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Medical Geneticists' Interpretations of Genetic Disorders in Roma Communities in Post-Socialist Hungary

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The article investigates the utilization of ethnic classification by human geneticists in Hungary, with a particular focus on the Roma minority. Drawing on qualitative expert-interviews, it analyzes how historically situated social imaginaries inform the production of genetic knowledge. The study explores how human genetics constructs heritable disorders as ethnic diseases, exposing the epistemological and ethical tensions inherent in translating sociocultural difference into biological terms.

▪ **Keywords**: population genetics, race/ethnicity, Roma, public health, East Central Europe

Avtor na primeru romske manjšine preuči uporabo etnične klasifikacije pri genetikih na Madžarskem. S kvalitativnimi intervjuji s strokovnjaki analizira, kako zgodovinsko umeščeni družbeni imaginariji oblikujejo ustvarjanje znanja o genetiki. Raziskuje tudi, kako človeška genetika konstruira dedne motnje kot etnične bolezni, pri čemer razkriva epistemološke in etične napetosti, ki so neločljivo povezane s prevajanjem družbenokulturnih razlik v biološke pojme.

▪ **Ključne besede**: populacijska genetika, rasa/etničnost, Romi, javno zdravje, vzhodna in srednja Evropa

Introduction

This paper examines the complex relationship between genetics, ethnicity, and public health, with a specific emphasis on the Roma population in Hungary. It provides an in-depth exploration of how human genetics has historically understood, classified, and addressed various health disparities and susceptibilities among ethnic groups, especially focusing on how these perspectives and methods apply to the Roma community. Research in population genetics seeking to map genetic predispositions to particular diseases among different ethnic groups has produced medically important studies, since its results can influence public health programs that help the management of epidemiologically significant diseases. In Hungary, the disparity in health outcomes between Roma and non-Roma populations has been particularly stark since the political transitions of the early 1990s. Roma populations face disproportionately high rates of unemployment, low educational attainment, and increased exposure to poor living conditions, which collectively impact their health. Consequently, research has shown that Roma individuals report lower health standards and greater difficulties in accessing healthcare services. Studies emphasize the importance of culturally aware healthcare practices, as Roma communities often encounter discrimination in medical settings,

exacerbating their health disparities. This acknowledgment led to various efforts, especially with Hungary's aspiration to join the European Union, which imposed criteria requiring improved minority health outcomes as part of a commitment to minority rights. Genetic studies, contributing to this strand of research, further underscore the need for targeted health interventions. Research has demonstrated that Roma communities have a higher frequency of certain genetic mutations that increase their susceptibility to a spectrum of conditions (i.e.: cardiovascular disease, congenital glaucoma). Population genetic maps reveal the statistically significant diseases, and thus genetic screening programs could mitigate these health risks by facilitating early diagnosis and encouraging preventive measures. However, researchers also caution against attributing all health disparities to genetics alone; socioeconomic factors, cultural practices, and historical marginalization are significant contributors to the observed health inequalities between Roma and non-Roma populations.

The following work begins with a brief historical overview of genetics, because it is important to demonstrate how genetics became a dominant scientific discourse in public health. After the Second World War, a paradigm shift occurred, and the production and use of genetic knowledge to address both individually and socially relevant medical problems became the accepted goal. This historical overview is important because the geneticists' narratives of the post-1990 discourse resonate with the scientific understandings of the state socialist period. Following a historical overview, the methodological principles, data collection, ethical considerations, and method of analysis are presented. The analysis addresses three main perspectives: (1) ethnic grouping (the ethnicization of medicine), (2) the medical meaning and interpretation of isolation, and (3) the medical issue of inbreeding. These aspects reveal the medical genetic logic of mapping diseases in relatively closed groups to promote the health of Roma. The logic of group formation and its medical interpretation will here be traced with the aim of developing an understanding how ethnicity is applied in medical genetics.

Genetics as a source of information for public health

Medical genetics began to establish itself as a distinct field in the latter half of the twentieth century. Harper compares this progression to the separation of human genetics from the broader discipline of genetics, which began in the 1940s. Although the roots of medical genetics can be traced back to the mid-1950s, its development was uneven. Focusing especially on the United States, Harper notes that medical genetics had emerged as a recognized discipline by the 1980s. In Europe, while the scale of development was comparable, it was more fragmented, as different nations evolved along separate paths following the Second World War. Harper defines medical genetics as a hybrid science, combining human genetics with medical and scientific applications

(Harper, 2008). With the advent of genetic analysis, it became possible to detect mutations that place individuals at increased risk for certain diseases (Kosztolányi, 2000). This allowed medicine not only to diagnose conditions but also to offer patients and clients information about lifestyle changes that could help prevent disease. This proactive approach represents what Kosztolányi describes as real prevention – the ability of medical genetics to identify genetic risks and guide individuals in maintaining their health (Kosztolányi, 2000: 2423). Initially, medical genetics concentrated primarily on individuals and families, diagnosing inherited diseases and providing options once a diagnosis was made. However, its focus expanded following the results of the Human Genome Project (HGP), which enabled the medical community to address conditions of broader epidemiological importance, such as asthma, cardiovascular diseases, and cancer. These conditions are significant because genetic analysis allows for the prediction of susceptibility, giving medicine a more active role in preserving public health and quality of life. Among the major achievements in this field, the HGP stands as a landmark in the history of medical genetics. Allison (2004) highlights the importance of its findings, noting that genetic research is now capable of clarifying the underlying mechanisms of diseases and developing targeted treatments. The latter is no longer a distant goal – according to Allison, such genetic knowledge is already being applied by researchers to identify genomic targets and create molecular genetic therapies.

