Contextualizing the Representations of Finland as a Population Isolate

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Abstract *The Finnish population is often represented in the scientific and medical literature as isolated and homogeneous. According to the proponents of this view, these characteristics resulted from bottleneck effect dating back some 4,000 years. Many scientific papers and journals have visually represented this asserted distinctiveness in maps or pictures, as have websites portraying the results of studies arguing for this thesis and depicting the country's population history from its perspective. In this chapter, we discuss these visual representations through three analytical lenses that each provide their own view on the narrative of population homogeneity and isolation. First, we will highlight the specific background against which the understanding of an isolated population emerged as part of wider research project on certain genetically enriched rare diseases understood to be particularly Finnish. Second, we will discuss the way Finns have been portrayed as outliers by discussing visual representations of Finns in the medical and scientific research literature. Third, we will discuss the characteristics of this supposedly isolated population that in fact indicate an inner heterogeneity, more likely pointing toward two (multiple) populations of Finns rather than one, clearly defined, isolated, and homogeneous population. Our analysis reveals how the concept of a population isolate has not been a stable and fixed concept through the years. Instead, the concept has drawn from several different methodological and conceptual approaches to what constitutes a population isolate, resulting in diverging understandings about that very same object of study. We suggest that this "flexibility" in the interpretation of isolation has helped researchers attract interest to Finnish genetics and create bridges to other population groups for purposes of comparison.*

Introduction

The Finnish population has been described as one of the "best-studied" population isolates in the world (Peltonen et al. 1999, 1913). In their article on Finnish population history, for instance, Palo et al. (2009, 1336) note: "As an isolated outlier population on the European genetic landscape, the Finns have attracted a great deal of interest among the geneticists." Drawing on the concept of isolation has served Finnish research in genetics

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well, but the promises of a population isolate for research has also evolved over the years (see Uusimaa et al. 2022; Kääriäinen et al. 2017).

Population isolates "are subpopulations resulting from the founder effect of a small number of individuals as a consequence of some type of bottleneck" (Arcos-Burgos and Muenke 2002). In addition to Finns, groups such as the Hutterites, old order Amish, and the Paisa community of Antioquia in Columbia are considered to be population isolates. In these populations it has been easier to identify the genetic causes of some diseases and medical conditions, such as asthma and multiple sclerosis. Historically, population isolates were seen as groups that had remained "unmixed" for generations. During the twentieth century, groups of people came to be seen as populations instead of races; the historical weight of eugenics and the race sciences of the late nineteenth and early twentieth centuries also prompted researchers to study human variation and human populations as dynamic rather than fixed categories (Lipphardt 2012, 2013). A focus on isolation was also part of this shift, as it helped establish certain ideas of "originality" to which later developments could be compared (Tupasela and Tamminen 2015; Lipphardt 2013).

In late twentieth-century biomedical research, population isolates were considered efficient in the study of monogenetic diseases (Hatzikotoulas 2014). Today, they are framed as promising in applications that include finding "entry points into the biology of common diseases through low-frequency, high impact variants" (Kurki et al. 2023, 508). Such recent framings serve to highlight the continued relevance of these populations for biomedical science and help identify why isolation sometimes plays such an important role in the characterization of populations. Understanding Finns as a population isolate characterized by "founder effect, genetic drift and isolation" (Peltonen et al. 1999, 1913) has been at the center of Finnish genetics research for decades, because of the small size of the country's total population. Finland's largest public-private genomics project to date, Finngen, is founded precisely on such characteristics (https://www.finngen.fi/en). Over the years, the project has highlighted several of these characteristics, such as the homogeneity of the population, as relevant, important, and particularly promising for genetic and genomic research both nationally and internationally (Helén et al. 2024). These claims, however, are not new. In a 2001 article, Finnish genetic researcher Juha Kere wrote that the Finnish population has "out of proportion for its size … by example shaped research in human disease genetics" (Kere 2001, 103). Even three decades earlier in 1973, pediatric researcher Reijo Norio and colleagues described the Finnish population as "rare flora in rare soil" (Norio et al. 1973) and, as such, possessing with certain unique attributes for biomedical research.

The history of Finns as a population isolate is thus in many ways both grounded in and a result of medical research. In this chapter, we want to contextualize how isolation has figured in different, yet related medical and scientific disciplines in Finland over time. We do not, for example, question the existence of a bottleneck effect as such, or that Finns may be a population isolate. Rather we aim to situate and discuss how this idea of an isolated population has been represented and what gives this population its characteristics. This includes examining how the understanding of a particularly defined population came to serve as a reference allowing Finns to be seen and claimed as unique, and how the inner heterogeneity of the population in Finland accords with the representation of a homogeneous population isolate. We argue that these discussions and scientific studies represent a preoccupation with two important aspects in genetic studies of populations. *First*, where do present-day populations come from? This is to say, what are the origins and "roots" of a people that have come to be known since the establishment of the Finnish nation-state in 1917 as Finns. *Second*, how have Finns, as they have been selected for various comparative genetic studies, been represented visually as relating to "other" populations. We see these two discussions as being fundamental to science and medicine, and more broadly to how the Finnish population is related to other populations in Europe and further afield. We also consider these debates and discussions to be central to the stabilization of Finnishness to a fixed time and place in history, enabling claims of origin and authenticity (Tupasela and Tamminen 2015).

