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Taking Account of Human Genetics in Identity Research

Introduction

It is sometimes remarked that the quest for identity is the real aim of genealogy. But what is identity; what does human genetics offer to the identity debate; and how does genetic genealogy impact on cognitive views of identity? Currently, mention of DNA tends to polarize rather than unite. For example, Brodwin (2002; pp. 323, 324) opines that … *to the dismay of anthropologists who fancy themselves as the cultural avant-garde, essentialist identities grow ever more powerful and seductive*. He takes the view that emerging knowledge in the field *… adds the cachet of objective science to the notion that one's identity is an inborn, natural, and unalterable quality. Rapid advances in sequencing and analyzing the human genome have strengthened essentialist thinking about identity ...* He adds *… Emerging genetic knowledge thus has the potential to transform contemporary notions of social coherence and group identity*. Comfort (2019; p. 170), on the other hand, remarks that [d]*efining the self only in biological terms tends to obscure other forms of identity, such as one's labour or social role*. Hauskeller (2004; p. 296) concurs and states: *Although DNA is no doubt real, it is clear that there are systematic problems in employing it for purposes such as establishing identity*. Clearly, the use of human genetics in identity is a controversial topic.

The most comprehensive article that I have come across about how genetics relates to identity shows that there is little agreement, even among scientists. The paper in question is a systematic scoping review of the concept of 'genetic identity' by Goekoop, *et al* (2020; pp. 1, 16). The authors found that: *… a clear understanding of the term is lacking … Overall, the diversity in the use of 'genetic identity' in the reviewed literature demonstrates that the term is used differently in different contexts, but also within each context the meaning of the term can vary widely* [and be used] … *in contradicting ways* (Goekoop, *et al*, 2020; pp. 1, 16).

The present paper provides a fresh look at this complex subject, taking care to define terms clearly and avoid—in as far as possible—the most obvious contradictions. It does not consider *artificial* interventions in human genetics. In other words, no medical therapies or interventions like organ transplants or blood transfusions are contemplated when considering biological identity. At this juncture, determining when a significant rupture has been caused and the biological identity of the individual has been compromised is open to interpretation and will be influenced by growing information about the functioning of our genome. However, this discussion is outside the remit of the presentation. In addition, the focus of the paper is on nuclear DNA rather than on mitochondria.

Hauskeller points out that *DNA is real.* But how do we account for it or incorporate it into wider identity studies? How does genetics impact on our cognitive or subjective views of identity which currently predominate? Intuitive approaches may appear more reasonable and compelling to many, than relying on what seems to be an indifferent chemical molecule to define the term. But surely DNA has a complementary role to play? It is only by knowing more about the subject that identity research will mature and answer questions about the role of human genetics in determining who we are. The purpose of this paper is to: 1) Provide a brief introduction to DNA and propose the human genome as **a** basis for genetic or biological identity; 2) Show how studies in human genetics and cognition can be compatible and complementary rather than irreconcilable or contradictory; and 3) Begin a discussion about how to capture mutually reinforcing benefits by amalgamating human genetics and subjective studies of cognition. The wider focus of the paper is on introducing biological or genomic identity as a base for identity, in contrast to cognitive perspectives, which are recognised here as expressions of how we identify

or are identified.

'Sameness'

The confusion associated with 'identity' can be traced to its late 16th century origins, combined with a conundrum that stretches into the remote past. The word emerged from Medieval Latin *identitas* or *idem* meaning 'sameness' or 'same'. Unfortunately, 'sameness' has been difficult to comprehend. This is well illustrated in the ancient Greek paradox of the Ship of Theseus, which was kept as a monument to its hero at Athens. As time passed the planks had to be changed one by one when they began to rot. At what point, philosophers asked, did the original ship cease to be itself? If every plank, rib and panel were replaced, along with the nails that held them together, how could it be the same ship? Surely it was only a replica. We know that the chemical constituents of our bodies are continually changing. As such, are we the same person as we move through time? In sum, there have been no adequate answers and thus 'identity' has suffered a crisis of meaning. Fortunately, the 'sameness' paradox is no longer a stumbling block in biological terms, at least in relation to the question: Are we always the same (physical) person?

