. Jennifer's first cousin Anna was in her mid-forties and had lost her mother (Jennifer's aunt) to breast cancer when she was in her twenties. She was awaiting the results of her BRCA mutation test. Anna also related the increased risk to historical reproductive patterns:

But it's interesting to understand this background because... presumably it's all through intermarriage. It's sort of fascinating... well it must be cousins, I presume it's coming through cousins, I suppose we have no idea of what background, I mean what sort of communities they lived in and one assumes it was an acceptable way of life in being introduced to what must be your first or your second cousin.

The existence of genetic breast cancer among Ashkenazi Jews has the potential to reiterate a collective historical narrative of Jews living in Eastern Europe, isolated in small shtetls and ghettos and practising endogamy. In this context, genomic knowledge does not transform or alter understandings of the past; rather, it serves to naturalize a social/cultural narrative about a shared past.

The passages from interviews below demonstrate how genetic disease can reiterate a sense of group belonging and boundaries based on blood, genes and biology. Lori was an American graduate student living and studying in London. She was in her mid-twenties and had lost her mother to breast cancer as a teenager. Although Lori was too young for genetic testing at the time of the interview, she was advised that it was something she might consider in future. Although Lori was not raised in a religious home – she described herself as secular and her Judaism as a 'cultural background' – she also said: 'I think that these potential genetic mutations, Tay-Sachs disease, breast and ovarian cancer mutations, I mean that's what brings it back to a biological level obviously.'

For Daniel, an unmarried, successful, retired businessman in his late forties, genetic disease among Ashkenazi Jews helped to define them as a distinct group:

I would have thought that if there are distinctive Jewish diseases then there must be distinctive Jewish genetic components. Now it may not be enough to found a whole identity, but... it makes it quite difficult to resist the conclusion that if one was trying to define a group, meaning draw boundaries between it and another group, even if they weren't clear boundaries... still it's an indicator!

Despite Daniel's acknowledgement that the boundaries would be unclear and porous when trying to define the group, it is hard for him to resist the conclusion that genetic mutations are an indicator that there are genetic aspects to being Jewish, and that this serves to reinforce the boundaries between Jews and non-Jews. For Lori and Daniel, disease-causing mutations, or 'genetic components', demarcate Jews in a certain way and reiterate a boundary between Jews and non-Jews. This 'disease-based Judaism' (Neulander 2006) demonstrates that the existence of supposedly Jewish genetic diseases reinforces the pre-existing boundaries and ways in which

individual 'Jewish' identity has been understood. Thus, this way of thinking about Jewish belonging did not challenge or conflict with the other social and cultural ways in which individuals also experienced being Jewish.

The Brazilian Context

The field of BRCA research in Brazil has been active since the early 2000s, when the 'unknown' contribution of BRCA genes to breast cancer incidence in the country became a source and site of inquiry and transnational research, contributing to the emergence of Brazilian *oncogenetica* as a clinical specialty (Gibbon 2013). From an early stage, there has been an explicit focus on identifying the frequency of possible founder mutations in the Brazilian population.

Initially, this included mutations identified in populations outside of Brazil, including those associated with the Ashkenazi Jewish population. The focus on these specific mutations was initially directly tied to the possibility of developing 'rapid inexpensive genetic tests' in the Brazilian context (Gomes et al. 2007) – an issue of considerable importance in developing cancer genetics in low-income country settings (Narod 2009).¹¹ A number of Brazilian studies have subsequently identified these mutations at variable but 'significant' frequencies in different regions of the country (Lourenco et al. 2004, Dufloth et al. 2005, Esteves 2009, Ewald et al. 2011, Costa 2008). Initially, this research suggested that 'a small number of founder mutations may be predominant' and called for 'screening for all Brazilian women with breast cancer' (Gomes et al. 2007).