We have previously discussed Finnish population within biomedical research from various viewpoints, such as population branding (Tupasela 2022; 2017), authenticity (Tupasela and Tamminen 2015), and the characteristics by which it is framed as valuable research material for biomedical research and development (Tarkkala 2019; Tarkkala and Tupasela 2018). In this chapter, however, we focus on representations of the Finnish population isolate and show the underlying dynamics that can contribute to the idea of isolated, homogeneous population. We also point out that "population isolate" is not a static concept but has varied over time and in different contexts in Finland.

Our discussion focuses on how the claim of an isolated population is represented visually in Finnish scientific work. Our interest in visualizations relates to how certain theoretical and methodological choices are represented visually, and how they simultaneously encapsulate certain ways of thinking and certain approaches in science, specifically in relation to the idea of the Finnish population isolate. We reiterate that the notion of population isolates might not prove to be valid if the selection criteria or pool of individuals selected for analysis were wider and less selective as it has been in Finland. In the following, we first will introduce visualizations to lay the groundwork for our analytical discussions, which will then draw attention to the way understandings of populations have been constructed and conditioned by the tools used, the availability of materials and methods, and the decisions made by researchers in various phases of their research. To this end, we augment our analysis of visual representations with a consideration of written and published materials from researchers working in this field. We have collected scientific articles on topic published between the years of 1972 and 2023 that discuss the characteristics of the Finnish population, either explicitly or as part of reporting results of certain analyses. We also utilize material from the internet that has been published by researchers and research institutions on their home page and contain information and visualizations about Finnish population history (Tupasela 2022).

Visual Representations in Genetics

In sciences, visualizations are a form of portraying evidence and condensing findings (Coopmans et al. 2014). Visualizations present data, expressing trends and relationships or even space-time interrelationships (Gross and Harmon 2014, 53). Visualizations might "witness what the scientist saw in the field, laboratory, office, or observatory" as well as "reveal the function of equipment" (Gross and Harmon 2014, 53). They can be produced as maps, images, plots, graphs, tables, and so on. In population genetics, visualizations are used for applications such as maps showing how people have moved from Africa to other continents, various kinds of evolutionary trees, or figures showing certain patters, variations, and connections (Sommer 2015; Palsson 2007).

More generally, images in science are usually produced based on certain instruments or imaging procedures, and they encapsulate the reasonings and methodological choices of their own time (Bredekamp et al. 2015, 1–3). As Bredekamp et al. (2015, 1) argue: "The transformation of observations, findings, and insights into images partakes actively in the constitution of knowledge." Images and visualizations can "create persuasive representations" (Burri and Dumit 2008, 297) for readers. In a similar vein, Lipphardt and Sommer (2015, 3) have argued that "images are at the heart of strategies of persuasion." Burri (2012, 54) calls for sociologists to pay attention to images, and while doing so "not focus on images alone but take the *social practices and contexts of image production, interpretation and use* into account" (emphasis in the original). Indeed, visual representations are always created and used in certain contexts, to deliver, strengthen, and stabilize certain understandings and arguments and portray them as stripped of too much complexity (Lynch and Woolgar 1990).

This is also the case in visual representations of the Finnish population and the population history claimed to have caused the isolated gene pool for this specific group of people (see Tupasela 2022). Some of the first notable images found in medical genetics in Finland date to the early 1970s, when doctors such as H. R. Nevanlinna (one of the most prominent doctors and geneticists in Finland to study population structure) published articles seeking to describe the history and development of early settlements in Finland. Similar maps can be found in historical and archaeological accounts of Finnish history. Historical texts and maps on the Treaty of Pähkinäsaari in 1323 were quite common in subsequent discussions on the genetic division of Finns between east and west and in creating a boundary between Swedish and Russian influence. This boundary also evinces striking correlations between Finnish dialects, medical reimbursements, as well as genetic differences (Palo 2020). Medical doctors such as Nevanlinna, however, began to link these forms of history to medical conditions and population genetics. Images of population structure and differences help to create distinctions that are easy to understand. The translation of similarity and difference to the visual realm, rather than offering only numerical figures, helps to convey the message of the researchers. Yet images and visualizations also help to conceal the meaning and methods necessary to create such images, thus producing a certain level of opacity.