While a substantial body of literature highlights the role of genetic knowledge in advancing individual and collective health promotion, it is important to recognize that pre-existing social inequalities and tensions may hinder the realization of these medical objectives. From a public health standpoint, the application of genetic knowledge is frequently referenced as a positive exemplar in the context of screening programs for conditions such as Tay-Sachs disease and β-Thalassemia. The earliest cases of Tay-Sachs disease (TSD) to be systematically examined occurred within the American Ashkenazi Jewish community. According to Ruth Schwartz Cowan (2008), the relative success of TSD screening programs can be attributed to their being organized, financed, and disseminated both within and by the Ashkenazi Jewish community itself. A critical factor contributing to this success was the voluntary nature of both the program's workforce and its participants. Equally significant was the support the initiatives received from religious authorities within the community, which further legitimized and encouraged participation in the screenings. A comparable example is the state-organized, clergy-supported β-Thalassemia screening program in Cyprus. Here, voluntary nationwide screening efforts have led to a substantial reduction in the prevalence of this hereditary condition within the Cypriot population (Schwartz Cowan, 2008; Kakuk, 2013). Both cases exemplify how community-based, culturally sensitive, and voluntary genetic screening programs can empower individuals – whether identifying as Ashkenazi Jews or Cypriots – to access genetic services when they perceive themselves at risk of transmitting hereditary disorders. In contrast, the history of sickle-cell

anaemia screening in the United States offers a telling counterexample. This genetically inherited, life-threatening disorder was first studied within African American communities. Despite its epidemiological significance, comparable to that of Tay-Sachs or β-Thalassemia, screening programs for sickle-cell anaemia were largely unsuccessful. Rouse (2009) identifies two principal reasons for this outcome: firstly, these initiatives were organized externally, without the active involvement of the African American community; and secondly, the programs unfolded within a broader historical context of systemic racism and medical mistrust. As a result, African American communities viewed the predominantly white medical establishment with scepticism, undermining the effectiveness of these public health interventions.

In the post-WWII period of state socialist Hungary, physical anthropological research persisted, with a primary emphasis on the description of the physical characteristics of various populations (Bartucz, 1954, 1955; Lipták, 1954; Nemeskéri, Deák, 1954). These studies often intersected with research on heredity, producing applications such as methods for paternity verification (Fehér, 1954) and investigations into the inheritance patterns of congenital anomalies (Szemere, Csík, 1962). Scholarly interest expanded to encompass the relationship between genetics and blood group distribution (Nemeskéri, Thoma, 1961; Rex-Kiss, Horváth, 1970), alongside broader inquiries into the population genetics of blood types (Rex-Kiss, Szabó, 1970). Advancements in biotechnology during this period facilitated the expansion of genetic studies to encompass larger populations and, by the 1980s, this work increasingly included the investigation of ethnic groups across Hungary. The studies were largely motivated by the pursuit of medically significant findings that could inform public health policy and practice. A significant contribution to this discourse was made by medical geneticist Endre Czeizel, who in 1982 published an article addressing the relevance of population genetics to the fields of medicine, history, and national identity. Czeizel defined population genetics as "the study of a population whose individual members belong to the same species" (Czeizel, 1982: 2271), framing its social utility primarily through a medical lens. He argued that it was widely accepted within the genetic sciences that distinct population groups – often delineated by race or ethnicity – exhibit divergent disease spectra as a consequence of long-standing patterns of endogamy. Consequently, Czeizel advocated for the systematic investigation of disease prevalence within the Hungarian population to enable more effective disease management through medical genetics (ibid.: 2276).

The 1980s witnessed the emergence of comparative studies assessing the distribution of genetic mutations among various ethnic communities. A notable early example is the work of Flatz and his colleagues, who examined lactose intolerance rates across ethnic groups in Hungary. Their findings indicated minimal variation between groups with the exception of Roma communities, where lactose intolerance prevalence was recorded at 56%, significantly higher than the 37% observed in the general Hungarian population (Flatz et al., 1984: 147). Further research attention was directed towards

the mapping of cystic fibrosis (CF) mutations within Hungary (Németh et al., 1996). By the early 1990s, five mutations had been identified as the predominant causes of CF in Europe. The Hungarian data revealed that the ΔF508 mutation accounted for 64% of the cases, a distribution closely resembling that observed in Poland and Finland. The prevalence of other mutations (G542X, G551D, R553X, and N1303K) was comparatively lower in Hungary relative to neighbouring countries. Raskó and Kalmár (2003) posited that mapping the genetic distances between ethnic communities constitutes an objective scientific method for establishing correlations between genetic traits and disease susceptibility. In a similar vein, Judit Béres, a population geneticist specializing in the genetic structure of Hungary's Roma population, has argued that certain genetic diseases disproportionately affect Roma communities. The justification for targeted genetic screening within these populations is frequently predicated upon the high incidence of genetic disorders and the persistence of culturally sanctioned endogamous marriage practices (Béres, 2003). Judit Sándor (2013) explains that the situation of the Roma minorities in Central-Eastern Europe needs much improvement in many ways; they have endured harm for centuries, and thus they treat any majority intervention with reservation. It is also widely acknowledged that mapping their healthcare problems could benefit their communities, and thus genetic research in Roma groups should not only be based on individual information distribution and individual consent, but should be based on broad public consultation (Sándor, 2013: 241). Scholars have further argued that, given the enduring social and geographical marginalization of the Roma in Hungary, such genetic mapping initiatives are in the best interest of these communities, facilitating more effective medical intervention and public health planning.

