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Implications of Racial/Ethnic Classification in the Hungarian Post-Genomic Medical Discourse

Racial/ethnic categorization in medicine presents challenges for clinicians and patients alike. Challenges arise because racial/ethnic identities do not match with objective biological traits, and at the same time, these identities do have medical consequences in a racially and ethnically stratified society. Three major epistemological approaches – biological realism, eliminativism, and constructivism – dominate scientific theorization on the consequences of racial/ethnic categorization in medicine. In this paper, I present a case study of Hungarian medical genetic discourse that focuses on the possible applications of race/ethnicity regarding Roma and non-Roma patients. In applying the methods of constructivist grounded theory, I recorded and analysed 34 expert interviews with human geneticists between 2011 and 2015. In this paper, I argue that the constructivist understanding of medical diagnoses must be complemented with materialist sensitivity, thus making sense of the contingent nature of race/ethnicity as factors that contribute to medical understanding.

Keywords: medical genetics, health equality, race, ethnicity, Roma.

Posledice rasne oz. etnične klasifikacije v madžarskem postgenomskem medicinskem diskurzu

Rasna oz. etnična kategorizacija v medicini pomeni izziv tako za zdravnike kot za paciente. Težave nastajajo, ker se rasne oz. etnične identitete ne ujemajo z objektivnimi biološkimi lastnostmi, hkrati pa imajo v rasno in etnično razslojeni družbi določene zdravstvene posledice. V znanstvenih teorijah o posledicah rasne oz. etnične kategorizacije v medicini izstopajo trije epistemološki pristopi: biološki realizem, eliminativizem in konstruktivizem. Prispevek predstavlja študijo primera madžarskega medicinskega diskurza na področju genetike, ki se osredotoča na upoštevanje rasne oz. etnične klasifikacije romskih in neromskih bolnikov. Ob uporabi metod konstruktivistično utemeljene teorije je bilo med letoma 2011 in 2015 posnetih in analiziranih 34 intervjujev s strokovnjaki za človeško genetiko. Prispevek zagovarja stališče, da je treba konstruktivistično razumevanje diagnoz dopolniti z materialistično občutljivostjo in tako rasno oz. etnično klasifikacijo upoštevati kot dejavnika, ki prispevata k razumevanju zdravstvenih razlik med prebivalstvom.

Ključne besede: *medicinska genetika, enakost na področju zdravja, rasna in etnična pripadnost, Romi.*

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1. Introduction

In the 1960s, it was found that members of Hungarian Roma communities were in such an economically marginalized situation and subject to further racial discrimination that their health standards were far below the national average. Since that period, significant medical and sociological research has been aimed at working out ways to ameliorate their living conditions, but because of systematic discrimination the research results were not put to use efficiently. The possibility of joining the European Union opened up for Hungary during the 1990s, and in 2004 the country successfully met the required criteria. As part of the requirements, it was mandatory to develop the rights and protection of minorities in Hungary (Kósa et al. 2002). The health protection of Roma was one of the key priorities, and some of the most important goals sociologists and healthcare professionals identified were the improvement of housing possibilities, access to employment, and access to healthcare services. Medical geneticists contributed to this discussion with their own expertise by claiming that to improve the health standards of Roma it is important to map the most prevalent inheritable genetic disorders in their communities, as well as the epidemiologically relevant genetic markers that cause them to be at risk for diseases. One of the conclusions that was drawn from the early epidemiologically important genetic studies is the fact that ethnic identity is only important because it meshes biological causes with social factors and is thus useful for better diagnosis and better healthcare service. Geneticists acknowledged that biological differences can be important for both patients and clinicians, and the socially marginalized situation of Roma should be considered during medical interviews. The everyday racism that they endure during healthcare services must also be taken into account when healthcare statistics are evaluated. Thus, in this paper, I will contribute to the understanding of race/ethnicity in the Hungarian medical genetic context.

In the international literature that deals with the applications of genetic results to improve health disparities it is possible to delineate two directions. The first approach considers connecting genetic patterns to race/ethnicity to be a scientific error, while the second approach embraces racial/ethnic classification of biological materials because it is argued that this technique speeds up and therefore helps medical diagnosis. Starting from these grounds, I rely on critical constructivist approaches that can accommodate the context dependence of racial/ethnic identities in medicine, but at the same time give a critical view on the essentializing tendencies of the genetic discourse and its possible drawbacks. Thus, in this paper I will briefly review the major framework regarding the conceptualization of race and connect it to contemporary epistemological discussions. My aim is to provide theoretical underpinnings to my argument that an essentialist understanding of race in the medical sciences could prove to be very dangerous in a society that structurally marginalizes and discriminates against

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racial minorities. In order to support my claim I will analyse the arguments that complicate the use of racial/ethnic classification in medical genetics. We live in a racially stratified society, and currently it is not possible and not desirable to get rid of racial categorization because it would seriously damage medical understanding and thus equality in healthcare (Rose 2007). Another reason is connected to this last problem; if a society wants to offer identical treatment[1] to all its citizens, it must take into account various identity categories, from class and gender to race/ethnicity (Risch et al. 2002). These issues are interconnected, and the suggested avenue is to tackle health problems with the present vocabulary and with the already established racial framework. However, medical geneticists do not accept racial/ethnic categorization in medicine unanimously and uncritically, and they do question the essential nature of race (Feldman & Lewontin 2008). There are also geneticists who suggest that a focus on individual genetic make-up will provide better information for establishing a diagnosis and thus providing identical healthcare, but at the same time race and ethnicity cannot be discarded just yet.

