Charles University

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Human Identity Through the Prism of Modern Science: Transformation, Reception, and Application of Molecular Genetics in Public Discourses

Lidská identita prizmatem moderní vědy: transformace, recepce a aplikace molekulární genetiky ve veřejných diskursech

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Declaration

I hereby declare that this thesis, titled "Human Identity Through the Prism of Modern Science" is the result of my own original research and has been written by me. This work has not been previously submitted for any degree or examination in any other university. All sources used are acknowledged in the thesis, and direct quotes are clearly designated as such. I certify that all information sources and data used during the production of this thesis are properly credited, and I have clearly distinguished between my own work and the work of others. The thesis was prepared under the guidance of Mgr. Tomáš Hermann, Ph.D., and it adheres to the ethical guidelines of Charles University.

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Abstract

This thesis explores how the advances in genomics and molecular genetics from the 1990s to the present have shaped human self-perception in public discourses. It looks closely at how the Human Genome Project (HGP) and subsequent advancements have altered public views on sociocultural ethnicity, health, disease, and ethics. By integrating multidisciplinary insights from molecular biology, sociology, and bioethics the text provides a critical examination of the landmark literature within the specified timeframe. The thesis argues that genetic progress not only provides an understanding of our biological foundations, but also raises important moral, legal, and societal issues that often challenge conventional notions of human self-perception.

Abstrakt

Tato práce zkoumá, jak pokroky v genomice a molekulární genetice or roku 1990 do současnosti transformují vnímanou lidskou identity ve veřejných diskurzech. Zaměřuji se na to, jak Projekt lidského genomu a navazující výzkum mění veřejné debaty ohledně sociokulturní etnicity, zdraví, etiky a práva. Kombinací multidisciplinárních poznatků z molekulární biologie, sociologie a bioetiky text kriticky zkoumá přelomovou literaturu zmíněného časového rámce. V práci argumentuji, že pokroky v genetice nám nejen umožňují lépe porozumět základům života, ale zároveň nastolují důležité společenské otázky a nutí nás přezkoumávat tradiční způsoby sebepojetí.

Keywords: The Human Genome Project, Bioethics, ELSI, Ethnicity, Genomic Privacy, Predictive Healthcare

Introduction

The dawn of the 21st century heralded a new era in the biological sciences, marked most notably by the completion of the Human Genome Project (HGP). This achievement not only advanced the biological sciences but also profoundly affected how individuals perceive themselves and their place within society. As the field of molecular genetics continues to evolve, it increasingly influences various aspects of daily life, informing medical decisions, legal frameworks, and personal narrative identities. By reviewing breakthroughs in genomic research since the 1990s and their underlying sociocultural ramifications, this thesis seeks to understand how molecular genetics has come to shape contemporary notions of cultural ethnicity, health, and ethical responsibility.

The thesis is structured into two main chapters. In the first one, I discuss the scientific underpinnings and achievements of the Human Genome Project and the public perception of genomic research from the 1990s to the present day. The second chapter discusses three aspects or fields that were profoundly impacted by the advances in genomic research: healthcare, cultural ethnicity, and societal policy including jurisdiction. Each section provides a semi-structured, curated review of the most influential literature on that specific topic ranging from the start of the HGP to the present.

Literature was sourced by searching appropriate keywords in academic databases (PubMed, Wiley, Semantic Scholar, ResearchGate, etc.) and aggregator tools (Google Scholar, ResearchRabbit, Consensus, Elicit). Generative artificial intelligence (GPT-4) was used for information processing and text generation in some parts of the thesis with all claims cited appropriately. The topic of interest is a vast one, suggesting an immense amount of literature often with different points of view and conflicting opinions. Therefore this thesis is by no means a complete review of the problematics, instead, it aims to offer a high-level overview of the academic and public discussion and its transformations.

Chapter 1

From Sanger Sequencing to Computational Biology

The dawn of the 1990s marked the beginning of an era that would see biology, particularly genetics and molecular approaches, evolve from its nascent stages into a scientific field that relies heavily on quantification and big data. This period heralded the onset of new methodologies, paradigm-shifting discoveries, and the emergence of influential figures and collaborations that continue shaping the field's trajectory to this day [Collins et al., 2003].