The focus on these mutations in Brazil reflects an international research agenda for the BRCA genes in which the Ashkenazi mutations are some of the most common identified worldwide. Nonetheless, given that nearly all of the Brazilian studies have been undertaken, entirely or in part, with individuals who do not identify themselves as Jewish, there has unsurprisingly been significant speculation in these published papers about the reason for finding these mutations in the Brazilian population. Common narrative explanations in published papers include the reference to the large Jewish population who used to live in Portugal and or Spain but who were deported during the Inquisition known as *conversos* (Gomes et al. 2007, Ewald et al. 2011). By contrast a study by Costa et al. in 2008 of the 'origin' and 'diffusion' of one so-called Ashkenazi mutation (5382insC) highlights the potential for a 'significant Eastern European contribution to the present day genetic background of the Brazilian population, contrary to an alternative hypothesis of a contribution from new Christians emigrating in the 16th century' (2008: 65). It is argued that this reflects the specificity of Brazilian migratory history, especially as this concerns German immigration to the region in the late nineteenth and early twentieth century.¹²

In a more recent article, Ewald et al. (2011) also turn their attention to the range of different founder mutations that they have identified in non-Ashkenazi women in their publications, including historical explanations for the identification of these mutations in Brazil. While acknowledging that a more detailed understanding of their entrance and distribution in Brazil remains to be determined, the article nevertheless counters prior speculation that those who carry the mutation may be *conversos* from Portugal. Instead they suggest that the more likely explanation is that the 'mutation probably originated in Scandinavia or Northern Russia...and could have entered Brazil through one or more several large European immigration waves in the 18th and 19th century' (Ewald et al. 2011: 5).

There is thus an understanding 'that the population from the Brazilian cities studied here show trihybrid ancestries that are distinct from Central American populations' (Ewald et al. 2011: 6).¹³ In the above

11 However, the development of NextGeneration sequencing and cheaper panel tests may, in the future, significantly change the parameters of the situation in the context of emerging and developing economy contexts such as Brazil as well as elsewhere.

12 While other South American countries have historically been associated with colonization from Spain, the southern part of Brazil, and to a large extent Argentina and Uruguay, have been associated with more recent immigration from other parts of Europe.

publication, such an acknowledgement becomes the broader context in which a position *against* population screening for the Ashkenazi founder mutations is made, as this would mean ‘more than 90% of mutation carriers would remain unidentified’ (2011: 6).¹⁴

We can therefore see how, in contrast to previous publications, instead of attempting to associate the presence of so-called Ashkenazi mutations with a particular national origin or specific migratory histories, such research contributes to knowing and making evident the ‘mixed’ ancestry of the Brazil population. This (re)framing of the meaning of Ashkenazi mutations is strongly consistent with long-standing notions of Brazil as a place of mixture, or *mesticagem*, which has been historically positively associated with Brazilian national identity. As emerging social science research in Brazil suggests, an emphasis on genetic mixture is being reconstituted through different domains of genetic research, conjoined to powerful, effective and moral alignments of national identity and nationhood (Ventura Santos and Maio 2006, Santos and Silva 2011, Gibbon 2013).

A brief overview of publications in the Brazilian context therefore illustrates the ways in which scientific and medical research associated with the Ashkenazi mutations is both shifting away from this association while it is also being incorporated into Brazilian cancer genetics. Nevertheless, observations and interviews with health practitioners in Brazil show how this continues to generate complexities at the clinical interface and ambivalence about the meaning, significance and utility of attending to certain categories of population difference through a focus on Ashkenazi mutations.

Practitioners

While the Brazilian census categories (black, brown, white, yellow and indigenous) were sometimes filled in on the family history forms used by practitioners in consultations I witnessed, questions surrounding *etnia*, or the more common term *raca*, were not commonly raised in clinical consultations as a matter of course. When they were discussed, often little or no information was given by patients beyond stating that they were ‘Brazilian’. In the southern city of Porto Alegre, discussions of family origin sometimes elicited further details, especially when it was obvious from a surname or during the consultation that a person had German, Eastern European or Italian ancestry. But there was little explicit discussion or even direct questioning of whether someone had Jewish ancestry. When I commented on this with one cancer geneticist in Porto Alegre, he explained this reticence in ways that also reflected the relevance and perhaps also moral worth of emphasizing and recognizing ‘mixture’:

I think there is a bit of fear about suggesting to the patient that perhaps you have a Jewish origin. I think we’re a bit different in that respect because we’re so mixed here we haven’t got ‘oh so you’re only Jewish’ – it’s difficult to find that in Brazil. You’ve got your African part and your Indigenous and European, but I think that helps too to have less prejudice.