Personal identity

Identity has an individual and a group dimension. Since 1987, thousands of forensic cases have been decided with the assistance of genetic fingerprinting (Roewer, 2013). Confidence about its application is generally based on the observations that our individual DNA sequences are unique. Besides, observations indicate that the nuclear DNA of every cell remains the same throughout the life of the organism. In other words, science reveals what is meant by 'sameness' in biological terms in who we are. If this is correct, then our DNA sequences provide a basis of our genetic identity at the personal level, which can be defined using older and more rigorous meanings of the word. For example, the Oxford English Dictionary (OED 2nd Edition, 1989), defined personal identity as: *The sameness of a person at all times or in all circumstances; the condition or fact that a person is itself and not something else*. It is the definition that is applied in this paper.

However, Dr. Alexander Hoischen of Radboud University Medical Center in the Netherlands, who was part of a research project into mosaic mutations stated that: *The textbook knowledge that our genome is identical in all the cells of our body is probably not true* (Azvolinsky, 2015). As sequencing practices are becoming more widely used and as techniques are improving, an increasing number of anomalies are being recorded. Despite advances, limitations of current technologies still hamper our understanding of the extent of these changes (Acuna-Hidalgo, *et al*, 2015; p. 67). Mutations may occur in early development at the zygote stage, but they may also appear in post-zygotic development. Account must also be taken of fetomaternal microchimerism which is a research field in its infancy and which refers to the bi-directional exchange of a small number of cells and cell-free fetal DNA through the placenta (Murrieta-Coxca, *et al,* 2022 and Rosner, *et al*, 2021). Although our knowledge is in its infancy, the exceptional intrusion of complex DNA appears to have important implications during and after pregnancy (Rosner *et al*, 2021). The repercussions of fetal DNA transfer and post-zygotic mutations on the definition of personal identity require careful consideration. Other considerations are the mutations that occur in a stem cell whose descendants become a specialised organ. The tissue of such an organ will appear different compared to all other parts of the body with respect to that mutation. The term for this anomaly is a 'mosaic mutation'. Anomalies can also arise, caused by the fusion of two zygotes during the early embryonic stage, forming a *fusion chimera* (Madan, 2020). The appearance of such differences has given rise to the suggestion that some of our cells carry different versions of our genomes (Azvolinsky, 2015) or that some people carry multiple genomes (Ledford, 2019; Lupski, 2013). However, for the purposes of the

present discussion, the genome is understood to mean *the complete set of genetic material present in an organism*, whatever its complexities. Therefore, all material—whether original, mutation, or even blended genotypes, as in the case of fusion or fetal chimeras—is included in the term and this is supported by the arguments that the personal genome is singular in expressing its nature and, furthermore, that the entire organism is a viable unit.

We know that mutations are relatively low in the human body. For example, in each new generation *one error in every 10^8 base pairs gives rise to 30–100 genome-wide de novo or new mutations* (Acuna-Hidalgo, *et al*, 2015; p. 67). This means that 0.000065 percent of base pairs are changed, leaving 99.999935 percent unchanged. Every mutation affects a minor part of the entire genome. In other words, a high level of structural conservation of nuclear DNA is retained in the bulk of our underlying genetic sequences across the whole organism. Nonetheless, the contribution of the post-zygotic mutation rate is unknown. New studies continue to emerge. One result reveals that early mutations were estimated to be 0.34×10^{-8} for one of a monozygotic twin pair and 0.04×10^{-8} for the other (Dal, *et al*, 2014). This shows that so-called 'identical twins' are not identical. Another study detected mutations among multiple samples obtained from the same individuals that included samples of blood, saliva, hair follicle, lining of the cheeks, urine, and semen. Results indicate that a clinically unremarkable person might harbour post-zygotic mutations corresponding to around 1.5×10^{-8}-4.4×10^{-7} per nucleotide per individual (Huang, *et al*, 2014; p. 1319). These are relatively low numbers and confirm the structural conservation of the bulk of our genome. It may seem reasonable, therefore, to base 'sameness' of the person on the observation that most of our genome remains the same. But no matter how small the modification is—it *is* a change, and the strict meaning of 'sameness' is infringed. This scrupulous attention to detail is important. After all, if a small mutation gives rise to a gene, or influences a group of genes that induce susceptibility to cancer or another malady, it is highly significant for the whole organism. So, even though the rates of change are relatively small, the implication that the genome is not the same *at all times or in all circumstances* in terms of its structural makeup is irrefutable in these instances. As mutations are part of the life of our genome, DNA sequences—alone—cannot be used to define personal identity ('sameness') in all cases. For this reason, the term must be qualified. The question we must now answer is: How do we know that we are the same person if post-zygotic mutations take place? In what way are we the same under these circumstances? It can be demonstrated that 'sameness'—though not contained in all DNA sequences—is present in the individual's genome, which conforms to a specific set of unchanging conditions. These include:

1. Continuity: development from its beginning to end-of-life is an uninterrupted series of events (in terms of life processes or metabolism)
2. Viability: the genome survives as a single entity
3. Individuality: the personal genome is singular in expressing its nature; and
4. Uniqueness: it is unlike any other genome

Continuity intimately links subsequent post-zygotic mutations to the same genome. A separate personal genome is not created because mutations take place. Mutations are part of the nature of many—if not most—individual genomes. Not only is the bulk of the underlying genome conserved in terms of its structure, it remains viable at all times. Viability is an important quality in that it overcomes the suggestion that the chimera or mosaic genome is, in fact, two or more people in one. A person cannot be divided up—particularly if vital organs are involved—and remain viable. And individuality is maintained from the point of fertilization until death. Furthermore, individuality is highlighted by the uniqueness of the makeup of the genome itself, which must also take account of changes in epigenetics (i.e., different genes being 'turned on' or 'off') which cause genomes to differ. On a separate issue: even monozygotic twins (or multiples) share a common beginning when the parental gametes unite. However, viability and continuity

in terms of life processes or metabolism, are not compromised during the shared stage for each genome. The case of conjoined twins, especially those sharing vital organs, represents an exception and requires further consideration.

The possibility of infringing 'uniqueness' where cloning has occurred is a perennial question, assuming that it was ethically permissible, which it is not. However, even in a case in which the nuclei of both donor and clone are the same, DNA reacts in conjunction with the cellular fluid (cytoplasm) in which it is embedded. Therefore, to be the same the nucleus of the clone would have to be introduced into the same host as that of the donor. Many discrepancies would arise as a result of epigenetic changes or gene expression. The developing cells would also be subject to random mutations and the effects of fetomaternal microchimerism. Furthermore, even a clone of oneself would differ from the donor in that its subsequent development would never replicate the original.

Another issue that is sometimes raised in relation to biological or scientific identity is the complexity of new information emerging from microbiology which forces us to take into consideration the relationship between the host and the microbiome (Liu, *et al*, 2021) especially in relation to symbiosis and pathogenesis. Comfort (2019; p. 169) goes as far as suggesting that the 'biological self' has been reframed as a cluster of communities rather than the individual. However, from the point of view of human identity, the integrity of host and microbial communities are maintained. Symbiosis or virulency are, after all, broad terms to describe *different* organisms interacting with the host (Eloe-Fadrosh and Rasko (2013) and Méthot and Alizon (2014)). Besides, a person's immune system is unique with regard to other living organisms and even when compared to other individuals of the human species, including a monozygotic twin. Consequently, individuality is manifest in the boundaries drawn by one's immune system (Pradeu, 2012; pp. 7, 8).