Public health standards of Roma and non-Roma Hungarians after the transition

After 1989 it was widely recognized by researchers that there was a huge gap between the health standards of Roma and non-Roma Hungarians. For example, in their study, the sociologist György Gyukits and his colleagues noted that the Roma population suffered the biggest losses during the transition (Gyukits et al., 2000). Unemployment rates were very high among Roma – estimated to be around 45 percent; thus, their socioeconomic situation largely determined their health standards. In their study, Gyukits and colleagues explored the causes of the very low participation rates of Roma women in lung-screening programs. They claimed that in order to understand how effective prevention programs are, the state must know about patient behaviour during those programs. Their results suggested that prevention programs and campaigns that target Roma people should be designed so as to distribute information and help preserve their health. Furthermore, a problem was identified they could not answer: there was

a sizeable difference between Roma and non-Roma Hungarians even from similarly lower-educated groups. The initial hypothesis was that education could play a key role in utilizing various healthcare facilities, but research results suggested the contrary.

Other researchers, similarly focusing on environmental factors, pointed out that the health of Roma living in slums is much worse than that of Roma living in average circumstances (Kósa et al., 2008). In addition to their detrimental social situation, slum-dwellers endure high rates of discrimination when they want to use healthcare services. In their study, Zoltán Kósa and colleagues compared the 45–64 age group and found that Roma people view their own health status in a worse light than those who live in a similar socioeconomic stratum but not in slums. They argued that the health status of the Roma must be improved through multisectoral programs (such as health education) that are designed to target specific communities. Without structural changes, their health status will remain well below average standards.

Elevating the health of minorities as a criterion for joining the European Union

Hungary's intent to join the European Union initiated numerous epidemiological studies. This is because Hungary had to meet the Copenhagen Criteria that "require the respect and the protection of minorities through the stability of institutions that would ensure it" (Kósa et al., 2002: 2419). Consequently, a government program dealt with the issues of Roma minorities, and researchers such as Karolina Kósa and her colleagues contributed studies to the health focus of the program by analyzing the general Roma health standards. In order to facilitate measures that would improve Roma health conditions, it was necessary to survey their demographic situation, their morbidity and mortality rates, their genetic specificities, and their health behaviour. According to Kósa and colleagues (ibid.: 2424), only those genetic factors should be examined that significantly influence life quality (such as inheritable disorders and epidemiologically relevant genetic factors). The aim of the studies would be to identify those genetic factors that pose a high risk. With the knowledge at hand, it would then be possible to design screening programs that boost the effectiveness of prevention. They also acknowledged what the data suggest: that the detrimental health situation of the Roma is the result of their marginalized socio-economic positions – "their ethnicity is only epidemiologically important as a confounding factor" (ibid.: 2424). They also noted that any study interested in the mapping of health status must take into consideration the data protection law of 1992[1] that makes the use of racial, ethnic, or national identity in a medical context dependent on the consent of the individual or on legal approval. Kósa and colleagues argued that the new law made it difficult

[1] The LXIII. law of 1992 on data protection of personal information and the accessibility of public data.

to collect precise information on the health standards of Roma, but on the other hand praised it as a progressive measure that places ethnic/racial identification in the sphere of personal autonomy.

Genetic studies point towards the need to focus on mutations that are perhaps more frequent in Roma populations because of their cultural isolation (Kósa et al., 2002: 2422). Geneticists found that most of the Roma choose partners from within their Roma communities, and also found that endogamous marriages are much more common among Roma, which makes the prevalence of mutations and thus certain genetic disorders more frequent in these groups. Furthermore, healthcare studies pointed out that Roma people have an increased susceptibility to heart and cardiovascular diseases. For example, the Leiden mutation that makes people susceptible to thrombosis is much more frequent in Roma living in East Hungary (12.2 percent of them carry the mutation in its homo- or heterozygous form, against only 9.8 percent of non-Roma Hungarians). Other geneticists, like Kiss and colleagues, compared Hungarian Roma (Vlachian gypsies[2]) with non-Roma Hungarian populations. They focused their studies on allele polymorphisms that decisively influence tumour development and thus mortality rates of those who carry these genes. It was argued that studies are already published in which scholars have pointed out the higher rates of congenital glaucoma, galactokinase deficiency, and polycystic kidney disease in Roma. The researchers' position was that studies of allele polymorphisms are vital in order to design targeted preventive strategies (Kiss et al., 2004: 69). Furthermore, by comparing their results to literature results from Indian and Caucasian populations, they found that Hungarian Roma people differed from non-Roma Hungarians regarding the prevalence of GSTM1[3] and p53[4] genes; finding also that regarding the NAT2[5] gene, the Hungarian Roma fall between the non-Roma Hungarian and Indian populations (Kiss et al., 2004: 72). Because of these findings, they argued that a part of the Hungarian Roma population, the Vlachian gypsies, are more susceptible to developing certain tumours than members of non-Roma and other Roma ethnicities. This is important because it implies that targeted knowledge distribution would be crucial in tackling the related diseases. Thus, targeted screening programs could be developed for use by people who identify as members of these groups to find out who carries the genes at risk and to manage their health more consciously.

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[2] The term itself is used by geneticists, as this is the official ethnonym of a subgroup of Roma within Hungary.

[3] Glutathione S-transferase mu 1 (National Library of Medicine, 2025); the mu class of enzymes joined with glutathione functions in the detoxification of electrophilic compounds (i.e.: carcinogens, therapeutic drugs, oxidative stress).