Another scientist who worked in the field of genetic epidemiology in clinical research projects associated with the BRCA genes expressed similar sentiments about the difficulties of categorizing risk in relation to unique population groups.

We’re so mixed here I think it’s difficult to find a unique influence, so we really don’t worry about this. We just don’t have this, the first time that I heard about the studies with specific Ashkenazi models I didn’t

13 They add here ‘additionally it has been demonstrated that even Mestizos from different Central and South American regions have intra-and inter-ethnic variability and admixture profile’ (Ewald et al. 2011: 6).

14 The authors also note it is a situation that they suggest is exacerbated by ‘lack of coverage through both private and public health based insurance of germline mutation testing’ (2011: 6), showing how such research is also linked to calls for expanded public health provision for cancer genetic services.

know what it was but then saw that there are lots of studies with this population so for instance the ‘Gail’ models¹⁵ with the race component if you are African or white. But it’s really difficult to do that here. When we use the model here to estimate risk, we never know what to calculate. We have in various moments opted to put all the population as white, because at least we can compare one woman with another.

There was also a sense among practitioners that patients in clinics often ‘didn’t know’ whether they had Jewish ancestry. As one researcher working with the cancer genetics team put it: ‘You could ask the patient directly – do you have Jewish ancestry? Then she’d reply, “No, no, as far as I know we don’t have,” but then you find out that she does’. A number of clinicians and researchers talked about the rapidly changing field of identifying BRCA mutations inside and outside of Brazil, which has raised questions about the extent to which certain mutations could still be described as exclusively associated with Ashkenazi Jewish populations. This was how one practitioner put it:

For example, that mutation that the team here found, that we thought was associated with the Ashkenazi population from the literature, now there are lots of articles that say there were possibly two entry points for the mutation. Perhaps it was a characteristic of the Ashkenazi Jewish population but perhaps it was from another that had different mutations. So imagine if you say to a patient that it was associated with Ashkenazi ancestry and two or three years later an article is published saying no it’s not – it gets complicated. So in science if it’s not something certain I don’t think we should be saying anything, otherwise we could cause more confusion. Of course, if they are really Ashkenazi and say they are, fine but if they’re not Ashkenazi then you’re going to be thinking, ‘Is it that they have an Ashkenazi origin or just that they don’t know?’ You might try to speak to them to ask them to see if they can discover something but afterwards you might find that that mutation isn’t associated in that way and it gets complicated.

Another professional reflecting on the ‘origin’ of these mutations in Brazil also underlined, as other international publications have recently suggested, that it might not be appropriate in Brazil to describe what was identified as Ashkenazi mutations or to use a stated Ashkenazi ancestry as a criteria for testing, as has been done in other countries.

You know those families that we have here who have Ashkenazi mutations –with rare exceptions they don’t recognize themselves as Jewish...but perhaps these mutations are very old. I think they could be Christian converts who centuries before were Jewish, and today culturally aren’t...so I think in Brazil we can’t call them ‘Ashkenazi mutations’ and as a result I’m a little bit reluctant to use this idea as an indication to test or not.

Practitioners’ comments reveal how a variety of concerns and issues emerge when considering the utility and value of Ashkenazi BRCA mutations in Brazilian cancer genetics. This includes the economic logistics of being able to undertake genetic testing in resource-poor contexts with the complex cultural histories of race, ancestry and ‘mixture’ in Brazil and perhaps most importantly with the inapplicability of population categories developed elsewhere for informing the significance and meaning of finding Ashkenazi mutations in non-Ashkenazi-identified persons and families in Brazil. The following sections reflect on how two Brazilian families, neither of

15 The Gail model is a statistical model for calculating the risk of breast cancer that incorporates age, reproductive history, breast biopos history and breast cancer incidence in first-degree relatives. Other models, such as BRCAPRO, incorporate Ashkenazi Jewish heritage.

whom identified themselves as Jewish, responded to the information given to them by practitioners that the identified BRCA mutations were associated with this population.