2. Methodology

I designed a qualitative study in which I interviewed 34 medical and clinical geneticists based in the cities of Budapest, Debrecen, Szeged, Pécs, Miskolc, and Győr. All of them had clinical experience with Roma and non-Roma patients, and some of them had even taken part in mapping population specific disorders. I executed the analysis of the collected interview materials and reported my findings according to the criteria of constructivist grounded theory (Charmaz 2003; 2006; Clarke 2005).

2.1. Ethics, Consent, and Permissions

This research was approved by the Central European University. I obtained consent verbally from all of the participants and I provided information about the aims of my research to everyone. I did not give any compensation of incentives to the interviewees.

2.2. Sampling and Data

I began collecting interviews through a series of lectures that were organized at the Semmelweis Medical University in Budapest. I collected the contact information of geneticists who had a focus on racial/ethnic issues in medicine. During the interviews, I asked for the contact information of other geneticists in order to contact those whose work in the field was valued by their colleagues. I chose to do semi-structured interviews because this method allows for constant

adjustments during the research process. I conducted the interviews between 2011 and 2015; the interviews lasted between 60 and 120 minutes. All of the interviews were conducted in Hungarian and were audio-recorded. Subsequently, I transcribed all of the interviews in Hungarian and I translated only the parts of the interviews that became relevant after the coding process. In the analysis, I coded all of my interviews as Personal Interview (PI) followed by the date it took place, in order to keep the professional identities of my interviewees safe.

2.3. Qualitative Analysis

In order to analyse my material I relied on the guidelines of Adele E. Clarke (2005) and Kathy Charmaz (2003; 2006). As Clarke suggests in her work, I used situational maps to complement the coding and memo-writing strategies traditionally used by grounded theorists. In my work, I went through three coding phases while I analysed the transcripts. At first my aim was to develop active short codes to capture the incidents told by my interviewees. Later, I developed focused codes that helped to clarify the most important and recurring themes that I eventually used in a comparative manner. In the third phase, I developed theoretical codes that became part of my final analysis. Charmaz states (2003, 261) that in order to provide a focused and sharp analysis researchers must employ the method of memo-writing as an intermediate step between coding and the final analysis. This served the purpose of elaborating on the analytical insights. With these methods, I mapped the elements at different locations, in different narratives that played a role in the production of various medical realities regarding racial/ethnic categorization.

3. Conceptualization of Race

There are three major approaches to theorizing race in philosophy of science: these are called biological realism, constructivism, and eliminativism (Haslanger 2008; Ludwig 2017). The most important distinction among these approaches is how theorists who work within these paradigms regard race: eliminativists, for example, consider race to be something that is non-existent, therefore it is counterproductive to discuss its relevance in any medical discourse. Their approach was criticized by many scholars for turning a blind eye towards skin colour; this approach, they claimed, would not solve the very real problems of everyday racism that people face in various societies. Social constructivists think that race is real in the sense that it is a social construct based on material differences, and although it is imbued with different values in different social and historical contexts, it has very real consequences for people experiencing discrimination or privilege because of a racial discourse. Biological realism holds that there are races that correspond to biological differences. This strand of theorizing has

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become revitalized as a result of molecular genetic studies. After the completion of Human Genome Project (Fridovich-Keil, Invalid Date), it is argued that within the differences mapped it is possible to find the reason for racial diversity that corresponds to racial subgroups which in turn, if recognized, contribute to health equality. In the following, I will overview very briefly the main shifts that took place in scientific thought about race.

During the Enlightenment, the work of Johann Friedrich Blumenbach entitled On the Innate Variety of Mankind, published in 1775, defined the direction of racial studies for the coming 150 years and it had a long-lasting impact on the twentieth century (Smith 2015, 253; Raskó 2015, 147). Blumenbach described four geographical varieties in his 1775 edition, then changed it slightly to five varieties in 1781, when he reworked his thesis, and finally concluded with five generic varieties classified as Caucasians, Mongolians, Ethiopians, Americans, and Malays in the final re-edition of his thesis in 1795 (Bhopal 2007, 1308). Justin E. H. Smith argues in his book that although Blumenbach was hesitant to claim that there is a biological reality according to which clear racial divisions can be made, he still maintained, through statistical measurements of human skulls that it is legitimate to support the racial classification of peoples into the racial taxonomic system that he proposed (Smith 2015, 259–260). This is why Smith (2015, 259) claims that Blumenbach was a "statistical racial realist", and his work lent itself easily to succeeding theorists whose aim was to establish accounts for the biological reality of race.