[4] Tumour protein 53 gene (or p53 gene); it is essential for regulating DNA repair and cell division (MedlinePlus, 2020).

[5] N-acetyltransferase 2 (National Library of Medicine, 2025); this gene encodes an enzyme that functions to both activate and deactivate arylamine and hydrazine drugs and carcinogens.

Kósa recalls that in the European Union, a complex public health program carried out between 2003 and 2008 was working on health equality; this was accompanied by a study that suggested looking for structural inequalities that hinder equality in health status among various populations. The European Committee issued a proposal in 2009 to start another complex program that would address these problems (Kósa, 2009: 334). The WHO began to research this topic already in 2005, and the results were published in 2008. Karolina Kósa summarized their most important findings on how to reduce inequality: (1) improve living conditions; (2) reduce the inequality in the distribution of power and economic resources; and (3) utilize targeted healthcare interventions. She argued that although reducing health inequality was the declared goal of the Hungarian public health programs, this would not be achieved. The goal was explicitly stated in the National Public Health Program adopted in 2003: the program aimed at helping the Roma, handicapped, and homeless gain access to healthcare. Kósa believed, though, that its strategic direction was deficient because it did not correspond to the suggestions of the European Committee. The problems that she emphasized relate to structural inequality; whereas the program was not addressing social and economic factors that determine the health of the marginalized groups. Thus, Karolina Kósa argued that access to health services by itself would not solve health inequality in the long term. One of the key arguments put forward to enhance the wellbeing of Roma people in Hungary is the inclusion of Roma perspectives in shaping and executing local policies (Fésüs et al., 2010: 317). Efficient contribution from Roma people – in which they could explain how they experience health problems and other relevant social issues – would help healthcare professionals, social scientists, and political decision-makers to design programs that include their needs, and thus prioritize economic and human resources in order to develop Roma living conditions, working possibilities, and ultimately their health standards.

One of the key starting points of the discussion was the European Union's Roma integration plan drafted in 2011; in this plan a key pillar of integration was understood to be healthcare. Among the various problems related to healthcare, the plan emphasized the need to improve access to health services, decrease infant mortality rates among Roma children, and improve the visibility of Roma issues in healthcare data (Balázs et al., 2011: 68). Balázs and his colleagues drew on the work of Marc Lalonde (who was the Canadian Minister of Health) in arguing that ethnic classification of healthcare data could advance the wellbeing of minorities. In his 1974 work 'A new perspective on the health of Canadians', Lalonde stated the importance of acknowledging that humans are biological and social beings, and thus ethnic specificities should not be left out from epidemiological programs, otherwise important information is lost to healthcare services. In this light, Balázs and colleagues argued for the integration of ethnic classification into medical research and service.

In their analysis, they emphasize that since 1989 there was still no healthcare database that would have collected health statistical data about Roma people in Hungary.

To place more weight on this issue, they cite the relevant public policy statute[6] enacted in 2003: "we don't know exactly the relationships between the Roma population and the healthcare system's specific institutions (general practitioners, clinics); and we don't fully know how biased the attitude of healthcare workers towards the Roma is, (and vice versa: what types of fears or biases Roma have towards healthcare institutions or their workers). Based on the data, it is possible to point out the effect that communication issues have in the doctor-patient relationship on judging the incidence of disease. It is an even more significant question that, in light of the data, general practitioners do not know the precise health condition of Roma people, including their mortality rates, hence it is not probable that they would pay greater attention to their Roma patients" (Balázs et al., 2011: 72). Based on this problem, a highly important question was raised in their paper: "What would happen if the attitude of general practitioners changed? What kind of Roma population data would they be able to use?" (ibid.). They posited that epidemiological studies are concerned with the mapping of real health problems of various populations within a society, and at the same time provide data on which primary and secondary preventive programs could be based. The authors argued that if the categorization of Roma identity is recognized as a potentially useful analytic variable for healthcare professionals aiming to meet epidemiological objectives that benefit Roma communities, then the refusal to employ ethnic classification in the collection of health data may represent a missed opportunity. From an anthropological perspective though, while rendering Roma health disparities statistically visible is indeed a necessary step toward addressing structural inequities, the use of ethnic categories in the context of so-called "ethnic diseases" raises significant methodological and epistemological concerns. In particular, medical-genetic interpretations that rely on historically constructed notions of isolation and inbreeding must be critically interrogated, as they risk reifying essentialist understandings of ethnicity and obscuring the socio-political determinants of health.

Methodology

This study adopted a qualitative approach to explore medical geneticists' perspectives on racial and ethnic categorization in clinical settings. Drawing on constructivist grounded theory (Charmaz, 2003, 2006; Clarke, 2005), 34 medical geneticists were interviewed from Hungarian cities: Budapest, Debrecen, Szeged, Pécs, Miskolc, and Győr. Participants were chosen for their clinical experience with both Roma and non-Roma patients and, in some cases, for their involvement in mapping population-specific disorders. The experts who contributed to the research by giving interviews received their medical degrees between the 1960s and 1980s. The main intention was to find experts who are

[6] Parliament declaration on the Epidemiological Program of the Decade of Health (46/2003, IV. 16.).

familiar with the historical background of their disciplines and are themselves widely recognized researchers and clinicians in their work field. With this focus, it was possible to make sure they will be able to reflect on the historical context, in which race and ethnicity as medically relevant identities have started to shape their practices.

Ethical considerations and consent

To ensure participants were fully informed, the study's purpose and objectives were explained, securing consent from each interviewee. No financial incentives or compensation were provided, preserving the voluntary nature of participation.