Hence in this paper, we are not interested in analyzing the images per se, but in their function as representations of certain scientific settings (methodological, theoretical, etc.) that also contribute to different ways of seeing and understanding the Finnish population isolate.

Understanding Finns as a Population Isolate—How Was This Understanding Formed?

The understanding of the Finnish population as a population isolate emerged as a by-product of an effort to understand certain rare diseases appearing in Finnish families. The studies on what is known as Finnish Disease Heritage (FDH), a group of over thirty monogenetic, rare diseases that have been overrepresented in Finland, formed a basis on which wider understanding of Finnish population history could form (Norio 2000). FDH was first introduced in the Finnish medical literature in 1972 (Perheentupa 1972) and two years later in an English-language publication (Norio et al. 1974). For this endeavor, certain inclusion criteria were applied to delineate which diseases could be included under the umbrella term. The inclusion criteria for the Finnish population in this context included that the conditions had to be overrepresented in the Finnish population, and that a minimum of ten cases have been clinically diagnosed. The study of FDH produced several narratives of specific causes for these rare conditions, which drew on notions of migration and isolation. Figure 1 provides an example of one such map where early migration history is used to explain the isolation and early settlement history of Finns, which has resulted in the emergence of various FDH conditions.

Reijo Norio, a prominent Finnish pediatrician and perhaps the best-known expert in FDH has noted that the criteria for FDH are not particularly strict, in that some have been included in the list since they were not found in the literature or elsewhere in the world but have a relatively high prevalence in Finland. For Norio and his colleagues, the method for studying rare inherited diseases related specifically to studying family pedigrees and inheritance. In many, but by no means all, cases FDH diseases were the outcome of a relatively small population in which there were carriers of recessive genes. In addition, those carriers had to reproduce with another carrier. In this sense, many FDH conditions have been the outcome of a specific gene pool and general isolation of smaller populations within Finland (Norio, 2000: 25).

So-called superisolates or subisolates within Finland provide an example of how some FDH conditions have come about. The people who originated from founder populations of Kuusamo and Salla (Varilo et al. 2000), for instance, are known to be carriers of several recessive genes. One study by Varilo et al. (2000) compares Finnish and Kuusamo-area samples. Kuusamo and its inhabitation history, Varilo et al. (2000) argue, is an example of how long distances have resulted in population pockets where a very small founder population can be identified, and where inbreeding invariably followed. The Kuusamo area, for instance, was inhabited at the end of the seventeenth century and "39 families survived the great famine of 1695–1697" (Varilo et al. 2000, 605). Thus, "all inhabitants of Kuusamo born before 1939 have ancestors from these 39 families and show numerous genealogical connections" (Varilo et al. 2000, 605). Local historical conditions such as this have left a mark in the genetics of the population. As Peltonen has noted:

> One additional feature in the population history of Finland has greatly emphasized the concept of isolation. After the first founders the slow inhabitation process of this relatively large country, broken by hundreds of lakes, resulted in the formation of internal subisolates. Typically, some 20–40 families formed a settlement which could remain

> isolated for centuries. Additionally, most eastern and northern parts of the country became inhabited relatively late, the main population expansion beginning as late as the 17th century when people from southern parts of the country moved towards northern and eastern regions. Population records such as taxation documents and church records are well preserved from the 17th century and reveal highly restricted immigration to many subregions of Finland as well as limited population expansion in these subisolates, predicted to result in the regional enrichment of some alleles and in the loss of others. (Peltonen 1997, 553–554)

Figure 1: "Figure 1 A" and "B" as published in Varilo et al. 2003, showing the internal settlement history of Kuusamo and Finland as well as the way the sampling was done based on the birthplaces of grandparents from both sides of the family as inclusion criteria.