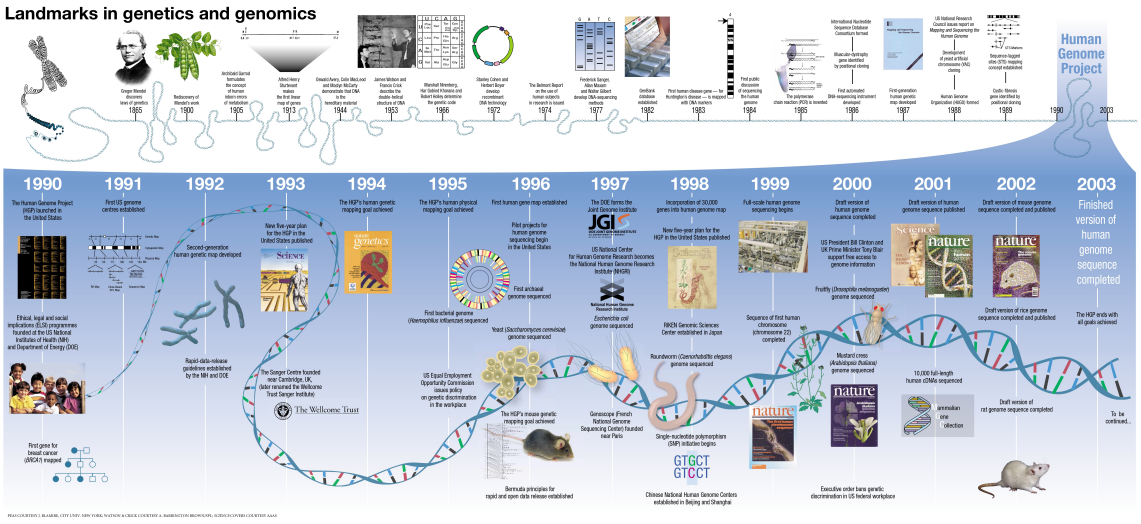

Figure 1.1: Landmarks in genetics and genomics, including the HGP Timeline [?]

1.1 The Human Genome Project (HGP)

Arguably, one of the most influential research projects of the 1990s genetics was the Human Genome Project (HGP) - an international initiative aimed at mapping and understanding all the genes of the human species, collectively known as the genome.

Launched in 1990 as a joint effort of the US Department of Energy and the National Institutes of Health (NIH), the HGP sought to provide a comprehensive blueprint of the human genetic material, offering unprecedented insights into the structure, organization,

and function of the complete set of human genes. The allocated funding was estimated at around $3 billion and the project was expected to take 15 years to finish. Although originally established and funded by the US government, the collaborating groups were located in many additional countries, including the UK, France, Japan, Germany, and China, forming the International Human Genome Sequencing Consortium. This international collaboration was coordinated by the Human Genome Organization (HUGO), serving also as a discussion forum. Among significant participating centers were for example the Sanger Centre, Washington University Genome Sequencing Center, RIKEN Genomic Sciences Center, or Max Planck Institute for Molecular Genetics [IHGSC, 2001].

Central to the HGP's data-sharing initiative was the establishment of public databases like GenBank and the implementation of the Bermuda Principles in 1996. Representatives of sequencing groups from 5 countries conducted 3 "Bermuda Meetings" from 1996 to 1998, during which they agreed upon how genomic data (nucleotide sequences) will be shared and publicized [Reardon et al., 2016]. To keep the HPG public and open, the working groups have consented to an immediate public release (within 24 hours) of newly sequenced data by posting a link leading to a database contribution. This kind of rapid data sharing was also intended to improve the collaboration between international groups and to differentiate the HGP from similar commercial projects [Cook-Deegan and McGuire, 2017].

The human genome, consisting of approximately 3,2 Gbp (per chromosome set) and an estimated 41 000 to 45 000 expressed genes [Das et al., 2001] posed a significant technological challenge at the inception of HGP. Overcoming these obstacles required the development of new sequencing technologies and bioinformatics tools, which allowed for the rapid and accurate analysis of large volumes of DNA sequence data. Initially, sequencing groups relied on the 1st generation Sanger sequencing method, developed by Frederick Sanger and colleagues in the 1970s [Sanger et al., 1977]. Sanger sequencing marked a breakthrough in genomics by enabling accurate DNA sequencing, crucial for the Human Genome Project. This method, which involves the incorporation of chain-terminating dideoxynucleotides, allowed for precise identification of reads up to 1 kb. However, the original Sanger sequencing was labor-intensive, time-consuming, and had low throughput, posing significant scalability challenges for large-scale genomic projects [Heather and Chain, 2016].

In the early 1990s, the original radioactive labeling [Sanger et al., 1977] was replaced by fluorescent dye, allowing the reaction to occur in a single capillary. This in turn allowed for digitalization and partial automation of the process, which meant higher throughput, and subsequent advancements in algorithms that assemble the sequence [Heather and Chain, 2016].

Since producing longer sequence reads was technologically complicated and expensive, the so-called two-phase paradigm was used to organize HGP on a large scale. The sequencing itself and subsequent finalizing represented two coordinated phases during the human genome sequencing process - the "shotgun" and "finishing phases. To this day, this two-phase paradigm continues as a prevalent approach regarding large-scale sequencing [IHGSC, 2001]. Shotgun sequencing is particularly useful when dealing with long sequences as it relies on cutting up the sequence into smaller fragments, which are sequenced independently with a high degree of redundancy - usually up to tenfold (shotgun phase). These are later algorithmically organized and unified into the final sequence (finishing phase) [Anderson, 1981].