Participant recruitment and sampling

The study's participant pool was initiated from the website of Semmelweis Medical University (Budapest), where the contact data of researchers who took part in the lecture series that focused on medical genetics was available. Using purposive sampling, medical geneticists with a specific interest in population genetics and the use of race/ethnicity in medicine were identified. Snowball sampling was then employed by asking participants to suggest academically respectable colleagues in this area of study, thereby broadening the reach to include additional experts.

Interview process

Semi-structured interviews were conducted between 2011 and 2015 to allow adaptability in the questioning process, accommodating the evolving nature of qualitative research. Interviews lasting between 60 and 120 minutes were conducted in Hungarian and audio-recorded. Once transcription was complete, only those interview sections were translated[7] by the author of this study that proved relevant after initial coding. To protect the privacy of participants, all identifying information was anonymized in published findings. In the interviews, the questions were articulated around the use of race and/or ethnicity in population genetics and in clinical genetic practice.

Data analysis

The analysis drew on Clarke's (2005) and Charmaz's (2003, 2006) methodologies relying on traditional grounded theory methods. This involved three phases of coding: (1) initial coding, where short active codes were developed that captured incidents described by participants; (2) focused coding, where the short codes were then refined into key themes that were compared across transcripts to reveal recurring patterns; and (3) finally theoretical coding, applied to integrate findings into a cohesive framework for the final analysis. Throughout these stages, memo-writing served as a bridge between coding and synthesis, enhancing analytical clarity (Charmaz, 2003: 261). The analytical

[7] All translations from Hungarian to English are the work of the author of this article.

work was centred around three main problems stemming from the narratives: (1) the articulation of the boundary between Roma and non-Roma people; (2) the medically significant problem of isolates or isolated groups sharing genetic traits that have medical consequences; (3) and lastly, inbreeding and its medical consequences. This method facilitated a nuanced understanding of how various medical realities around racial and ethnic categorizations are produced by the participants during their work. The analysis maps those interpretations that frequently occurred during the data collection process.

Discussion of research results

Making distinctions between the Roma and non-Roma communities of Hungary

In the early works, geneticists were interested in finding out the genetic structures that make Hungarians Hungarian. For this reason, they wished to compare different ethnicities but mainly wanted to follow the lead of physical anthropologists and ethnographers who suggested that people in the Őrség region[8] might be the ancestors of ancient Hungarians, since they had kept their separateness throughout the centuries. This lead eventually failed, and it was concluded that it is not possible to differentiate ethnic Hungarians from other white Europeans – relevant information in the rationalization of disease mapping.

> *In researching the genetic structure of the original Hungarian settlers, the geneticists found that the genetic structure is a mixture. There is no real difference between different ethnicities, once the Jewish and gypsy samples are excluded. They concluded that in Central-Eastern Europe, the present populations are genetically kin populations, which is good for the society because there is no specifically Hungarian genetic disease. It is well known that there are Jewish diseases, gypsy diseases, Celtic, and even French-Canadian diseases, and researchers know about Finnish diseases as well. Basically, from the perspective of genetics, the more mixed a population, the healthier it is.* (PI 20121031)

One of the most easily observable directions in the interview is the distinctiveness of Hungarian Roma and Jewish samples from the majority. The explanation is connected to international literature, implicitly stating that this is an accepted narrative in the international medical genetic discourse. Certain ethnic communities within a society maintain their social isolation; and it is even more understandable when the explanation is that geographical barriers stop people from biologically mixing with

[8] Őrség is a geographical area in South-West Hungary on the border of Slovenia.

other ethnic communities, such as is the case with Finland and the so-called "Finnish diseases". The separation of majority Hungarians from Roma and Jewish Hungarians is primarily explained through historical narratives as we can see in the following excerpt:

> *In Hungary, one can distinguish between three ethnic groups of Roma: Vlachian, Romungro, and Beasi gypsies. It is clearly evident that they joined the Hungarians later, and that they lived in isolation. This is like the Jewish case: firstly, there is a religious law which prescribes that a Jewish woman can only bear a child to a Jewish man; and secondly in 1096 King Szent László sanctioned that Christians can only marry Christians, so he also restricted the choices of Jewish individuals; hence they chose partners from their small communities. As there were not so many individuals in these communities, certain kinds of inbred genetic characteristics evolved. When living in inbred communities, for example where cousin marriages are very frequent, which is typical for both Hungarian gypsies and Jews, a flawed gene from a common ancestor makes it much more probable that these genes resurface. For example, we know that there are Jewish-only diseases – there are seven such diseases – which is interesting because in Israel, all the newborn babies are screened for these diseases. Our research confirmed that there are similar gypsy diseases. Congenital glaucoma, buphthalmos, is such an example; it is much more prevalent in their communities. Of course there are advantages as well: there is no sclerosis-multiplex in gypsies. When we analyzed these gene signals it emerged – it was verifiable – that they came from elsewhere; they lived as isolates, in inbred communities. This was similarly verifiable regarding Jews.* (PI 20121031)

This position, that there are Jewish-only, or Roma-only diseases, underscores the belief in biologically meaningful ethnic/racial differences and recreates the dividing line among racial/ethnic populations. Although it acknowledges that the cultural context played a decisive role in shaping disease frequencies, it glosses over the fact that these communities are not biologically closed reproductive groups; and on the individual level, where the medical diagnosis takes place, this might differ case by case.