Communal identity

According to Keogh (2019; p. 17): *In biological terms, a species is generally defined as a group of organisms capable of successfully exchanging genes, or in other words, capable of interbreeding to produce fertile offspring. Ability to reproduce with our own kind is, therefore, the essential constant that distinguishes us as human. This means that communal identity becomes manifest through the 'trinitarian' act of reproduction, in which two personal identities of the opposite sex give rise to a new personal identity.* The main question to answer here is: How does communal identity 'become manifest' through procreation? Focus is placed on the formation of the individual genome irrespective if the parents produce other offspring at another time or several unique genomes through the one reproductive event. The instant that gene fusion takes place, which describes the essence of reproduction, is expressed by Condic (2008; p. 3): *Following the binding of sperm and egg to each other, the membranes of these two cells fuse, creating in this instant a single hybrid cell: the zygote or one-cell embryo … Cell fusion is a well studied and very rapid event, occurring in less than a second. Because the zygote arises from the fusion of two different cells, it contains all the components of both sperm and egg, and therefore the zygote has a unique molecular composition that is distinct from either gamete. … These modifications block sperm binding to the cell surface and prevent further intrusion of additional spermatozoa on the unfolding process of development. Thus, the zygote acts immediately and specifically to antagonize the function of the gametes from which it is derived … Clearly, then, the prior trajectories of sperm and egg have been abandoned, and a new developmental trajectory—that of the zygote—has taken their place*. Clearly, the essence of reproduction occurs … *in less than a second* and, although communal identity becomes manifest through it, the underlying 'sameness' of the group is ongoing and not confined to a single instant. How, then, is this 'sameness' or identity to be understood?

The collective dimension in procreation is underpinned, firstly, by the degree of randomness associated with the parental encounter, which involves the union of two representatives of the opposite sex from the segment of the genomic spectrum that is potentially fertile at that moment. Even though panmixia is only theoretically feasible, the entire component of humanity that falls into this category, is open, at least in biological terms, to the possibility of interbreeding. Random events make potentialities concrete. Irrespective of the capacity—or incapacity—of offspring to procreate, all humans are connected to the reproductive process in that they have been created by it.

Turning to the wider picture: DNA patterns do not correlate well with the normal divisions of humanity like race, ethnicity, culture, or nation. This does not rule out genetic patterns that are more common in certain groups. However, ethnicity and race are shown to be non-genomic (Kim, *et al*, 2023). Genetics is the new classifier and shows that it is not possible to recognise these categories in terms of our DNA because individual genomes are part of a virtual continuum of genetic variation around the world (Marshall, 1998). The interconnectedness of modern humanity resulted from our origins in a small bottleneck of people in Africa—our *ancestral singularity*—some 200,000 years ago and subsequent developments (Keogh, 2016; pp. 146 ff). Even the probable interbreeding with Neanderthals and Denisovans failed to establish a separate species and inhibit intermixing. From a genealogical viewpoint, an individual is composed of multi-dimensions in their many genetic pathways out of the past. In other words, genomic composition is made up of DNA mixes that often emerge from sub-groups that intertwined uneasily in society. However, knowledge that we share a common biological identity helps alleviate divisiveness in terms of logic, but often requires a long process of emotional reconciliation before a satisfactory acceptance is established between contested components in our makeup.

The entire human genome, which is composed of unique and therefore different individual genomes, constitutes the complete spectrum or range of DNA sequences that exist. It follows that no human sub-groups—no matter how large or small—are made up of individuals who share a 'sameness' that is exclusive to that cohort. The communality within sub-sets that we observe across this spectrum contain similarities that may be helpful in the study of human behaviour, but they do not constitute separate identities. Resistance to the notion of intrinsic internal divisions in the human family—beyond the individual—means that communal identity is located exclusively at the universal group level, which is composed of all living individuals or personal genomes. The *nature* of the characteristics that imbue the individual with personal identity (continuity, viability, individuality and uniqueness) are shared by parents and offspring. In like manner, this nature is shared by all people and provides us with a basis for expressing communal identity or 'sameness' at the universal level. Consequently, collective, universal or biological identity may be defined as: *The sameness of the group at all times or in all circumstances; the condition or fact that the group is itself and not something else.*