After a debate on which implementation of shotgun sequencing should be used, the

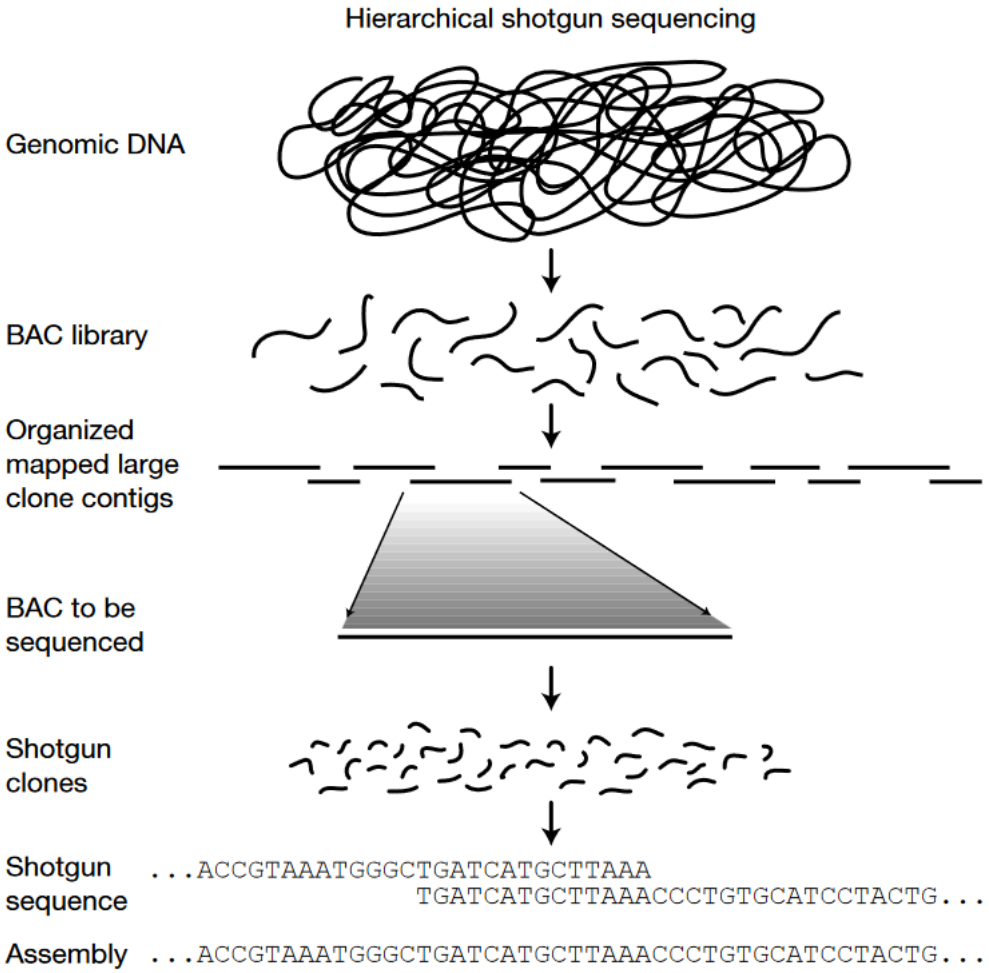

Figure 1.2: Hierarchical shotgun sequencing diagram for large-scale genomic projects [IHGSC, 2001]

hierarchical shotgun sequencing (Clone-by-Clone) method was chosen for the purposes of the HGP [Green, 1997]. This strategy adds some steps into the original paradigm - namely the initial mapping of the sequence by cloning larger fragments of into vectors (such as BACs or YACs) and creating a library. A physical map of the sequence is generated, which then acts as a scaffold for the subsequent sequencing and assembly. This particular variant of shotgun sequencing is considered more precise, but also time-consuming and costly [IHGSC, 2001].

By 1998 less than 5% of the human genome had been sequenced. American biotechnologist Craig Venter, prompted by the slow speed of the HGP has then revealed his plan to launch a business venture (later known as Celera Genomics) and complete the rest of the sequencing in 3 years by using whole-genome shotgun sequencing together with recently released fully automated sequencers ABI PRISM 3700 and new assembly algorithms. The plan was to utilize BAC sequence fragments generated by the publicly funded group to accelerate the project and eventually sell further genomic data to pharmaceutical and biotechnology companies [Venter, 2001].

Both the public and private HPG groups produced and published nearly complete human genome sequences in 2001. International Human Genome Sequencing Consortium has published its findings in Nature [IHGSC, 2001], while the Celera group pub-

lished a similar article in Science even earlier that year, listing Venter as the first author [Venter, 2001].

1.1.1 Ethical, Legal, and Social Implications (ELSI) Program of the HGP

The Ethical, Legal, and Social Implications (ELSI) program, established alongside the Human Genome Project, represented a research policy-making entity within the US National Institutes of Health (NIH) with the effort to integrate ethical, legal, and social considerations into genomic research. Around 3% of the overall HGP funding was allocated to fund the research of ethical, legal, and societal implications of the new genomic findings, organizing public events, and developing educational materials [Kathi E. Hanna, 1995].

Several high-priority areas were explored, particularly concerning privacy, equality, and the usage and misinterpretation of genomic information [McEwen et al., 2013]. As genomic research evolved and gradually entered into clinical practice, the expected benefits revolved also around understanding its implications by medical professionals, patients, corporations, and other parties outside of the direct research field. Questions surrounding genetic testing, potential legal issues [Friedland, 1997], discrimination, ownership of genomic data [McEwen et al., 2013], and many more dominated the public discourse.