The problem of isolated populations

Knowledge about different disease frequencies in populations is produced by the methods of population genetics. The information that is gained through its techniques is applied in two main fields of medicine: in clinical diagnosis and in genetic counselling. The central organizing principle of population genetics is the Hardy-Weinberg equilibrium, which allows researchers to predict the ratio of gene frequencies in different ideal and

real populations. The main benefit of this model is that researchers can predict that disease frequencies will be more or less the same in a given population on condition that (1) it is large and mating is random, (2) there is no observable mutation so allele frequencies remain the same. In other words, there is no selection against any genotype and there is no significant allele frequency that has been contributed by immigrants to the endogenous population (Nussbaum et al., 2007: 192–193). Population geneticists identified three factors that disturb the Hardy-Weinberg equilibrium (ibid.: 195–196): (1) stratification of a population, which means that subgroups of a large population remained genetically distinct from each other during the last centuries (this perspective is applicable to race/ethnicity); (2) assortative mating, meaning that individuals tend to choose partners with similar characteristics (from the perspective of medical genetics this contributes to the suspension of the Hardy-Weinberg law since mating partners possess similar traits); and lastly (3) inbreeding, similarly to the previous two factors, effectuating an increase in autosomal recessive diseases.

In the literature, the medical significance of isolates is explained by examples such as colour-blindness on the Pingelap Atoll. This problem is highly frequent there and the mutation that causes the phenotype can be traced back to a single individual. The history of this mutation is connected to an environmental disaster – a typhoon killed most of the inhabitants on the atoll around 1775 – and from those who survived, one person carried this genotype and passed it on to the next generations. Another similar example is the shared asthma disease of the inhabitants of Tristan da Cunha, an island in the middle of the Atlantic Ocean. It was found in 1961 that more than half of its 300 inhabitants carried the gene for the same asthmatic problem. Researchers comparatively analyzed the samples and found that the mutation can be traced back to the same person (see Raskó, 2015). The latter examples are medically significant in pointing out that genetic mutations accumulate in small populations which are closed to genetic inflow. These problems are relevant in any mainland region as well. One of the classic examples for genetic analysis is the Ellis van Creweld syndrome that was first identified in Amish populations in the United States in the 1960s by Victor McKusick (2000). But there are several other examples of other social isolates: the β-Thalassemia in several at-risk populations in the Mediterranean (Cao, Galanello, 2010) or Tay-Sachs in Ashkenazi Jewish communities in the US (Nussbaum et al., 2007). Such medical genetic knowledge is argued to be significant for Hungarian populations as well. To emphasize its relevance, one of the informants considered Finland and pointed out that because of generational immobility of Hungarians a couple of decades ago, perhaps similar problems could be detected.

> *A good example can be Finland for this question, because around the Arctic Circle obviously there are some villages with a couple thousand inhabitants who inter-married, hence there was no significant mixture. In these cases, genes were accumulated which characterized the populations.*

> *When a genetic disease occurs, its incidence is much higher in such an isolated population than in a population where the choice is wider. However, there are isolates on the mainland as well: it is enough to think about small villages a few decades earlier, whose inhabitants could only reach the next villages to choose a partner. Thus, geography is one but ethnicity is another factor, because there are ethnicities who choose partners only from their own ethnic community.* (PI 20130321)

In this explanation, besides the geographical determining factors, racial/ethnic identification and the internalization of traditional values play a significant role. Although it is widely accepted that members of ethnic communities tend to choose partners from the same ethnicity, it must be noted that these are not only determined by internal forces (such as community tradition, family values, peer pressure) but also external forces such as xenophobia, racism, classism, and linguistic discrimination that play key roles in shaping individual reproductive choices. Although other researchers who were interviewed acknowledge its medical usefulness, not all of them considered the question of closed populations easily explicable. They do think that it is indispensable to make use of this term and its tenets, but on the other hand they also shed light on its obscure nature. It is not possible to take hold of a closed population in a finite manner since they are constantly changing in emergent ways.

> *It is hard to define. In reality, I think, there are no closed populations because external relationships always occur in any community. The question is rather their frequency.* (PI 20130214)

To clarify this point further, it was necessary to inquire about the methods and actual practices whereby geneticists choose homogenous populations, and how difficult it is to find such isolates, especially Roma isolates, in Hungary. To this inquiry the following answer was given by one interviewee:

> *We focused on Vlachian communities, choosing villages where the ratio of the population was more than fifty percent Vlachian gypsy. Which is, to a certain extent, a homogenous population*. (PI 20131119)

The above demonstrates that this endeavour is not easy. It also relates back to PI 20130214's position, in which it is stated that there are actually no homogenous populations. It is rather how strictly researchers interpret homogeneity, and how they construct (or circumscribe) the boundaries of a population, take samples, and find a common shared problem whereby the members of the group are linked together, as is evident from another excerpt:

> *When individuals choose partners from within the same group, there is no inflow and outflow of genes. Hence, on a given genetic trait, everybody is in genetic kinship relation with everybody else*. (PI 20100527)

In the explanation, it is glossed over that inbreeding is similar in effect to consanguineous marriages but not equal to this cultural custom, as its biological effects are larger in scale. Inbreeding is rather the result of geographical isolation from the majority population and hence the inflow and outflow of genes is minimal – the inhabitants of the Pingelap Atoll, mentioned above, is a classic example from the literature, where the population of the island lived in significant isolation for an extensive time period. In Roma communities, none of the above discussed factors – geographic isolation, social exclusion, and endogamous practices – are fully applicable regarding their reproductive processes.