Power of DNA

The founder of the modern science of genetics was Gregor Mendel who discovered the principles of biological inheritance between 1856 and 1863. But the field only evolved rapidly after the derivation of the structure of the double helix in mid-20th century (Pray, 2008). Another significant recent advance was made by sequencing the entire human genome in 2003. Our rapidly growing knowledge has initiated what is popularly called *the DNA revolution*. The revolution is advancing through the increase in our understanding of how DNA works and how it can be manipulated, which is not without risks. We now know that DNA contains the biological instructions that make each species unique. It is not a passive molecule; *it is highly dynamic, enwrapping layers of complexity … Its function as a universal genetic*

material is among the most highly conserved qualities of living things. Its system of four bases, when overlaid with spatial and temporal controls, governs biology across the entire scale of life and actively contributes to all living processes (Duzdevich *et al*, 2014; p. 3072). It contains the instructions needed for an organism to develop, survive, thrive, reproduce and pass on genetic information from one generation to the next (hereditary). The roles played by nature versus nurture or—more likely—the complex interactions between the environment and the way DNA assuages its impact to give the organism the best chance to stay alive and prosper, must be considered. It is also known that DNA influences personal attributes. Plomin (2019; p. viii) states that he is *not aware of a single psychological trait that shows no genetic influence*. Duncan *et al* (2019; p. 1518) reinforce the link between disease and genetics when they point out that *all major psychiatric disorders have now been shown to be polygenic*. Furthermore, specific loci have been identified through genome-wide association studies (GWAS) rather than by way of specific candidate genes. However, these authors also note that this approach requires *massive increases in sample sizes (i.e. encompassing tens of thousands of participants, or more)*. The advantage of very large sample sizes is that they tend to avoid the dangers of non-repeatability in experimental outcomes, which is a stumbling block to scientific progress, especially considering the current crisis of replicability (Pashler and Harris, 2012). [U]*ntil now, psychologists have had to rely on behavioural symptoms to diagnose disorders*. Genetics is beginning to offer *a causal basis for predicting disorders rather than waiting until symptoms appear and then trying to use these symptoms to diagnose disorders* (Plomin, 2019; p. 66). These results are likely to improve as more GWAS—using very large samples—become available.

However, a central role for DNA in personal and communal identity tends to trigger antagonism based on concerns that it is a form of essentialism that equates with **genetic** determinism or reductionism. And these concerns are intensified by the assumption that human behaviour is, therefore, under the full control of an individual's genes at the expense of the roles of the environment, learning or free will. One way to dispel the notion of genetic absolutism is to consider a scholar who becomes convinced by an argument and is converted to a new way of thinking and acting after reading a ground-breaking scientific paper or book. Genetics allows the subject to see the hieroglyphs on the page, translate them into meaningful thoughts, commit them to memory and compare them to knowledge already accumulated, thereby forging associations, stimulating comprehension, and coming to conclusions that are independent of the personal genome. The incoming words and ideas are the principal agents activating segments of the brain during this process, not the underlying DNA. Julian Baggini (2015), reflecting on human genetics, comes to a nuanced conclusion, which discounts an 'either/or' argument in which either genetic absolutism, the environment or unrestricted human freedom rule supreme. A balance between these aspects reflects the human condition more accurately. However, historically, we are in the difficult process of determining the limits of these factors. On the other hand, 'scientism'—or the belief that material science is the only source of authentic knowledge—is unsound and may be discounted.

Discussion

Most of this paper has been dedicated to introducing the genomic base for genetic or biological identity. In what way might the approach impact on studies in the humanities that rely mainly on subjective cognitive analysis to define and understand identity? Modern classical thinkers and philosophers tended to avoid answering the question 'What is identity?' in terms of material content and, instead, sought to find 'sameness' in intellectual traits. In simple terms, the main question they tried, and failed to answer, is: Does our 'sameness' persist over time; and do we possess an unchanging 'essence'? Hume attempted to overcome the conundrum by suggesting that sameness resides in 'memory'; Descartes favoured 'thinking'; Locke opted for 'consciousness'; Erikson advocated 'ego'; while 'sense-of-self' gained popularity among a wide spectrum of social scientists. The problem in these cases is: How to account for