For healthcare providers, ELSI supported the development of continuing education programs focused on genetics, covering ethical considerations in genetic testing, patient counseling, and the interpretation of genetic results [Dressler et al., 2014]. An important component of this education was ensuring that patients too understood the implications of genetic tests, provided informed consent, and were supported in interpreting and acting on their results [Greely, 1998].

Last but not least, philosophical and conceptual debates were raised in light of unprecedented discoveries that the HGP brought. This included funding research to explore the implications of genetic determinism and reductionism. Public forums and ethical debates facilitated by ELSI encouraged discussion on the value of genetic diversity, the definition of health and disease in the context of genetic variation, and the ethical boundaries of genetic modification [Adams, 2016].

1.2 Significant Development Trends

The technological and scientific progress made during the HGP has undoubtedly altered both the research as well as the public discourse [Hamdoun and Ehsan, 2017]. In order to better grasp the overall societal, philosophical, and legal implications, let us now review major development trends and directions in which the research (and its perception) seems to be moving.

First and foremost, the open communication between international working groups facilitated by Bermuda principles was a significant step towards the development of an open science approach in genomic research. Other subsequent regulations and agreements were developed, notably the Fort Lauderdale Agreement (2003), the NIH Data Sharing Policy (2003), and the Toronto Statement (2009) [Arias et al., 2015].

The inception of 2nd and 3rd generation DNA sequencing methods has further accelerated the generation of enormous amounts of genomic data, showcasing the underlying need for database and algorithmic infrastructure. Sequencing technology in par-

allel is since becoming cheaper, faster, and more accessible even for use outside of academic research (direct-to-consumer genetic testing) [Ku and Roukos, 2013]). Bioinformatics and computational biology are just two examples of emerging fields that contribute to a new positioning of genetics towards a data-driven and exact field of study [ElSayed et al., 2021].

Our deep comprehension and mechanistic modeling of living organisms have served us well in recent years, allowing the development of unprecedented practical applications [López-Rubio and Ratti, 2021]). Synthetic biology and genetic engineering approaches nowadays are enabling the mass production of pharmaceuticals, designer crops, or even entire synthetically-made organisms [Gibson et al., 2010]. These technological exploits represent a paradigm shift in our perception of nature, and by extension, ourselves, bringing an entirely new set of ethical, legal, social, and philosophical problems [Bennett, 2020].

1.3 Public Perception of Genomic Research

The advent of genomic research in the 1990s has brought a notable popularization and a change in public perception of many topics related to genetics and biotechnology. Interestingly enough, the Human Genome Project itself was not at the forefront of public discourse - it was the potential applications and societal implications driving the public interest.

Most of the data reflecting public perception and understanding of genetics in the early days of HGP comes from media coverage and social surveys conducted in a specified location. Although this data is by no means definitive it can help us determine general trends and key topics discussed outside of the academic context.

The level of public understanding in this field has been determined multiple times during the 1990s - for example a study conducted by [Miller and Pifer, 1993] has shown that only 20% of participants in the US were able to construct a viable definition of the term "DNA". Other studies have focused on the perceived benefits and risks of applied genetics [Lippman, 1991], such as gene therapy, genetic screening in medical practice [Marteau and Croyle, 1998] employment, genetic engineering, DNA fingerprinting, etc. Applied genetics has shown to be a polarizing topic with certain aspects being endorsed and supported by the public, while others appeared to elicit controversy.

As shown in the figure 2.3, the "promising" subjects were associated with clinical treatment of genetic disorders, such as Down syndrome or cystic fibrosis, and with supporting jurisdiction. Therefore the positive outlook on applied genetics appeared to be centered mainly around equal social conditions and fair treatment. On the other hand the notion of genetic "improvement" or "discrimination" has been shown to cause concern in the public discourse in Britain [Durant, 1993]. According to the comparative analysis conducted by [Macer, 1992], people from the US, Europe, Japan, and New Zealand shared a positive outlook toward gene therapy while being increasingly more aware of both potential benefits and risks [Marteau, 2000].

More recent studies suggest that "genetic literacy" - knowledge of genetic concepts outside of academic research has been generally on the rise, but still has room for improvement. This trend could be attributed to the increasing number and affordability of direct-to-consumer genetic services, such as genetic screening or counseling. In the US, for instance, the majority of the population reports a high degree of support for scientific research in the field of medical genetics, while simultaneously exhibiting comparatively