Inbreeding and the problem of the founder effect

It is accepted within the field of human genetics that on a theoretical level, humans are all in consanguineous relationship with one another since it is possible to trace back our ancestors to common progenitors; but close consanguinity is an important factor in determining the occurrence of medical problems. Taking this standpoint into account, medical geneticists consider the problem of inbreeding to be relevant only in groups where closely related consanguineous marriages occur. Knowledge about inbred populations is important in calculating the F coefficient factor that helps in the prognosis of the occurrence of any genetic effect that has already accumulated in the studied population (Tóth, Hegyesi, 2007: 144–145). The F inbreeding coefficient factor is the half of a first-level familial relationship that means fifty percent genetic similarity, and in the case of inbreeding it means twenty-five percent genetic similarity. This shows why it is hard to talk about genetically homogenous populations, on the one hand, but it also shows that there are populations where genetic similarity among members on given traits can lead to the accumulation of genetic disorders.

> *It is very difficult to talk about completely homogenous groups, it is only possible to talk about approximately homogenous groups. It was revealed by molecular genetics methods that even if I should take a group to be homogenous according to their diagnosis, they won't be homogenous genetically. This means that the same disease can be generated by different mutations, different genes, or even different mutations of genes. There are inbred groups whose members marry each other. In these cases, certain rare genes accumulate. These founder effects influence the genetic characteristic of a population or a race in a given geographical area, because it is obviously true that the Roma population is, decisively, endogamous.*

> *But it is not the fault of the Roma, similarly as it is not the fault of the Ashkenazi Jews that Tay-Sachs is more frequent in their communities*. (PI 20140307)

In population genetic studies, founder mutations are significant factors divided into two kinds. One kind occurs only in the same population or same group of people, and there is another kind that can be traced back to its origin in two different populations. An example of this might be a mutation which was found on the Iberian Peninsula, in a Roma individual in South Portugal; at the same time this type of mutation was found in Spain in a non-Roma individual, yet these two individuals had nothing in common and researchers could prove through the analysis of their respective BCKDHA[9] genes that they did not share common roots – they developed the mutation in separate historical and geographical contexts (Quental et al., 2009). There are individuals described by these researchers who do not have anything in common. One identifies ethnically to be Roma and the other is non-Roma; they both carry the c.117delC-α mutation, but it has developed separately in space and time. These can be termed parallel founder mutations. The concept entails that the same mutation can develop as a response to environmental factors and then can be passed on to descendants, and it means that researchers can retrospectively trace back the same genetic disorder to separate groups of people, and to ethnically and geographically independent individuals. A similar example was given in one of the interviews:

> *In Szeged a similar mutation was described. Researchers found a mutation for a very rare multiplex dermatological disease – tumours appear in diverse locations on the patients' bodies – and within the same gene, the same type of mutation was described in England, in English families; thus it is impossible that one founder mutation spread from here to there or vice versa. This is a very rare event, but it can happen independently. On the other hand, there are founder mutations which are typical of populations. Let's take an example: cystic fibrosis, where the most common mutation is the lack of phenylalanine in an enzyme, in a protein. This mutation is prevalent in 85 percent of Danish patients, whilst in Hungarian and other South European patients this type is present in only 40 percent. In these populations, there are other types as well which cause the disease.* (PI 20130328)

[9] Branched chain keto acid dehydrogenase gene, or the BCKDHA enzyme complex is responsible for one step in the normal process of breaking down three protein building blocks, it is found on chromosome 19 (MedlinePlus, 2020).

In the above example, the rare dermatological disorder supports the idea of parallel founder mutations. On an individual level, these are already recorded multiple times, but on a population level, statistically, there are mutations which are epidemiologically meaningful. Cystic fibrosis is used as a genetic disorder that is ethnically/racially typical of certain populations, but even in this case, it is not excluded by the interviewee that populations whose social self-identification varies can carry the same mutation (see for example, Roberts, 2011: 99; where she discusses the CF case of 4-year-old African-American girl, when CF is predominantly viewed as a Caucasian/White disorder). Hence some population geneticists (see Raskó, 2022: 24) caution using race and ethnicity, because these terms are imprecisely defined cultural categories and thus their use in genetic research could yield imprecise results.

Conclusion

The use of genetics to address population-level health disparities has increasingly been framed as an ethical necessity, particularly as researchers turn their attention to ethnically identified populations with the aim of promoting health equity. This is significant for two reasons: (1) this orientation reflects a growing awareness that health inequalities are not solely biomedical phenomena but are fundamentally shaped by intersecting structures of economic marginalization and racialization; (2) it also shows that the medical genetic discourse operates with anthropological categories – such as ethnicity – that cannot be equated with biological phenomena. Nevertheless, within this context, genetics becomes a contested yet potentially productive site for negotiating justice in public health.

In the paper, this is demonstrated through the case of Hungary and its renewed focus on minority health. Hungary's 2004 accession to the European Union marked a significant political shift, bringing minority rights, especially those of the Roma population, into sharper institutional focus. National and EU-driven efforts sought to improve housing, employment, and access to healthcare for Roma communities. Medical geneticists participating in these initiatives undertook the task of mapping the distribution of genetic disorders and health vulnerabilities perceived to be concentrated among Roma populations. Drawing on both population genetics and empirical fieldwork, researchers pointed to phenomena such as geographic and social isolation and endogamy – conditions understood as shaped by both cultural practices and the structural violence of systemic exclusion – as factors influencing reproductive processes and health outcomes, such as the accumulation of genetic mutations, and thus the resulting health disorders.