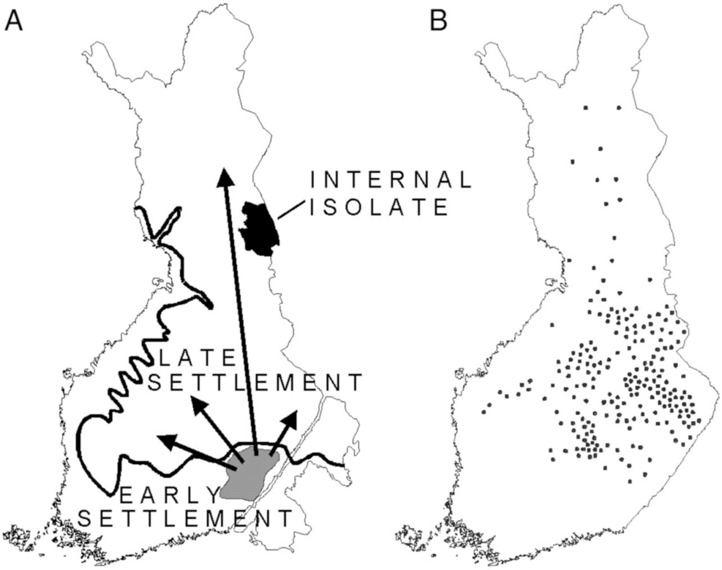


Figure 1 also shows how the different settlement histories of early and late settlements have been identified as key factors for population formation. These have been reproduced in several articles showing borders, areas, and arrows at the side of a map of birthplaces of grandparents specific to the late settlement that formed the essence of what the article identifies as the "Finnish population isolate." The frequent reproduction of these images in other sources has, moreover, constantly reinforced this understanding. Similar maps containing essentially the same information seen in figure 1, as taken from Varilo 2003, have been circulating since at least the 1970s (see Norio et al. 1973).

One result has been the emergence of an understanding of a genetically homogeneous population isolate of Finns. In his book *Suomi-neidon geenit: Tautiperinnön takana juurillemme johtamassa* (The genes of the Finnish fair maiden), Norio emphasizes both national isolation and regional isolation as causes for many of the inherited rare diseases listed in FDH (Norio 2000, 26). The work done on FDH in Finland served specific purposes in both consolidating rare disease research in Finland to provide critical mass when there were relatively few patients per condition. Norio additionally notes that being listed as an FDH condition also afforded publicity and visibility to conditions that needed support and funding from public authorities.

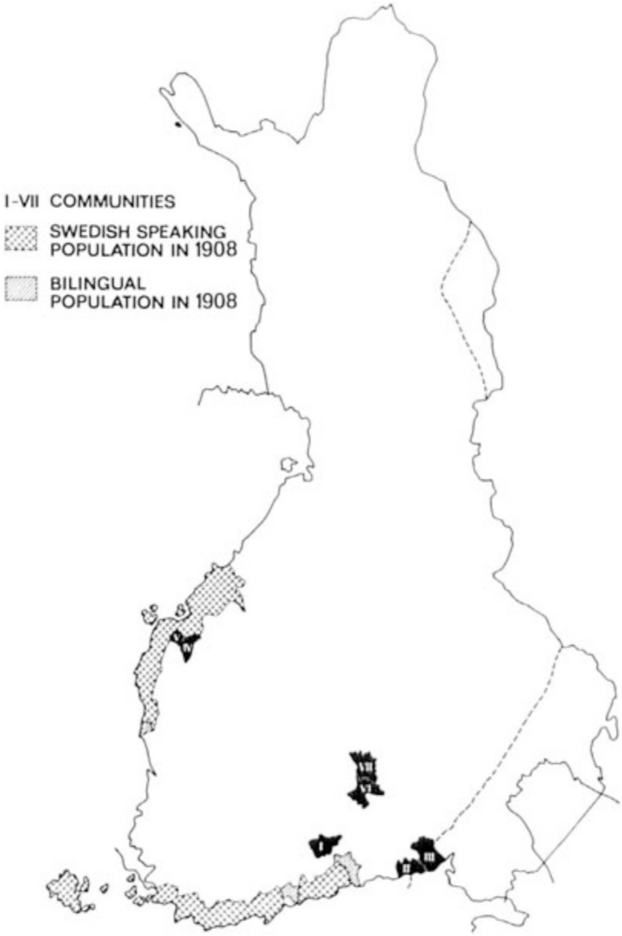
In addition to being referred to as a population isolate, in genetics the Finnish population has also been described as originating from a "founder population" (Lim et al. 2014; Wang et al. 2014; Järvelä et al. 2021). The specific term "founder population" refers to a specific group of people who usually traveled and settled to an isolated area, and whose isolation over several generations led to the enrichment and possible inbreeding of individuals who did not know they were related. As a concept it is more historically grounded and specific than isolated population as such. The same goes for the concept of a "bottlenecked" population (Minton 2023), which has been used to refer to specific events in Finnish history that have had an impact on the country's population structure and size. The notion of a founder population does not necessarily signify that a population has been isolated over the years, but rather that a small number of individuals originally inhabited a specific region.

Selecting Finns for Research

Early studies of rare diseases were always premised on following family pedigrees based on patients and families who had been identified as carriers of specific rare diseases. As mentioned above, the emergence of the concept of Finnish Disease Heritage came to represent a group of diseases that were thought to be overrepresented in the Finnish population. As Reijo Norio has noted, however, it could not be ruled out that such conditions did not exist elsewhere, since not all countries had healthcare systems that reported and studied rare conditions such as those found in Finland (see Tupasela 2016).