The view that there are essential racial types started to become more and more incompatible with developments in biological research. First Charles Darwin's evolutionary theory questioned whether it was possible to pinpoint toward racial types, and then later Mendelian genetics provided rational arguments in favour of abandoning the essentialist approach. By the early twentieth century, the essentialist concept of race was supplanted by a geographical concept that divided races into subdivisions according to their geographic origin. For example, William Z. Ripley, in his work published in 1899, divided the people of Europe according to three different geographical regions: Alpine, Mediterranean, and Nordic (Marks 2008, 22–23). A series of works appeared in this paradigm from which perhaps the work of Theodozius Dobzhansky was the most significant in 1937, titled Genetics and the Origin of Species (Dobzhansky 1982). Although the Second World War had an important impact on the reconceptualization of race, it is needless to say that the Unesco Statement on the Nature of Race and Racial Differences (UNESCO 1952) described three major races – European, Asian, and African – and an unspecified number of subdivisions, which still adhered to the geographical type paradigm. This understanding dominated scientific thinking until the 1960s.

The civil rights movement in the 1960s pushed the theorization of race in a new direction. In 1962 Frank Livingstone argued that it is best to understand

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racial difference in terms of clines, which are "geographical gradients of features in natural populations" (Livingstone 1962, 279, cited in Marks 2008, 24). In a similar manner, Richard Lewontin (1972, cited in Marks 2008, 24) quantitatively compared populations from a genetic perspective. He concluded that intra-group differences are larger than in-between group differences, thus deconstructing the race-as-geographical-type concept. As noted by Marks (2008) Lewontin still holds that despite the developments in molecular genetic studies, abandonment of the biological race concept in medical research was well-supported. Marcus Feldman and Richard Lewontin (2008, 90) state that if medical professionals understand race/ethnicity as a social construct, they can gain important knowledge about the social-environmental factors that determine the health status of patients. In particular, knowledge about race/ethnicity can be informative in order to fight discrimination on the micro-, meso-, and macro-levels of society.

The third and perhaps most radical shift occurred during the 1990s, when researchers suggested that roughly 7 % of our molecular genetic difference could be racially relevant (cf. Sesardic 2010, 148–149). They argued that perhaps a more thorough understanding of this part of the human genome could contribute to making sense of human racial differences. The process through which race is projected onto the molecular level is called the molecularization of race (Kahn 2008; 2012; Fullwiley 2008; Duster 2006). It would be a misunderstanding to see the present process of racialization as unreflective of racism – it is the opposite; scientists apply race in genomics as a biosocial reality but at the same time they are working towards the genetic explanations and rebuttals of any kind of racism; this is called the biosocial paradox of race (Bliss 2011, 1019). The primary aim of racial classification that is advanced by scientists who work with genomic-level data is to overcome health inequality that is caused by systematic racism.

4. Arguments for Classifying Genetic Material According to Racial/Ethnic Identity

Although scientists widely acknowledged that it is hard to define what race/ethnicity means in biomedical research, at the beginning of the twenty-first century, Nikolas Rose argued that it is inescapable for researchers working in the field of biomedicine to categorize through racial/ethnic identity (Rose 2007, 172). Skin color is one of the markers (besides hair texture, and bone structure) that define racial belonging following the work of modern naturalists. Although numerous scholars point out that skin color or bone structure cannot be used to group people together for medical genetic purposes because it is a very arbitrary marker (Gould 1981), it is still viewed by others as a medically valid factor of cat-

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egorization. It is claimed that folk racial/ethnic categorization is useful because it shows that members of such groups share medically relevant genetic histories. The following quote from an interview emphasizes that skin colour indicates deeper medical relevance than superficial racial resemblance:

> Skin colour is not only a superficial marker that accidentally helps, obviously, Caucasians or white people resemble each other more than white people resemble black people. But within the white population, or black or Asian, a Japanese is utterly different – not fundamentally of course – but represents a significant genetic difference in contrast to an Indian or Nepalese. This is because of the migratory routes of different populations. Those who lived together for a long enough time developed genetically unique characteristics (PI 20130311).

I do not interpret the above claim to be in tension with the medical genetic knowledge that within-group differences can be larger than differences between groups. This position rather entails that, although there are no fundamental differences among human beings, there is medically useful information that can be teased out with the use of racial/ethnic categories.

In the Hungarian social context, numerous racial and ethnic groups have been analysed by geneticists (cf. Béres 2003). Within these populations, in most interviews Caucasians were viewed as the medically most significant group, and besides them Roma and Jewish groups were highlighted. In the excerpt below, it is noted that superficial racial/ethnic characteristics are not helpful in directing the caregiving process because in some cases they are not visible, or they do not diverge from the dominant look of individuals in the country. In these cases, self-defined ethnicity is understood to be crucial for precise diagnosis.

> In Hungary the most significant ethnic group is the Caucasian, and there are a few Roma and Jewish groups. But I only inquire about ethnicity in the case of concrete diseases, when those are more prevalent in certain ethnic groups; because I can't always recognize from the appearance of the patients which race they belong to (PI 20121207).