persistence when we sleep, or if we happen to be in a coma, or if we have the misfortune of getting Alzheimer's Disease? And how do we account for persistence before we had a remembered memory of any sort, as in our time in the womb? On the other hand, does our sense-of-self not change over time? What about personality? **Surely** that, too, can change. Personality modifications may occur when certain physical changes are induced in the brain. Considering these objections, many philosophers began to avoid the suggestion that we have an unchanging essence that makes us who we are. The reason for the turnaround is based on the conviction that 'sameness' is difficult to prove, and therefore, is unsolvable. But failure to reach a satisfactory conclusion exacerbated the problem and deepened the 'identity crisis' as it morphed into what have been called postmodern and constructivist strands of academic thought. These trends began in the 20th century and have made the topic more puzzling by sharing … *a fundamental disbelief in the existence of any 'objective truth' and 'definiteness' – conceptual or otherwise* (Prusch, 2017; p. 10). Consequently, identity is portrayed as not being static, is subject to fluxes and does not exhibit a *sensu stricto.* This inference *… provides the basis for understanding that people's identities may have many different facets, can change all the time and might even contain contradictions*.

However—echoing Hauskeller's observations—*DNA is no doubt real*. It is unwise, therefore, for social scientists to ignore scientific observations in their research. One possible way forward is for the academic world to acknowledge that revelations in biology point to an underlying physical sameness in the individual and in the community. Given that this approach can be demonstrated independently and repeatedly through empirical means allows it to be applied to research in social science and studies of the modern classical thinkers and philosophers, as well as genealogists. The collective reasoning of these specialists provides intermittent glimpses of continuity of the person through memory, thinking, consciousness, ego, narrative and sense of self. The principles on which biological identity are based offer an underlying framework to affix the views of all these scholars and practitioners, thus strengthening their theories. However, 'sameness at all times' transfers to the material content of the personal genome. This change allows sociologists to advance with greater ease in new directions because they are unhindered by the need to reify identity from subject notions of self. Clearly, studies in human genetics and subjective cognition, are compatible and complementary rather than irreconcilable or contradictory. The amalgamation of biological and subjective views will—I believe—also help to determine when a significant rupture has been caused in biological identity through artificial interventions that interfere permanently with or eliminate cognitive continuity. However, clarifying this and many other aspects of identity lies well into the future. To have any chance of success there is an urgent need to begin a serious discussion about the merger of empirical and subjective studies in identity so that the mutually reinforcing benefits of both human genetics and subjective cognition are maximised.

Conclusions

The reluctance to engage in human genetics was highlighted in a presentation I made at the 29th Annual Conference of the International Society for Research on Identity (ISRI) at Tufts University in Boston in 2023. In a rapid and informal oral response from my audience, around 80% were sceptical, had doubts, or knew little about the application of human genetics in identity studies. One participant mentioned its misuse in the past. The varied responses reflect the ongoing ethical and social discussions that often accompany advancements in genetic research and its potential implications. However, an African American participant was enthusiastic about the use of genetics, citing its value as a method of tracing origins in Africa for a people whose history was denied to them due to the slave trade. Her comments echo a wider reality. Genetic genealogists and many family historians have little difficulties in fusing DNA testing with traditional genealogy, oral history, and documentary records, because it has proven to be a powerful research tool. It offers the potential to overcome the challenges and obstacles that often arise

when tracing ancestry through purely non-genetic methods. However, like social scientists, genealogists often view cognitive perspectives as 'identity', which can cause confusion, particularly if identity is viewed as unchanging. Tests results have often produced surprising results, forcing practitioners to change their views of self. It would, however, be incorrect to suggest that their underlying genetic identity has changed; they have discovered something extra about it. For simplicity the dilemma is avoided if cognitive perspectives, which are subject to change, are recognised as expressions of how we identify or are identified, rather than our 'identity'. Viewed from this perspective, genetic genealogy may be seen as a forerunner or vanguard in that it has the potential to merge biological and cognitive perceptions on identity, leading to a more comprehensive understanding of who we are and where we come from, with wider implications for reshaping our understanding of identity itself.

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