In contrast to these studies on rare diseases, other early studies into the Finnish population structure drew heavily on the idea of selecting individuals from rural areas. As H. R. Nevanlinna noted in an article published in 1972: "In order to eliminate the effect of different groups on the national level (and that of individuals not belonging to the original population in the communities), the sampling was made as a weighted one from the rural communities only" (Nevanlinna 1972, 199). The notion of an "original population" is not exactly clear in this context, in that Nevanlinna does not specify in his publication whether he is referring to a founder population or another type of criterion regarding how long someone has been living in a specific area. This selection criteria appears rather problematic given that, only several lines earlier in the same article, he observes: "The Swedish-speaking urban population seems to be of mixed origin" (Nevanlinna 1972, 198). Although the urban population represented a relatively small portion of the whole population, its diversity was significant in that it included Jews, Russians, and Roma. Despite the small sizes of these groups, arguments for who counts as a Finn have played an important role in subsequent selection criteria in Finnish studies. In addition to excluding Sami, early studies thus sought to clearly define which individuals could be considered to represent Finnishness at a genetic level. The exclusion criteria also included women, as well as all individuals who lived north of a diagonal line that went approximately from the northwest of Finland to the southeast (see figure 2). The criteria that were introduced in these studies have played an important role in stabilizing who is included in studies of population genetics in Finland.

Figure 2: Selected populations for study



Source: Nevanlinna 1972, 201

Figure 2 provides an example from Nevanlinna's article. The type and form of mapping found in this piece was used quite prominently throughout the end of the last millennium and can be also found in subsequent articles and publications by such prominent doctors as Reijo Norio (2000) and Jaakko Perheentupa (1972). More contemporary use of population genetic images is based on methods such as principal component analysis and often also includes reference populations from other countries (see figure 3 for an example).

Certain images and the information they contain are consistently used in publications on the topic (e.g., figures 1 and 2), which reinforces the persuasiveness of the image and what it represents. But the reuse of images and narratives tends to detach the images from the original methodological and sampling-related discussions and framings. This can be seen, for instance, in several newspaper articles that have sought to popularize the study of Finnish genetics and the notion of Finns as "two people" ("kaksi kansaa") (see Kotkavirta 2019; Puttonen 2017).

These early selection criteria have contributed in part to the notion of Finland as an isolated population. One might go as far as argue that the early selection criteria served to solidify the notion of Finland as a homogeneous population. Interestingly, the early exclusion of the Finnish-Swedish population found along the western coastline of Finland would also provide the basis for later findings relating to genetic differences among the Finnish population.

Connecting Rare Diseases and Population Genetics

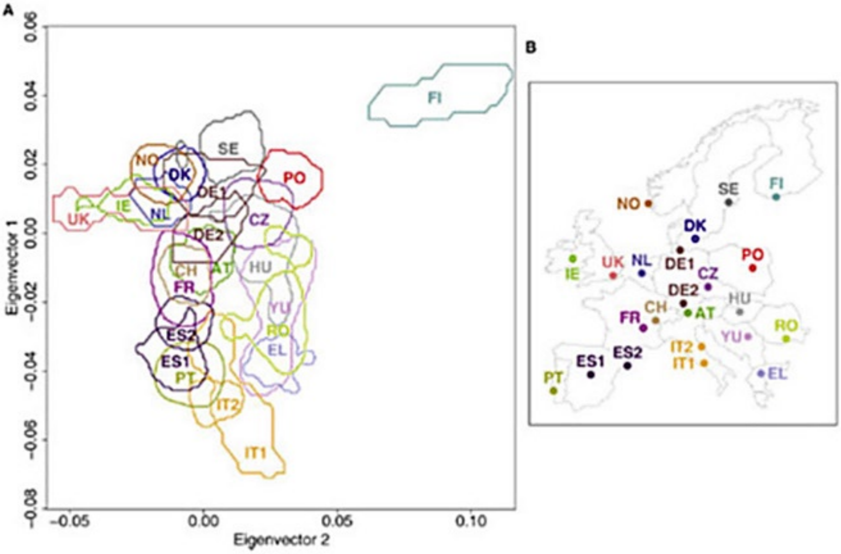
The historical narratives of Finland as a population isolate developed through the study of Finnish Disease Heritage were later connected with the broader field of population genetics and molecular genetics in the scientific literature in the mid-1990s. The work of Finnish geneticist Leena Peltonen has especially emphasized and drawn on the connection between the history and development of rare diseases in Finland and the more general population and its impact on national genetic structure and substructure (Helén et al. 2024). But the shift from the study of rare diseases, which focused on families and family pedigrees, to the study of more complex diseases required that connections needed to be made regarding the relevance of isolation to complex diseases and to the identification of genetic loci that played a role in their development. This "translation" from monogenic to complex on the one hand, and rare to common on the other, is an important bridge between the two periods and methods of studying disease in Finland (see Tarkkala 2019).