To put it differently, medical genetic knowledge cannot be applied by clinicians in certain cases when there is no knowledge about ethnic belonging and only a medical hypothesis exists about a possible diagnosis based on the medical interview and examination of the patient. This medical hypothesis rests on the previously existing racial stratification of people. Thus, self-defined race/ethnicity is viewed as a very good marker for the medical understanding of the health status of the patient (Risch et al. 2002). Risch and his colleagues argue that most of the research that addressed the genetic and epidemiological validity of race is not objective, hence these studies, they claim, cannot contribute to the scientifically grounded reformation of healthcare. They acknowledge that because of the past

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and present racial/ethnic discourse of the United States, racial/ethnic minorities are disadvantaged and their disadvantageous social position largely defines their health standards; they say that besides the genetic structure – about which they claim that racial/ethnic communities resembling their own members more than non-members is empirically provable – it is needless to take into account the environmental factors (overall health, education, lifestyle, support system, and socioeconomic status); together these define the genetically understood health needs of an individual. Therefore, they claim that it is vital to consider the self-defined racial/ethnic identity of patients in order to provide them with identical – though not equal – healthcare, because human beings are different.

In the case of Hungarian ethnic groups, for example, the homogenous racial/ethnic identity category of Caucasian does not address certain problems. The distribution of the cystic fibrosis gene ΔF 508 is such an example. In a study conducted by scientists in Debrecen, the authors claim that the F508del mutation is the most significant variant in Hungary with 61.2 % and it shows a decreasing north-to-south gradient in its distribution in the country (Ivády et al. 2015, 50). This means that other mutations are also prevalent in the population, and the diagnoses would be helped with further knowledge of the geographical ancestry of the individual.

In addition to this, one must note that population genetic studies that aim at mapping the diversity of certain diseases within various populations is very useful on a local level to shorten the time that is needed for diagnosing a given problem. Let us look at the following argumentation that points out differences between Roma, non-Roma, Hungarian, and non-Hungarian Caucasian populations to the West of Hungary, regarding the above detailed cystic fibrosis disease. What I want to emphasize from the quote below is the distinctions that the author makes regarding ethnic boundaries and the uncertain claim about the prevalence of the disease in Roma communities.

> In our work, we noted that a high percentage of our patients have a certain mutation which is very rare in the international literature. This could mean that this mutation is a highly frequent one in Hungary, but it could equally mean that this is a mutation which is frequent in a Hungarian gypsy minority population – because we don't know the ethnic background of our patients. However, we have data about ethnic Hungarian patients regarding cystic fibrosis. We know that these are from ethnic Hungarians because the clinicians sent the samples that way. By the way, cystic fibrosis is not really prevalent in gypsy populations. So, we have two mutations, which are practically non-existent to the West of Hungary. I would say, where there are no Slavic populations. This is beautiful evidence of the mixture of ethnicities. This mutation occurs in 5 % of our patients, and this is zero in England, in Sweden, and Spain. And this is very important because it can help us to offer fast and cheap diagnoses for our Hungarian patients. Because we know that this exists in Hungarian patients, and in a diagnostic kit, which is composed in England – and let's say we use that – that is not included because it is not typical in that population (PI 20140210B).

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The claim regarding the prevalence of two genetic forms of cystic fibrosis in Hungarian populations suggests that medically relevant ethnic boundaries cross state boundaries. A medically valid argument was put forward by the interviewee to create disease diagnostic kits that are relevant for the Hungarian and neighbouring populations' genetic make-up, since it would make diagnoses faster and much more precise. The argument supports, in my view, the idea of mapping the variations of different mutations for the same genetic disease in local populations in order to design diagnostic kits to treat patients effectively.

This argument stands for members of the Roma and Jewish groups as well, since their racial/ethnic diversity is also important when discussing the medical relevance of sharing certain problems. The dominant approach in researching racial/ethnic populations is connected to the works of Cavalli-Sforza (2000), who suggested analysing genetic data that is collected from communities defined by their shared geographical location, cultural behaviour, and linguistic practices. He argued that these factors determine the reproductive practices of community members and, hence, the gene-flow within the population. Regarding Roma diversity, three main ethnic groups reside in Hungary; these are the Vlachian, Romungro, and Beasi communities scattered across the country. These communities came to Hungary on various migratory routes, and they differ from each other culturally and linguistically. In addition to this, they are located in different geographical regions of the country. An example that helps to elaborate my point is the Beasi community: they came to Hungary from two directions: (1) from the south, particularly from Croatian-Slovenian regions and (2) from the east, from Romanian territories. Their cultural customs were different, they spoke different languages, and they settled in different parts of Hungary. The Beasi Roma who came from Croatian regions mostly settled in Baranya and Somogy counties, while those who came from Romania first settled in Szabolcs and Szatmár counties and then moved to the Tiszafüred region (Kemény 2005, 50–51). István Kemény, through accepting Katalin Kovalcsik's differentiation, identifies three ethnic groups within the Beasi ethnicity. These are the Mucsán, Argyelán, and Ticsán communities. Members of the Mucsán group live around the Hungarian-Croatian border, and their linguistic dialect still uses Croatian words. Members of Argyelán group speak a Transylvanian dialect (called Bánátian), and they also live in Baranya and Somogy counties, while the Ticsán communities came to Hungary through Szabolcs and Szatmár counties from Romania, and they live around Tiszafüred nowadays. This anthropological differentiation suggests that medical genetic problems could be very different for the members of the Beasi Roma communities living at a significant distance from each other and possibly mixing with non-Beasi Roma communities; but it still supports the idea that self-identified and precisely used identity categories could be medically beneficial.