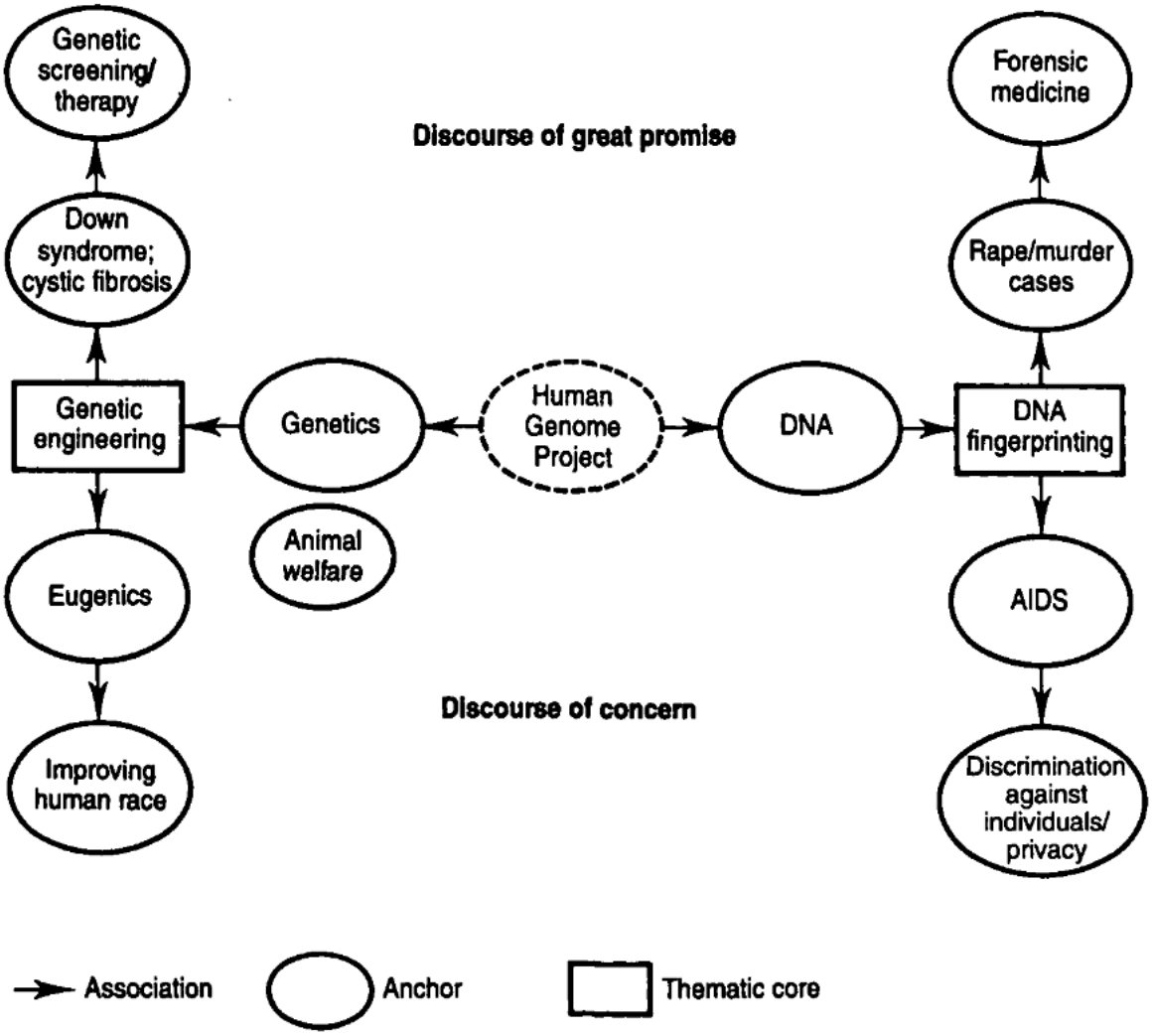

Figure 1.3: Thematic visualization of public perception of the Human Genome Project in Britain 1992 [Marteau, 2000]

low levels of factual genetic knowledge [Little et al., 2022].

According to Haga et al., the ethical and legal implications of genetics were not a significant concern for Americans. Rather it was the speed of scientific research and innovation, coupled with the fear of losing traditional values that generated the most concerns [Haga et al., 2013].

A meta-review of public knowledge and attitude toward health-related genomics conducted by [Pearce et al., 2024] has aggregated data from studies published from 2016 to 2022, including a variety of articles, reports, and surveys. The review offers us insights into multiple aspects of this relationship, including awareness, attitude, technical knowledge, psychological implications, concerns, and more. Two main vectors for increasing public awareness of genomic research and applications appear to be mass media, such as TV and the Internet, together with word-of-mouth transmission [Eum et al., 2018]. However, higher awareness was mostly accompanied by little knowledge of the full implications of genetic testing and data handling practices in medical and research fields [Riggs et al., 2019].

Among the general public in recent years, the majority feels that applications of

genetics in medical practice do help healthcare professionals to make the right decisions [Muflih et al., 2019]. Parallel to this sense of stability and trust, there also was a plethora of reported negative emotions associated mainly with the interpretation of genomic data. In one study, as much as 88% of participants with colorectal cancer or type 1 diabetes have reported induced worry, anxiety, stress, and psychological burden related to hypothetical genetic assessment [Nicholls, 2016].

As the genomics-related public discourse matured since the early days of HGP, the views and attitudes toward applied genetics in healthcare have generally remained positive. The applications, such as screening, gene therapy, D2C genome sequencing, etc. have been introduced to medical practice on a global scale, shifting the view from a perspective of promising technology to a more individually concerning topic. Research shows us that genetic literacy of the general population is rising, however many a large part of the population appears to have unrealistic expectations [Eum et al., 2018] and does not fully comprehend the meaning and implications of genetic data.