One such article establishes a connection between the major causes of rare diseases (isolation, bottleneck, and founder population) and "major loci" contributing to complex diseases. The piece focuses on the examples of Aspartylglucosaminuria (AGU) and infantile neuronal ceroid lipofuscinosis (INCL) to make this link. As it notes: "In both of these lysosomal accumulation diseases one major (point) mutation is carried by 98% of diseases alleles in Finland" (Peltonen et al. 1995, 699). The connection between rare disease research and the causes of common diseases is an important step within the Finnish context as it helps to reaffirm and accentuate the narrative of an isolated population. Despite the significant differences in relation to methodology and sampling between rare diseases and common/complex diseases, the story of isolation, bottlenecks, and founder population has played an important role within subsequent population studies in Finland. This linkage is especially significant as, during the past decade, Finnish researchers have had to "backpedal" somewhat on the claims of homogeneity and uniqueness to make Finnish samples and data of more relevant internationally (Tarkkala and Tupasela 2018).

After the mid- to late 1990s, the sequencing of the whole human genome sequence introduced a host of new techniques and methods for studying complex diseases in Finland. An important development during this time concerns the ways in which visual representation also began to develop to include new types of methods and techniques in analysis and representation. A significant feature of this shift was the introduction of comparison to other populations as a means of understanding Finnish uniqueness and difference.

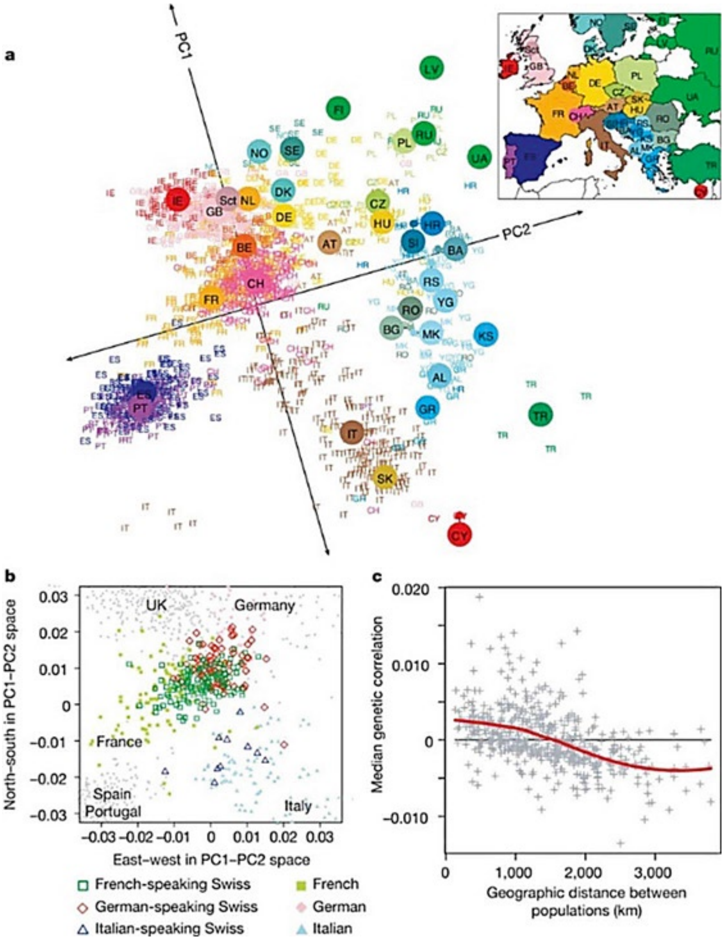
One example of the representation of Finnish isolation in relation to other European populations can be found in a comparative study of European populations by Lao et al. (2008). As seen in figure 3, a comparison of twenty-three European subpopulations generated an image where Finns are presented as distinct from other European populations. Images such as this have played an important role in enforcing and emphasizing the unique and isolated nature of the Finnish population. The image in figure 3 is somewhat misleading in that similarity and difference are represented on two axes. On the y-axis, Finns are quite close to Swedes, Norwegians, Danes, and even the Polish population. On the x-axis, similarity is with Poles and Hungarians. The image is an important milestone in the study of populations, however, due to the way populations are represented visually and in relation to others.

Figure 3: SNP-Based PCA of 2,457 European individuals from twenty-three subpopulations



Source: Lao et al. 2008

Figure 4: A statistical summary of genetic data from 1,387 Europeans based on principal component axis one (PC1) and axis two (PC2).