If we choose to analyze concrete medical situations, wherein a quick and precise response is of crucial importance, the argument to use ethnic/racial markers

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in the sampling and diagnostic process seems to further strengthen this position. In the case of bone marrow transplantation, for example, racial or ethnic ancestry is suggested to be valuable information. The interviewee quoted below is on the same theoretical footing as Neil Risch and his colleagues (2002) referred to above: in order to help patients to have an equally positive outcome one has to take into account the ethnic diversity of the population.

> Gypsies are different genetically from the surrounding white Hungarian population, and in order to help them, we must analyse precisely their genetic background. For example, it is necessary to map gypsy bone marrow donors, because gypsy and white Hungarians are so different immunologically from gypsies that they cannot get bone marrow transplantation from Hungarians because they would die. In order to be able to treat them properly because of their increased risks we must do these genetic assessments. There is no discrimination in this (PI 20131119).

Here, the primary racial/ethnic differentiation happens through skin colour, and gypsy people are viewed as being isolated, surrounded by the majority white population without any intermixture.[2] This explanation stems from the fact that because gene-flow does not occur significantly, the biological difference regarding bone-marrow structure between a Roma and a non-Roma patient is significant enough to cause the death of the recipient of the transplant. In this case, because there are fewer bone-marrow donors among Roma individuals than among non-Roma, in order to help Roma patients who are waiting for bone-marrow transplantation and to shorten the waiting period, it is medically useful to create a bone-marrow donor bank in which the donated bone-marrow is from Roma people. This practice aims to counter unequal care. Another interviewee further argues that transplantation donors must be identical in the relevant genetic markers, otherwise the transplant will probably be rejected by the recipient's body. Race/ethnicity helps medical professionals to find acceptable donors from a more reliable pool of sources. This is because of the unique mutational events (founder effects) that took place in the bodies of people who belong to the same ethnic group, since they lived and travelled together across the same geographical landscapes.

> In the case of bone marrow transplantation, donors mustn't be only approximately identical in order to be accepted by the recipient's body; it must match a lot of genetic details. This means that there may be genetic characteristics in the Roma population for which it is better for a Roma person to receive the transplant from another Roma. It will be identical with a higher safety margin. With this approach we have higher probability rates because of the founder-effects. In other words, if we need a donor, we had better look for the donor in the same ethnic community because this way we have a better chance of finding an identical match in a shorter time period. It is not possible to exclude the chance of finding a donor from the Caucasian population but we would need to analyse many more samples (PI 20140307).

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However, when speaking about ethnic/racial boundaries, it is important to note that this standpoint does not exclude the possibility of finding a bone-marrow donor with identical markers from a different racial/ethnic community – it is only assumed that it would take more time to run into an exact match. This position does not create biologically grounded and divided races or ethnicities; according to this direction, it is possible to imagine that people who have been living in the same geographical area, who have similar dietary habits, but perhaps who have rarely chosen their reproductive partners from another ethnic community, would still be a match for bone-marrow transplantation for a racial/ethnic other.

Race or ethnicity in itself is not a sufficiently precise marker to draw any medical consequences. The following quote from one of my interviewees suggests a limited usefulness: genetic studies on ethnic/racial ancestry would suggest the use of these markers in medicine but with a restriction that would mandate the inclusion of geographical ancestry. This counters the view that skin colour is not a superficial marker; it rather states that because skin colour variation is the result of multiple genomic combinations, it is of no use for precise diagnosis in medicine.

> [A]s I said, skin colour, and other superficial characteristics, are defined by multiple genes, and this disease is also defined by multiple genes. From a medical perspective, it is important to know the ancestry of a human community. I think it is important to do population genetic studies that can result in the ascertainment of disease susceptibility that is higher in a given geographically defined population than in another one (PI 20130328).

In this sense, skin colour is understood to be superficial, which means that population genetic studies are perhaps designed in a manner wherein race and ethnicity is considered, but without any information on geographical ancestry, disease susceptibility cannot be adequately defined. This approach entails that perhaps skin colour on the molar level acts as a dividing factor, thus we need molecular level information in order to provide medically precise diagnoses for ethnically different patients with the same genetic disease.