1.3.1 Research and Genetic Determinism

Genetic determinism can be characterized as a belief that genes are the sole (or at least main) causal factor in determining the characteristics of a living organism. It promotes the idea that individual characteristics are discrete and unchangeable by the environment or social setting. Genetic determinism is sometimes used interchangeably with genetic reductionism and essentialism - although these terms are related, they do not describe the same position. Genetic reductionism is more focused on explaining phenomena by reducing them to their fundamental aspects (genes) and genetic essentialism promotes a point of view in which each person has their own unchanging "essence" based on genes [Harden, 2023].

In the early 1990s, the HGP ignited hopes and expectations that unlocking the human genetic code would rapidly unravel the mysteries of diseases, behaviors, and traits. Some authors spoke of the human genome as our "blueprint" and compared the HGP to a culmination of the reductionist line of thought in molecular biology of the 1960s [Vicedo, 1992]. However, as the sequencing was approaching its end, the total number of genes was estimated to be a mere 22,500, far less than initially expected and even less than Arabidopsis thaliana (around 25,000). The belief that the complexity of an organism is directly correlated with its number of genes was therefore disproven - it was necessary to search for the cause of this complexity elsewhere. In the end, instead of confirming genetic deterministic views, the findings from HGP have refuted them to a large extent [Hub Zwart, 2007].

Contrary to popular belief, many studies have found that genetic determinism is not a widespread position in society, nor is it correlated with a certain socioeconomic status [Gericke et al., 2017]. For instance, according to [Shostak et al., 2009], a nationally representative US sample of socioeconomically advantaged people has exhibited about the same level of genetically deterministic views as other groups - that is directly connecting a specific genotype to a phenotype. Similarly, also the knowledge of the genetic field and gene-environment interactions did not appear to be correlated with deterministic views of society. The basis of determinist thinking is often established early in education, which prompts us to pay attention to the way that the basics of genetics are conveyed [Donovan et al., 2021].

Multiple sources have reported that while genetic determinism may not be that

widespread in society, it can still be reduced early on by modifying basic genetic curriculum. Teaching the classical Mendelian approach to genetics has resulted in the same level of determinist opinions as it is notable in the general population. However, the groups of students who underwent a specialized education program in genetics showed a notable decrease in determinist and essentialist thinking [Jamieson and Radick, 2017].

Chapter 2

Self-perception in the Genomic Era

2.1 Genetic Conception of Health and Physiological Identity

The post-HGP era has shuffled our understanding of health and disease to a big extent. Prior to this, health and disease had been described primarily by visible symptoms and bodily reactions to illness, from a more contemporary perspective, the borders separating the states of health and sickness are much more blurry and continuous [Canguilhem, 1989]. Genetic screening and genome sequencing as applied in clinical practice have underscored the uniqueness of our physiological states, making it difficult to classify the "normal" and "healthy".

2.1.1 Predictive and Personalized Healthcare

The 1990s, supported by the HGP have seen great success in uncovering the aetiology of many genetic diseases down to their molecular mechanics. These include Duchenne muscular dystrophy, cystic fibrosis, Huntington's disease, myotonic dystrophy, and others, growing the total of the time to about 100 - 150 known and researched diseases. Knowledge of disease causes and mechanisms has been greatly utilized in the then-emerging field of pharmacogenomics - tailored drug design and delivery based on the genetic makeup of the patient [Van Ommen, 2002].

Many diseases have been mapped to their underlying gene effects with the help of a so-called "positional cloning" method. This method allowed for effective mapping, even in cases of limited knowledge of the gene function. Apart from novel drug design, it has been argued that disease-causing genetic polymorphisms can be detected before the actual symptoms emerge, allowing for preventive therapies or lifestyle changes. However, even penetrant single-gene disorders with mendelian inheritance, such as breast and ovarian cancer do not have an absolute predictive power [Collins, 1999].

The psychological situation that patients face during the predictive healthcare process is an unprecedented one. DNA testing may reveal predispositions for diseases that are not yet effectively treatable, potentially eliciting feelings of anxiety and fear [Bondy and Mastromarino, 1997]. Now, is knowing our susceptibilities and predispositions worth the additional psychological burden? This topic is often discussed in bioethical circles as a sort of "predictive medicine dilemma".

So far genetic counseling pre and post-testing has proven to be crucial in educating