The empirical work that this analysis relies on, semi-structured interviews with geneticists, revealed ambivalence about the use of ethnicity as a category of analysis. Interviewing is a valuable tool because it enables social scientists to capture the continuity of a scientific

idea through personal accounts. Predominantly, geneticists were in agreement with the use of ethnic identity for data collection purposes, but there were participants who stated their concern by claiming that these identity categories are ill-defined for genetic use. It must be emphasized that there was no reflection in the data on the historicity of these categories. The knowledge articulated and produced in a historically different time was carried on by geneticists and thus informs their practices in a discourse that has changed radically in terms of both biotechnology and culture. Although it is true that certain mutations may appear more frequently in socially isolated groups, through the analysis, it became clear that foregrounding ethnicity risks reinforcing essentialist understandings of biological difference. From an anthropological and bioethical standpoint, such practices demand critical scrutiny: the use of ethnic classifications can obscure the historical and political forces that shape health outcomes and may inadvertently re-inscribe racialized imaginaries under the guise of genetic objectivity.

Ethically informed public health genomics must therefore resist the reification of "ethnic diseases" and instead engage in nuanced, community-based approaches that centre lived experience and social context. Monitoring the prevalence of specific genetic conditions across populations can be a valuable tool – particularly when it facilitates informed consent, autonomy, and participation among historically marginalized groups. However, such efforts must be accompanied by strategies that challenge structural inequities and ensure that genetic knowledge is communicated in ways that are both accessible and culturally respectful. The integration of genetic research into public health must be socially reflexive, ethically grounded, and attuned to the complex entanglements of race, identity, and power.

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Research data statement

The author states that the article is based on ethnographic research materials that are not classified as research data. All additional information concerning the ethnographic research materials are available on reasonable request with the author.

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Kako medicinski genetiki interpretirajo genetske motnje v romskih skupnostih postsocialistične Madžarske

Članek kritično obravnava uporabo rasnih in etničnih kategorij v medicinski genetiki v postsocialistični Madžarski. Osredinja se na apliciranje biološke kategorije rase/etničnosti na romsko populacijo. Preučuje klasifikacijo in interpretacijo genetskih razlik v javnem zdravstvenem raziskovanju in klinični praksi v spreminjajočih se družbenih in kulturnih kontekstih. Raziskuje, kako se je medicinska genetika uporabljala za pojasnjevanje in obravnavo bioloških razlik ob vztrajni zdravstveni neenakosti med romskimi in neromskimi Madžari.

Članek se začne s kratkim zgodovinskim pregledom razvoja medicinske genetike na Madžarskem po drugi svetovni vojni in njenega postopnega uporabljanja v zdravstvenih ukrepih na populacijski ravni. Od osemdesetih let 20. stoletja so primerjalne genetsko-genomske študije, osredotočene na romske skupnosti, odkrivale višjo prevalenco nekaterih genetskih mutacij v teh skupnostih, in sicer v povezavi z boleznimi, kot so bolezni srca in ožilja ter prirojena glavkom in laktozna intoleranca. Te ugotovitve so bile pogosto pripisane trajnim vzorcem družbene in geografske izolacije, endogamnim porokam in družbeni izključenosti. Čeprav so te študije prinesle epidemiološko dragocene podatke, so znanstveniki opozarjali, da zdravstvenih neenakosti ne smemo pripisovati izključno biološkim dejavnikom, in poudarili pomembno vlogo socialne izključenosti, revščine in sistemske diskriminacije.

Proces pristopa Madžarske k Evropski uniji je z zahtevami po izboljšanju manjšinskih pravic in zdravstvenih rezultatov dodatno spodbudil epidemiološke študije o romski populaciji. Raziskovalci so dokumentirali očitne zdravstvene pomanjkljivosti, vključno z višjo obolevnostjo in umrljivostjo, slabšim dostopom do zdravstvenega varstva in neugodnimi življenjskimi pogoji v romskih naseljih. Trdili so, da bi učinkoviti programi javnega zdravja morali vključevati več sektorjev, ne le zdravstva, da bi morali obravnavati strukturne neenakosti, ki so

podlaga za slabe zdravstvene rezultate, ter vključevati kulturno občutljive in skupnostno-odzivne strategije.

Pričujoča študija temelji na analizi polstrukturiranih intervjujev s 34 madžarskimi medicinskimi genetiki, s katerimi je bil vzpostavljen neposredni stik prek njihovih institucionalnih naslovov. Intervjuvanci so bili izbrani na podlagi njihovih strokovnih dosežkov in nadalje z uporabo metode snežne kepe glede na priporočila kolegov. Vsi so strokovnjaki z večdesetletnimi kliničnimi in raziskovalnimi izkušnjami. Z analizo so bile opredeljene tri osrednje teme: (1) konstrukcija razmejevanja med romskimi in neromskimi skupinami; (2) pomen populacijskih izolatov v medicini; (3) razmerje med endogamijo, učinki ustanoviteljev in genetskimi motnjami. Genetiki so Rome pogosto uvrščali v ločeno genetsko skupino in jih primerjali z drugimi izoliranimi populacijami, kot so Aškenazi in finske skupnosti za arktičnim krogom.

Analiza postavlja pod vprašaj pojem biološke homogenosti etnične/rasne populacije ter kaže na kompleksnost in dvoumnost opredelitve »izoliranih populacij« za genetske analize. Čeprav geografska in socialna endogamija lahko prispevata k razširjenosti nekaterih genetskih bolezni, pa slednje vedno oblikujejo prepletajoči se zgodovinski, kulturni in družbeno-ekonomski dejavniki. Poleg tega zanašanje na stroge etnične klasifikacije v genetskih raziskavah tvega prekomerno poenostavitev medicinske realnosti in ohranjanje stereotipov.