5. Difficulties of Racial Classification in a Clinical Environment

In the early 2000s as the Human Genome Project approached its final years, during a conference on June 26, 2000, President Bill Clinton evaluated the results and placed emphasis on the finding that humans share 99.9 % of their genome which renders racial differentiation genetically meaningless (Bliss 2012). This politically significant position was supported by the human geneticist Craig Ven-

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ter, who said that they "have shown that the concept of race has no biological basis", and only one year later, Francis S. Collins further emphasized that "those who wish to draw precise racial boundaries around certain groups will not be able to use science as a legitimate justification" (Bliss 2012, 1). Catherine Bliss, who is a sociologist of science, claims that it is observable globally that geneticists are trying hard to give new meaning to the concept of race on the genomic level. This racial turn in genomic thinking about the concept took place in the second half of the first decade of the new millennium: scientists are busy looking for medically applicable data regarding someone's racial identity.

Genetic tests are useful on different levels and for different purposes. They are useful, for example, in gaining knowledge about an individual's health prospects, they are useful for couples who want to know genetic data about their reproductive capacities, they are also useful for individuals to get medically relevant information about their newborn child, and genetic tests are useful on an epidemiological level to manage the healthcare of the population. The primary direction in which researchers began to work was toward epidemiological screening. Population screenings were first introduced in the United States during the 1960s. These first screenings were phenylketonuria (PKU) screenings, which were introduced over the course of the following ten to twenty years in countries that had systematically organized healthcare systems (Kosztolányi 2013, 70–77). This was the case with Hungary as well, where they were introduced in the 1980s, and since then, because of rapid biotechnological developments, compulsory screenings were supplemented with 25 other genetic problems and have been tested for since 2007, following the decree of the Healthcare Ministry 44/2007 (EüM Rendelet ...).

There are arguments put forward by researchers for designing screening protocols that target ethnic communities. Perhaps it is sufficient for this argument to name two racial/ethnic target communities with their respective genetic problems (here I rely on Kosztolányi 2013, 72–74). It was observed in the United States that sickle-cell anaemia is more prevalent in the members of African American communities than in non-African Americans, so this was integrated into the screening programs of several states. In a similar manner, it is argued that cystic fibrosis (CF) is a disease that occurs more frequently in Caucasian populations than in others. Particularly the ΔF508 mutation is responsible for roughly two-thirds of the occurrences. In these cases, it is argued that it is both rational and economically beneficial to design racially/ethnically sensitive screenings that would help these communities to tackle these genetic issues. In the development of genetic screenings in Hungary, biological averages were used to define the thresholds of acceptability.

> In Hungary every newborn is screened for certain deficiencies. It doesn't matter if the newborn is German, Dutch, Russian, or Ukrainian; this is a compulsory screening for

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> every newborn. Blood samples are collected on a filter paper and half of the country's samples are sent to Szeged, and the other half to Budapest. Only those will be notified whose results diverge from the norm. This is not a diagnosis, this is a precaution. These people are requested to return to the clinic and subjected to a focused examination. It is possible to set up a diagnosis only after the examination. There is absolutely no distinction on ethnic grounds (PI 20121018).

The whole population was studied to establish mean values for various problems, such as PKU, to be able to give meaningful medical answers to those who are affected by the disease. It is important to note, however, that today there are arguments provided by researchers, for example about CF, that there are various forms of cystic fibrosis mutations but different populations are affected by only a given set of these. And certainly, this can be true for various ethnic groups within a larger population, so the perspective to screen a given community with the same parameters may well be imprecise. However, the question remains: how do ethnic or racial identities best serve the medical needs of community members?

Here, another relevant question comes up regarding the medically-guided distinction of the Hungarian population on genetic grounds. Roma people are mainly referred to in the literature as Asian, regarding their ancestry. Recent studies argue that the ancestors of Roma people presently living in Europe can be traced back to their ancestral geographical origins in Northwestern India (Pamjav et al. 2011; Martínez-Cruz et al. 2016). In opposition to this position, other geneticists argue that this type of differentiation is not tenable or useful.

> Geneticists do not take Roma people to be an Asian group; with this mindset we Hungarians could be Asians, too. I don't know about any genetic abnormality which has a higher frequency in Roma communities than in non-Roma communities. According to our present knowledge from the perspective of diagnostics, there is no difference between a Roma and a non-Roma: we must take them to be of Caucasian ethnicity. It would be an exaggeration to consider them to be Indians. In everyday screening practice, there is no difference between the white population and the Roma population (PI 20121210).

The counter argument centres on the tacit linguistic, anthropological, and historical knowledge that Hungarians migrated to their present geographical area from Asia. Despite this accepted view, ethnic Hungarians are classified as Caucasians or Europeans. Importantly, there is no significant genetic difference regarding disease prevalence in the Hungarian Roma population that the above geneticist knows of. And this also entails that there is no official disease panel suggested by the Hungarian Human Genetics Society that would recommend racially focused screenings. This shows us the untidiness of boundaries that genetic discourse creates for white Hungarians and non-white Roma Hungarians as possible identification schemes, because these are still based on classic racial markers such

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as skin colour or hair texture when it is widely acknowledged that there are no biological grounds for racial differentiation (Raskó 2015, 147–148). However, this entails, as Raskó (2015) argues, that there are mutations, which accumulate in various groups who intermarry for a long time, and this prompts researchers to suggest further sensitivity in screening and providing diagnoses. This perspective is explained below with a joint problem called Bechterew's syndrome that is perceived to be more common in the members of Roma communities.