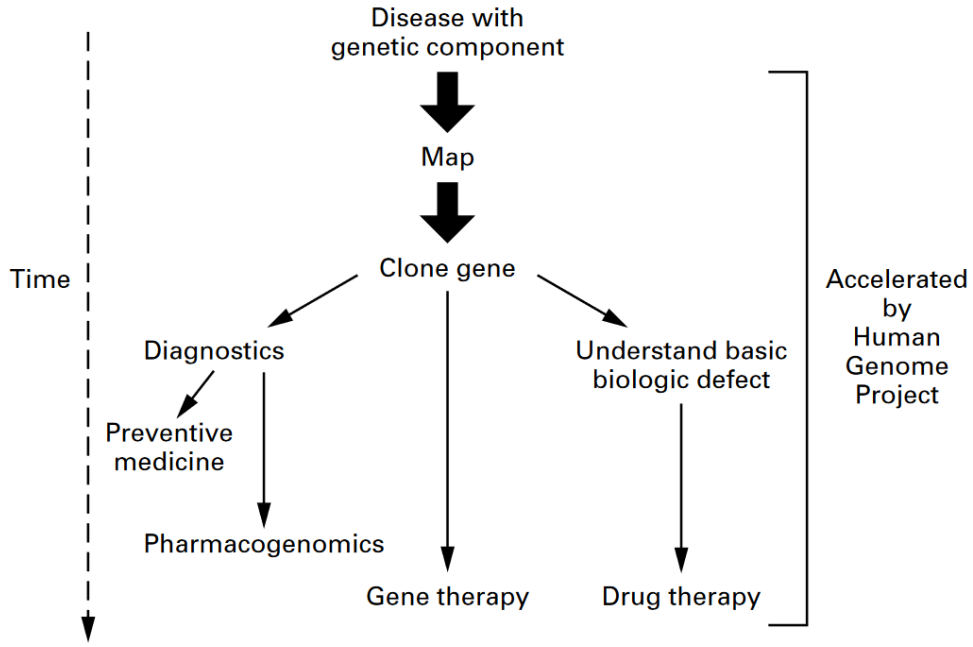

Figure 2.1: Schema of gene-disease mapping and subsequent development of individualized therapy[Collins, 1999]

patients and assessing their capability to give informed consent [De Wert, 1998]. Patients not only have to be knowledgeable of personal implications but also have to understand the influence that genetic findings may have on their biological kin, which adds another layer of ethical difficulty [Di Pietro et al., 2004].

2.1.2 Personal Identity, Normalcy, and Genetic Enhancement

Authors like Buchanan Allen et al., in "From Chance to Choice: Genetics and Justice," have significantly influenced the debate on genetic interventions, challenging the clear division between therapy and enhancement [Buchanan, 2001]. They suggest these interventions exist on a continuum, contrasting with traditional medical ethics and prompting a reassessment of genetic intervention principles. This distinction between therapy and enhancement relies to a great extent on the notion of normality to determine whether a state is pathological and therefore requires treatment [Scully and Rehmann-Sutter, 2001].

Historically, the concept of normality has been based on statistical averages and medical norms established through population health data. However, as argued by Erik Parens in "Enhancing Human Traits: Ethical and Social Implications", the advent of genetic engineering introduces a dynamic where normality is no longer a static or universally agreed-upon standard. Instead, it becomes a fluid concept, susceptible to changes in medical technology, cultural values, and individual preferences [Parens, 2007].

This position of accepting enhancements on the grounds of physical autonomy has been further developed by Nicholas Agar, in "Liberal Eugenics: In Defence of Human Enhancement". Agar embraces the potential for enhancements to extend human capabilities, advocating for a liberal approach where individuals have the freedom to choose enhancements. Agar's stance emphasizes the right of individuals to pursue augmentations as a means of self-improvement and self-expression if accompanied by a voluntary

decision to do so [Agar, 2005].

As argued by Habermas the identity of an enhanced individual may lead to existential questioning about free will and the authenticity of one's abilities and achievements, especially if the enhancements were decided and implemented by others, such as parents or guardians in the case of germline modifications[Habermas, 2008].

Michael Sandel highlights the concern that genetic enhancements could devalue personal effort by disconnecting it from the result. In a society that enables the selection and enhancement of traits, the link between effort and accomplishment may become blurred, potentially diminishing appreciation for individual achievements. Achievements could be attributed to genetic engineering rather than personal effort, elevating engineers and doctors to the role of creators of human capabilities and complicating the dynamics of achievement and recognition [Sandel, 2009].

Commercial entities developing enhancements as consumer goods could introduce market dynamics and commodification into human traits. Those could become items for purchase, influenced by trends and market demand, similar to the cosmetic surgery industry [Elliott, 2004]. This commodification risks transforming social goods into symbols of status and wealth, where the value of enhanced traits is determined by supply and demand, potentially exacerbating social divides.

The widespread diffusion of genetic enhancements could establish a new standard of perfection, rendering unenhanced traits inferior or even pathological. This shift towards a genetically engineered ideal poses significant risks to self-esteem and social cohesion. Individuals unable or unwilling to undergo enhancements may face discrimination or social exclusion, while those who do enhance may encounter greater expectations and pressures to conform to an ever-changing ideal of perfection [Brey, 2009].

2.1.3 Health Insurance and its Transformations

One of the implications of better understanding our genomic information is that we can predict to an extent whether a person is susceptible to a certain disease. This notion has helped to advance predictive medicine, however, it has also altered the paradigm of health insurance companies, potentially fostering discrimination and exclusive access to such services [Morrison, 2005].

The business of life insurance revolves largely around carefully calculated risk and long-term strategic investment. Preventive genetic testing done either by insurance companies or their customers may in theory cause a shift in risk distribution. This scenario has been known and discussed for many years before direct-to-consumer genetic testing became available [D. Rodriguez-Rincon, 2022].

For instance, the concerns about scenarios of insurance discrimination have been prominent during the Human Genome Project as Carol Lee writes in the early 1990s. The idea at the time was an image of the future, where the DNA testing of the illness markers is done directly by insurance companies in order to determine the correct fee rate for each person (and their biological kin). Certain groups might even become "uninsurable" solely on the basis of their genetic testing. Alternatively, these genetic tests might be used as a justification for different treatment of certain ethnic groups as specific diseases might be associated with their genetic heritage [Lee, 1993].