Elenice and Angela

Elenice and her sister Angela attended a consultation in the mixed private/public hospital in Sao Paulo. They had travelled several hundreds of miles from the neighbouring state of Parana for a follow-up clinical appointment to receive the results of the BRCA test they had (in this case) paid to have done. Both the sisters were in their mid-to-late-40s, had each been diagnosed with cancer ten years earlier and had many cases of breast cancer in the family.

During the consultation, they were told that an initial mutation test had positively identified a specific BRCA mutation. They were in the consultation with five of their daughters, all of whom were in their 20s or late teens, to discuss the possibility of genetic testing for them.

Before discussing the result that she'd received that morning, Elenice talked about what she thought had caused her cancer, pointing out that, although genetic factors were significant, she believed that the interaction of other factors with genetic aspects were important. For Elenice, 'external factors' such as 'smoking and stress' were, she said, 'together in this'. In other words, in her view genetic factors were necessary but not sufficient to explain the incidence of cancer in her family.

Responding to an open question at the start of the interview about how she would describe her ethnicity or race she immediately asked if this meant 'ancestry'.¹⁶ This being affirmed, she went on to say 'the ancestry well what we discovered in all this process is that the genetic mutation that we have amongst us came from the Jews so we discovered that we have Jewish ancestry'.

She pointed out that she had a brother who had investigated this also and discovered Jewish ancestry on both sides of the family. When asked directly if they were Jewish, Elenice immediately said, 'No, no, we're Brazilian. I might have a [Jewish] genetic characteristic but I'm Brazilian'. Probing this response further and asking what being Brazilian meant to, her she added, 'I think that being Brazilian means being Indian and I'm more on that side.' Toward the end of the interview, she described in more general terms how the information that they had received today about the test result would affect how she saw her family history or the identity of the family. She was hesitant and uncertain, pointing out that this was still 'new information', but said, 'I think there will be changes, yes...internal...personal, psychological perhaps.'

Elenice's younger sister Angela also directly responded to the initial question about ethnicity at the start of the interview with information about ancestry and cancer risk, saying

We have a little bit of Italian with Portuguese but my oncologist was saying that there is a very rare cancer that was detected in Curitiba that was discovered in 1800 linked to the Portuguese so Elenice discovered just a while ago we have Portuguese in the family...we never worried about this but now we have to worry about this, go back in the family.

For Angela the information they'd received in the consultation about having a mutation associated with particular ancestry seemed in this case to have been linked to other information they had been given by a different oncologist.¹⁷ This was even more evident when Angela pointed out how their current surname 'was one of the names the Jews used to change their name that really isn't Portuguese its Jewish'. Unlike Elenice, who saw the causes of cancer in her family as a mixture of factors, for Angela genetic aspects seemed to dominate, as she

¹⁶ This question forms part of the collection of more socio-demographic data collected at the start of the interviews with those attending the cancer genetic clinics.

said: 'It's all in the blood [*tudo esta no sangue*] – not just disease but everything, isn't it – something that you carry for ever'. At the same time, when asked what this meant in relation to the ancestry she had recently discovered in the family associated with the risk of cancer, it was clear that a notion of Brazil as a place of population mixture also informed this understanding of at-risk ancestry. As she said, talking of Brazil, 'There isn't just one ethnicity or population, there are unions [*unioes*]. I think that one union of one ethnicity with another creates this [risk]...because [here] the mixture is very big.'

For both these sisters, the information that they'd been given in the clinic about the BRCA mutation that had been identified in the family as being associated with Jewish ancestry was contextualized in a variety of ways with differing repercussions. While for Elenice, genetic risk factors were nested in a complex mix of other causal aspects, it was clear that there might be consequences associated with knowing this risk information in terms of 'identity' (for her, read through an idiom of 'internal' psychological factors) for the family. Yet, like Angela she also situated information about genetic ancestry associated with the BRCA genes in the broader context of Brazil being a place of population mixture such that, at least for Angela, it was hard to think of 'just one ethnicity', even if the explanatory weight of genetic knowledge was for her a powerful one.