> There can be biological differences regarding ethnic belonging. Let's take an example: in gypsies the occurrence of Bechterew-syndrome is much higher than in the non-gypsy population. So, when a gypsy young man comes and tells us that his waist hurts him, this is the first thing we have to think about because almost every second gypsy man will have this problem. And this is not racism. This is an empirical fact. And it is right to think about why this has developed this way, it is right to think about it, however this is how it is (PI 20130311).

The initial symptoms of Bechterew's syndrome are lower back pain or back pain that usually occurs during the night with changing intensity but it can worsen in the morning or with inactivity (Brent 2018; Sáfrány 2010). Its occurrence in populations is 0.1–1.4 % and it is more prevalent in males than in females.

The precise cause of the syndrome is unknown, but it has been shown that familial transmission of the gene is frequent. In addition to this observation, many point out that the presence of the HLA B27 allele can be detected in most pathological cases. However, this HLA B27 allele can most likely only be held responsible for 20 to 30 % of risk factors that cause the disease (Sáfrány 2010, 13). Enikő Sáfrány, a medical geneticist, studied nine single-nucleotide-polymorphisms of the IL27R gene. The IL-23R is a transmembrane protein that can be detected on the short arm of the first chromosome (1p31.3) (Parham 2002, cited in Sáfrány 2010, 6). Sáfrány found that certain SNPs are higher in frequency in the population that has the disease in comparison to the control group. The rs11805303 T allele, rs1004819 A allele, the rs10889677, and the rs2201841 SNPs are detected to be more frequent in populations that exhibit the disease. She claims that in the case of the IL27 R haplotype, in connection to the development of the syndrome, the ATCACAG and ATCACAA haplotypes consisting of the rs1004819, rs7517847, rs7530511, rs10489629, rs2201841, rs10889677, rs11209032 variants were understood to be susceptibility factors; while in the case of the patients who carried the B27 allele, only two haplotypes showed connection with the disease. The GGCATCG haplotype was proven to be a defense factor, while the ATCACAA haplotype was understood to be a risk factor in the examined Hungarian population (Sáfrány 2010, 30–31). In the papers that were published by E. Sáfrány et al. (2009), and later by Sáfrány herself (2010), regarding this problem, neither racial nor ethnic categories were

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mentioned by researchers to classify their subject material. However, it is possible to make further distinctions by using racial/ethnic categories regarding the frequency of the disease in various communities.

Investigations aiming to understand the links between various immunological diseases and the above-mentioned SNP variants of the IL23R gene were initiated by Richard Duerr and his colleagues (2006); in their study, they compared the IL23R gene variants in samples taken from Jewish and non-Jewish patients. They stated that there is significant correlation between the function of the gene variants in the development of Crohn's disease. Similar studies have been conducted across the world. Researchers examined various SNPs regarding the IL23R gene mutations that can be relevant to Crohn's disease in Brazilian populations, in New Zealanders, in Koreans, in Chinese, and in Germans (listed in Magyari et al. 2014, 150–151). This is the direction taken by Hungarian researchers when they began investigating the prevalence of the IL23R SNPs in ethnically identified samples. Magyari and her colleagues compared the IL23 receptor gene variations in Roma and Hungarian population samples.

> [We] examined five susceptible, one protective and two neutral variants of the IL23R gene, and found significant increased genotype and allele frequencies in rs10889677, rs1004819, rs2201841, rs11805303, rs11209032 in Roma samples compared with the Hungarian population, and the rs7517847 showed significantly decreased genotype and allele frequencies in the Roma samples compared to the Hungarians (Magyari et al. 2014, 151).

Because various studies pointed towards the correlation between the susceptible variants of the gene and the disease, their finding implies, they argue, that hypothetically Roma people are more prone to develop the Bechterew's syndrome than non-Roma Hungarians.

In the above discussed case, medical genetic findings are paramount for the better public health for both Roma and non-Roma Hungarian citizens. The question is how one understands it and how citizens are capable of using the information. I would argue against the starting position of my interviewee, who stated that when a Roma male individual with lower back pain enters his office, they (the medical professionals) must consider the possibility of Bechterew's syndrome. It would be misleading for both parties to consider Bectherew's syndrome only when racial/ethnic identification matches the description of the patient and the perception of the doctor. Those who are non-Roma but similarly carry the gene variants that make them more susceptible to develop the disease are left out in this perspective. According to Sáfrány (2010, 29), 47 % of the healthy control group had the same genetic variant, namely the presence of the rs11805303 T allele in one instance, and also the rs1004819 A allele variant was more frequent in groups who had the disease than in the healthy control

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groups. With these results, geneticists argue that (Sáfrány et al. 2009; Sáfrány 2010; Magyari et al. 2014), because of the difference in frequency of the variant between Roma and non-Roma carriers, most probably Roma people are more susceptible to Bectherew's syndrome. This information can work to the advantage of both Roma communities and health professionals, but only on the condition that they are careful to screen non-Roma patients with similar symptoms for the same genetic variants. And with this move the work that geneticists do can be seen as a contributing force in re-thinking the divide between Roma and non-Roma Hungarian communities. Members of both vaguely defined communities carry these variants, and the only medically significant difference is the fact that one in one of these groups' carriers is more frequent.