Source: Novembre et al. 2008

This image can be compared to another similar study published by Novembre et al. (2008), which includes more subpopulations. In this study and the accompanying image (figure 4), the Finnish population appears far less isolated with the inclusion of samples from Russia and Latvia. The way the visualization uses dots, rather than circles, also plays an important role in how visualization contributes to how we understand similarity and difference, and thus isolation as well. The role that reference populations have played in the representation of difference and similarity has been similarly influential in constructions of Finns as a population isolate. The inclusion and exclusion of specific reference populations impacts the relative distance that the chosen population representatives are from each other. Therefore, adding a new reference population may either increase or decrease relative distances from other populations, which will either emphasize or de-emphasize the visual representation of the isolation of specific populations. This impact can be seen in comparing the differences between figures 3 and 4. As more populations are added to comparisons, the relative genetic differences between each population becomes less and less distinct.

In genomics and medicine, understanding a certain population as distinct, and thus as different from another population, can play a significant role in perceptions and practice. Each population can thereby be seen to carry exceptional characteristics, which might prove to be important for certain scientific findings, methods, and approaches, as happened in the case of Finnish Disease Heritage. Certain methods, together with certain samplings of Finns, produced a corpus of knowledge of these monogenetic, rare diseases: linkage disequilibrium studies, for instance, were conducted for this purpose (Hästbacka et al. 1992; Varilo et al. 2000).

Each population gains its specific, exclusive characteristics from the comparison to other populations, seen to embody something different. This logic is, at its core, simple. However, when 99.9 percent of human DNA is shared, the effort to draw lines between groups of people is a significant task requiring careful sampling and scientific precision. In medicine, to repeat what was noted above, these differences might be significant, and scientists continuously fine-tune their approaches to be able to detect even rare variants (Martin et al. 2017). Yet because many studies have been done in European populations in the past, there has been a call to diversify the field, as the "lack of ethnic diversity in human genomic studies means that our ability to translate genetic research into clinical practice or public health policy may be dangerously incomplete, or worse, mistaken" (Sirugo et al. 2019, 26). Nevertheless, the question of comparison and of the reference point against which a certain group is compared is always meaningful for settings of genetic research. Potential "noise" in data often needs to be controlled, and in a homogeneous population there is less noise to start with due to high level of similarity—a fact that Finnish biomedical research has leveraged. As we have shown in our previous studies (Tarkkala and Tupasela 2018; Tarkkala 2019), exaggerating the uniqueness of the Finnish population may produce a conclusion that the Finnish population does not provide the necessary means for comparison if it is considered as mere outlier population in which results based on analyses of other populations cannot be reproduced or replicated. However, in European comparisons Finns often seem to stand out (Novembre et al. 2008). The emphasis of Finns as unique and isolated has therefore required a kind of balancing act on the part of Finnish researchers.

The inclusion of Finns as "European" is important for Finnish biomedicine. There has been a concern about whether the Finnish samples could be compared with other samples—that is to say, whether they have been handled and stored in a way that does not add variation in the analysis, and whether findings in the Finnish population can be replicated in others (Tarkkala 2019). European samples have been used in biomedicine to a great extent and have been considered useful; hence it is important to include the following:

> European GWA studies have produced many successes that can be replicated in different sets of individuals from the same European population as that in which the association was originally detected; and associations in one population of European descent

> are often replicable in other European populations, sometimes in groups that are quite geographically distant within the European continent. (Rosenberg et al. 2010, 358)

It is worth asking whether the perceived uniqueness of the Finnish population as an isolated population would remain as strong if samples from a broader pool of individuals living in the geographical area of the country had been included from the beginning. The role of visualizations and relevance to European GWA studies cannot be overstated, however, as Finnish genetics has sought to find a place in the global market for genomics research using the strengths that it has developed over decades.

One, Two or More Than Two Populations

Even though Finns have been widely represented as a homogeneous population isolate, the population in Finland is in fact also heterogenous, as several publications have documented (Salmela et al. 2008; Huyghe et al. 2011; Kerminen et al. 2017; Virtaranta-Knowles et al. 1991). Discussions centered on homogeneity and heterogeneity are crucially related to both the reference populations used for comparison and the scale at which population structure is analyzed. And yet these discussions are also related to how isolation is understood and represented. Within the context of what constitutes a population isolate, there is no commonly agreed upon definition or standard against which isolation and homogeneity are compared. Therefore, there are multiple and sometimes even different definitions of what constitutes homogeneity within a population. And since there is no agreed upon definition of what scale of analysis is meaningful for claims of homogeneity or heterogeneity, the term ends up being somewhat fluid and fuzzy. As Kerminen and colleagues note:

> Our haplotype-based population assignment using a data set that evenly covers a major part of Finland therefore provides unprecedented information on the fine-scale genetic structure of the Finnish population. In general, the fine-scale structure that we detected is highly geographically clustered with little overlap between the populations. (Kerminen et al. 2017)

This observation points to the way in which scale can play an important role in how a larger population isolate, such as Finns, can further be subdivided into smaller distinct populations depending on the scale of analysis.