Yvonete and Viviane

Yvonete was in her mid-20s and worked in a pharmacy in the city of Porto Alegre in the southernmost state of Rio Grande do Sul. She had been attending the cancer genetic service of a government-funded SUS hospital there for the last few years and had recently heard that she was (like her mother who had had breast cancer a number of years before) carrying a BRCA mutation, although she herself had not developed the disease.

Initially Yvonete mentioned nothing about the information she had been given about the mutation, which had been associated with the Ashkenazi Jewish population. This emerged more directly during a discussion about how she would describe her ethnicity. She initially described herself as being 'half German and half Italian...and all the Brazilian outside influence as well...so I'm really well mixed'. When asked if she associated ethnicity or ancestry in any way with an increased risk of developing the disease, the subsequent exchange followed:

Yvonete: I think yes...in fact the geneticist said something to my mum about this gene that they have been studying appearing in a Jewish community that existed...that all the women had it...all of them had cancer. So it appeared initially that it was obvious, but my German grandfather nearly fainted, you know saying that it had come from the Jews!!

SG: So you are not Jewish then?

Yvonete: No! No, well, you see my grandfather is German, so we started teasing him 'you know you have Jewish ancestry'...but it looks like it started in that Jewish community. But really I've never stopped to think about this very much...well I suppose it's interesting that its with us now and we don't know how...or we'll never know...But what we have to think about at the moment is how not to pass it on right? If there existed that possibility, that's what I would like to know now.

Yvonete's mother Viviane later also talked about her ancestry as a 'mixture' of 'German, with Brazilian and Portuguese' while also emphasizing her German ancestry. But when asked if she associated ancestry as a risk factor for the disease she was more hesitant, saying:

17 There is an association in Brazilian oncogenetics with a different mutation R337h also linked to a high incidence of cancer in the southern region of Brazil and in research publications has been associated with Portuguese ancestry (see Gibbon 2013). Angela seemed to have mixed these narratives in discussing issues related to ancestry.

I've never thought about this in a direct way. I've heard people speaking about this...but now that I'm talking I remember that the doctor mentioned this to me that some races [*racas*] have more diseases or types of cancer. I'd never heard this before, never thought about it. I guess those who have lighter skin have more skin cancer in that sense...but in relation to my history it could be but I wouldn't know to say for sure.

While this response suggests that she recalled something about the association with Jewish ancestry that her daughter also remembered, Viviane does not directly mention this link. More explicitly, later on when asked directly whether the test result had affected the way she thought about her ethnicity or identity, she said emphatically and explicitly, 'No, it's got nothing to do with this.'

Thus, while both mother and daughter remembered in part some of the information they had been given – that the identified mutation was associated with Jewish Ashkenazi ancestry – this was in both cases nested in a sense of having mixed European and 'Brazilian' ancestry. While Viviane explicitly rejected the relevance of that information to the family's identity, Yvonete suggested that it initially had repercussions in the family, given the family's sense of having particular 'German' origins.¹⁸ Yvonete, nevertheless, like many others, seemed less concerned with recent or older family or ancestral origins than with making future preventative histories for herself and her children.

Conclusion

Through a comparison of ethnographic research in the UK and Brazil, this chapter has examined how changing scientific and medical understandings regarding the origin, genealogical history and patrimony of the so-called Ashkenazi mutations have been diversely taken up and put to use in clinical/research contexts. It has explored the very differing consequences this can have for the way health care practitioners, scientists, patients and their families engage with and incorporate knowledge about hereditary BRCA mutations into scientific narratives, clinical practices and understandings of clinical/familial risk and identity.

We have shown how the recently discovered presence of Ashkenazi mutations in non-self identified Jewish individuals from diverse geographical regions reveals the complexities and fluid boundaries of categories such as population, race and ethnicity. In particular, it reveals the complexity and incommensurability of translating scientific findings and categories originating in European/North American contexts to other diverse regions.