Following on from this, two problems arise regarding the use of race/ethnicity. The first problem lies in the difficulty of identifying someone's ethnicity, and the second problem is the precise application of said knowledge. It is important to address this issue, because it is possible that an individual cannot precisely define their racial or ethnic ancestry. This can be for various reasons but let us take one: incomplete ancestral information was passed down across generations. In this case, what is the best solution? And how can this approach accommodate the individuals' freedom to choose their ethnic or racial identity according to their social circumstances? Let us say a white immigrant from Africa who lives in a European country permanently, perhaps even without planning to return to their country of origin, chooses to identify as White African. Immediately, a similar question emerges: if geneticists use stratification more and more, then to what extent should they rely on folk race/ethnic categories? In what ways can we secure precise information flow regarding these social categories in medical settings when we must ensure precise diagnoses? In order to be inclusive, the medical response must begin on the biological grounds that there are different genetic mutations that must be identified, since these are crucial for successful treatment. Ethnic/racial markers understood by either patients or health professionals in a superficial manner can lead the diagnosis astray. Let me use an example of a patient suffering from cystic fibrosis. CF is understood primarily to be a Caucasian problem, one that largely affects white people, and as such this understanding directs the medical gaze of health professionals along racial identities.

Arguments to use racial/ethnic markers put forward by geneticists include time efficiency in a clinical setting, where the patient's interest is to find medical solutions to their problems as soon as possible, hence medical professionals need reliable markers that efficiently guide the therapeutic process. In addition to this criterion, it is often argued that it is simply not economically efficient to screen a patient for everything, since it is a costly procedure, and it is also inefficient for the state for the same reason. Therefore, many suggest that racially or ethnically different populations be screened for health problems that are more

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prevalent in specific communities. Dorothy Roberts discusses an exemplary case where the racial bias of healthcare professionals caused harm instead of fast and efficient treatment (2011, 99). Roberts tells the story of "Lela, who was described by doctors as a '2-year-old black female with fever and cough' and later as a '4-year-old with another pneumonia', as she continued to suffer from an unshakable respiratory ailment" (Roberts 2011, 99). After six years of continuous imprecise diagnosis, at the age of eight her chest X-ray was read by a radiologist who identified her condition as cystic fibrosis. The radiologist did not have any previous knowledge about her; Lela was not classified as a black patient by this radiologist; it was only the X-ray image that allowed the doctor to give a racially unbiased diagnosis of her respiratory problem. In Lela's case, it is highly probable that if she had been white, she would have been diagnosed early on with CF and treated correctly. Roberts emphasizes that because of the racial lenses that crude statistical data provide about various racially or ethnically significant diseases, medical professionals mistreated Lela because they interpreted her racial difference in a way that was translated into the clinical practice that CF is a predominantly white disease, which means that black patients very rarely suffer from it. Thus, her doctors never considered checking her for CF despite the symptoms that she had shown during her physician office visits.

6. Conclusion

One of the turning points in medical genetic studies was the completion of the Human Genome Project. Its results have changed our thinking about the possible applications of genetic knowledge in medicine. One of the key fields where it had a significant impact was epidemiology. The knowledge gained made it possible and ethically necessary to address population-based health problems with the tools of genetics. Thus, partially, racially/ethnically identified populations became the focus of such studies, in order to provide equal healthcare for everyone. Furthermore, it became widely accepted that in order to provide equal healthcare, it was necessary to map population differences in any social context. This approach embraced the idea that economic marginalization, racial discrimination, and gender inequality contribute to diverse health issues and unequal access to healthcare. Thus, it became mandatory to find ways of tackling these empirical problems. Social categories, such as race and ethnicity, might provide useful guidelines both for clinicians to reduce the time that is needed for a precise diagnosis and to offer medical services, and for patients on how to change their lifestyle in order to better attend to their health. However, it is important to be vigilant about the social processes that reduce certain diseases to race- or ethnicity-based problems. An essentialist understanding of race/ethnicity in medicine can constrain the medical gaze and thus interfere with the diagnosis and the treatment process.

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Notes

[1] Identical treatment is a term suggested by Risch and his colleagues (2002) to explain that different social groups have different treatment responses, therefore, they argue, treatments must be adjusted to their specific needs.

[2] There are sociological and anthropological studies that explain sensitively the social immobility of Roma people living in segregated areas (see for example Rozgonyi-Horvath 2018).

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