Interestingly, the division between people originating from western Finland and people originating from eastern Finland is clear-cut and has even been said to point to two distinct populations. This internal heterogeneity is important to the notion of Finns as a population isolate, as when the heterogeneity is accounted for, many Finns have much closer ties to other populations, such as Swedes, than previously claimed. The dichotomy is also something that Nevanlinna noted in his early studies and recognized as contributing to difference within Finland, and thus as impacting selection criteria for early studies. Another example of how internal heterogeneity can be generated comes from an article by Salmela et al. (2008), which sought to study the population structure among western Finns (most of whom are considered Swedish-speaking). To understand the differences among this population, the authors compared samples from western Finns with those of Swedes. They note:

> 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) (emphasis added)

The generation of such differences within the Finnish population, and all populations for that matter, raises two important questions. *First*, what reference populations are relevant in studying the population structure of any given group of people? *Second*, when differences are created through comparison, which differences can we consider to be significant and on what basis is that significance determined? How, for example, might one compare the significance of one so-called isolated population to another? What criteria of quality or quantity signify an important difference or benefit that one population may have compared to another?

A further issue of concern in relation to definitions of isolation is the criteria that are relevant for defining isolation and differentiation from other populations. As the article by Kerminen et al. (2017) shows, the scale of analysis can have significant impact on delineating what constitutes the borders and composition of a specific population cluster. Which individuals are included or excluded from the group and why?

The discussion of similarity and difference within Finland has further implications for international comparisons and studies of populations. What is the role of difference and similarity in these studies, and what significance does it have? What relevance does it have, for example, in identifying disease-causing genes? Furthermore, to what extent should studies include more diverse populations and what impact might this have on generation of noise and the need for larger samples sizes? Or to what extent does the introduction of strict selection criteria mask and hide the identification of other disease-causing genes that may have less relevance for a small population like Finland, and more relevance for larger populations?

Discussion: Changing Representations of a Population Isolate

In this chapter, we have traced the origin and changes of the concept of a population isolate in Finland. We focused first on the late 1960s and early 1970s, when the term was first associated with the idea of Finnish Disease Heritage. Over time, especially after the turn of the millennium, the concept became increasingly linked to molecular genetics and population genetics more broadly. This historical arc demonstrates that the concept has not been scientifically stable. Instead, it has been quite flexible, proving adaptable to various perspectives, methodological approaches, and selection criteria when determining which people to study in Finland. Within the Finnish context, some aspects of isolation have been constructed both methodologically and conceptually.

We have shown how these various concepts of population isolation have been popularized and represented visually, in certain images that have been widely reproduced and circulated in both academia and popular literature. These images are often intended to persuade readers to accept certain claims without fully understanding the underlying methodological and conceptual choices. However, visual representations can also be misleading and confusing, as they may obscure the criteria used to select individuals deemed representative of a given population.

The combination of historical perspective and more recent forms of visualization serves as a powerful tool of persuasion. In the Finnish context, this persuasion has been crucial in the construction of knowledge (Bredekamp et al. 2015), presenting a unified narrative despite the availability of alternative possibilities and explanations (Översti et al. 2019; Översti et al. 2017). As several scholars have noted (Burri and Dumit 2008; Lipphardt and Sommer 2015), images play a significant role in this process by condensing information and simplifying complex issues (Lynch and Woolgar 1990).

Additionally, we have extended the understanding of images of populations isolates by highlighting the role of social practices and interests that underpin the generation of these images. In Finland, different notions of population isolation have often been tied to the criteria and focus of analytical methods and selection criteria. This is evident in early studies by Nevanlinna (1972), as well as more recent research by others who examine the genetic characteristics of Finland's population structure in studies on Y chromosomes (Palo et al. 2009). Our study does not argue that isolation has never been a feature of people living in different parts of Finland. Instead, it suggest that the concept of isolation and its connection to genetic analysis is heavily influenced by social factors, including researchers' decisions on selection methods and which parts of the genome to explore for similarity or difference.

Since population isolates have historically been seen as groups that have remained 'unmixed' for generations, we suggest that the criterion for this determination is highly fluid and susceptible to interpretation. As Lipphardt (2012) has argued isolation or 'unmixing' has usually been based on other factors than genetics first. Therefore, issues related to sampling selection, scale as well as reference populations are not brought forth or raised as possible concerns when images are circulated. Thus, the images seek to convince individuals of a specific truth related to notions of isolation. As said in the beginning, our intention is not to question the validity of genetic drift, founder population, bottleneck effect, or isolated homogeneous population as concepts applicable to Finland or elsewhere. Rather, we have sought to contextualize these notions within a broader perspective on populations and in relation to visual representations that have circulated for decades. By doing so, we demonstrate that the notion of an isolated population is not uniform but has some flexibility, as highlighted by the examples given in this chapter.

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