At the same time, genomic knowledge is temporally configured by bringing narratives of migration, ancestry and colonization into view alongside the medical, scientific and social discourse surrounding the origins of the BRCA mutations (cf. Adams et al. 2009, M'Charek 2013, Gibbon 2013). In this sense, BRCA founder mutations have the ability to constitute not only promissory futures but also to de/re/construct or 'abduct' genomic pasts (Adams et al. 2009, Palmie 2007), 'telescoping' both the past and future into the lived present (see Schlünder in this volume).

While several researchers examining the turn to ancestry and history in the wider field of genomics, particularly in relation to commercial ancestry testing, have warned about the potential in these developments to lend scientific legitimacy at the molecular level to 'unscientific' constructions of the past (Palmie 2007, Abu El Haj 2008), others point at different dynamics, where the 'strategic' selection and use of genetic genealogy and

18 This region of the southern part of Brazil is historically associated with German populations who themselves have a complex history in Brazil. As a group, German immigrants to Brazil at the turn of the twentieth century have a history of being culturally suppressed but are also more recently celebrated as having a distinctive history within southern Brazil. In this region there is a popular association between having German ancestry and conferring to specific phenotypic features, such as blue eyes and a 'whiter' skin tone. This is set against a contrasting broader background of 'mixture', which is valorized as being more Brazilian.

ancestry is complexly unfolding. Nelson (2008) has shown that there is considerable variance in how individuals interpret genetic ancestry information. That is, scientific data is not always accepted as definitive proof of identity, but is rather often interpreted within the context of personal experience and the historical politics of identity.

Such variation in the significance and meaning of genetic ancestry information is particularly notable in the examples discussed in this paper that reveal the effective and moral ramifications of the transnational reframing of Ashkenazi mutations, which move far beyond a simple telescoping of past and present. In Brazil, the disjuncture in alignments between Ashkenazi mutations and being Brazilian is evident in the hesitancy and ambivalence of the cancer genetic practitioners, as they negotiate newly emerging understandings and gaps in epidemiological knowledge associated with the BRCA genes. Here, a focus on Ashkenazi mutations facilitates transnational research and a reflection on migratory histories while it can also be used to valorize and make material 'hybrid' ancestries. At the same time, we see how knowledge of the mutations associated with Jewish populations and history is diversely incorporated within an understandings of being 'Brazilian' as being of mixed ancestry by families in the cancer genetic clinics, where newly discovered specificity and solidity of family history generates discomfort or ambivalence.

In contrast, for Ashkenazi Jews in the UK, genetic information about BRCA mutations and Jewish history are consistent with the pre-existing ways in which individuals self-identify. While such knowledge may also be nested in other understandings of risk (such as diet or environment), genetic research and medical practices reiterate narratives of shared ancestry and collective belonging as Ashkenazi Jews. On the one hand, it is plausible that, for historical reasons (such as the Holocaust), genetic research and discoveries might be met with scepticism or anxiety regarding 'biological' thinking about Ashkenazi Jews. In the UK context, however, this appears not to be the case for the majority of individuals. Knowledge about genetic mutations does not necessarily lead to ambivalence or a reconfiguration of identity, but rather allows individuals to situate genetic disease within a complex sense of self and community that includes shared history, ancestry and belonging.

As BRCA testing continues to expand and to be undertaken in diverse global arenas it will become increasingly important to monitor the extent to which we may see further reclassifications of the so-called Ashkenazi and other founder mutations and/or the way that historical narratives of colonization and migration of other populations are (or are not) used in the explanatory scientific and medical discourses associated with these new understandings. As the examples in this paper suggest, the socio-cultural consequences of inferred Jewish origins from genetic research can be highly variable. While for some, this may be unproblematic or even culturally valued, for others such information may sit uneasily with pre-existing national or regional histories, creating an unsought and sometimes undesired solidity of what are in fact heterogeneous and fluid identities and modes of identification. Equally, we may see a more cautious approach to categorization, or a redefinition, of categories with reference to continental ancestry or geography (Neulander 2009, Fujimura and Rajagoplan 2009) that could help constitute new spaces of identity and identification. At the same time, practical issues such as cost and available resources may continue to encourage researchers to look for founder mutations, or other population-wide risk factors, that may help ease the economic burden that incorporating genomics into health care raises for many countries.

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