In the early 1990s insurance companies faced societal fear and accusations of potentially misusing genetic testing for reducing economic risk. The public perception of the genetics and insurance issue was emotional and misaligned with the scientific knowledge

of the time. However, to tackle this public response and negative publicity, insurance companies opted for an indifferent position regarding genetic testing and focused on educating the public and policymakers [Thomas, 2012].

This strategy stemmed from a "public deficit model", which claims that skepticism and resistance come from a lack of public understanding of the issue [Wynne, 1992]. However, the passive stance and reliance on the public deficit model has backfired in the implementation of restrictive legislation, which, in turn, challenged the foundational principles and practices of life insurance underwriting [Joly et al., 2010]. This situation forced the insurance industry to adopt a more proactive stance, resulting in various national regulatory compromises [Van Hoyweghen et al., 2005].

A review conducted by [Joly et al., 2013] suggests that the individual cases of genetic discrimination in life insurance revolve mainly around five well-studied hereditary conditions, namely Huntington's disease, breast and ovarian cancer, hemochromatosis, hypercholesterolemia and colorectal cancer. The number of individual discrimination cases, however, was small, which could be attributed to occasional errors rather than systematic exploitation.

From patients'/insurers' point of view, the information about potential discrimination may influence their will to undertake genetic testing. In a study working with colorectal cancer patients from 1999 to 2006 less than half reported that they would undergo genetic testing with the knowledge of potential discrimination in the field of health insurance. This suggests that the fear of discrimination may potentially discourage people from undergoing genetic testing, even with the benefit of cost-effective and early detection of diseases [Keogh et al., 2009].

Overall the knowledge of potential genetic discrimination in life and health insurance has been discussed in an emotional public debate since the early 1990s. This concern gave birth to a restrictive regulation prohibiting unequal treatment dependent on hereditary information in many parts of the world. Actual cases in which these concerns have manifested are happening, however, their consequences are not by far comparable to the hypothetical scenarios painted during the 1990s.

2.2 Genomic Research and Cultural Ethnicity

Cultural ethnicity, race, ancestry, and similar expressions have been somewhat controversial in the context of genetic and health-related research. The attempt to objectively define and aggregate certain subgroups in a population based on their genetic information is (1) often in conflict with the traditional sociocultural conception of ethnicity and (2) has historically led to societal stratification [Kevles, 2004]. The tension arises primarily because genetic data can reveal connections and distinctions among populations that may not align with sociocultural identities or self-perceptions [Foster and Sharp, 2002].

In the post-HGP era, this tension has prompted researchers and policymakers to acknowledge this differentiation by implementing terminology and research procedures that respect both sociocultural individuality as well as new genetic discoveries. In this section, I will further focus on examining these distinctions in perceptions of cultural ethnicity, how they relate to each other, and how they have changed since the early 1990s.

2.2.1 Sociocultural and Genetic Identity Dichotomy

Genetic literature and public discourse still partially treat ethnic groups as a scientific category, rather than a sociocultural construct [Foster and Sharp, 2002]. Genetically viewed ethnic groups are usually determined by ancestral lineage and shared genetic markers, as well as phenotypical traits.

In the sociocultural context, however, ethnicity is conceptualized as a complex, constantly negotiated structure that encompasses shared history, culture, language, and social practices among groups. This conception extends beyond mere biological or genetic markers, instead, it is viewed as an identity continuously negotiated and reconstructed through social interactions and historical contexts [Nagel, 1994].

After the completion of the Human Genome Project, new research findings prompted a discussion about racial and ethnic terminology of the time. It was clear that the established categories carried very little biological or anthropological meaning, suggesting that a new framework was needed to reflect these findings [Oppenheimer, 2001]. Some authors argue that self-identified race might serve as useful data for determining genetic homogeneity and different health predispositions [Foster and Sharp, 2002]. Others are opposed to using these self-identified categories in biomedical research as they rely more on individual perception and a sense of belonging rather than common ancestral lineage [Cooper et al., 2003] [Tishkoff and Kidd, 2004].

As of the current state of biomedical research and clinical practice, the most frequently used racial categories for self-identification come from the US Office of Management and Budget (OMB) directive NO. 15 - this directive specifies the following ethnoracial groups: American Indian or Alaska Native, Asian, Black or African American, Native Hawaiian or other Pacific Islander, or White, and Hispanic/Latino or not Hispanic/Latino [of Management and Budget, 1977].

The US Institute of Medicine has also explicitly added that races should not be considered a biological reality, but instead "a construct of human variability based on perceived differences in biology, physical appearance, and behavior" [Smedley, 2003]. While not inherently biological nor anthropological, these categories have become a sort of gold standard in biomedical research, enabling large-scale epidemiological studies and exploring new hypotheses in population genomics [Burchard et al., 2003].

Some Implications of Genetic Ethnoracial Misappropriation

In academic research within the social and biological sciences, the use of poorly defined racial categories can introduce bias, skewing results and leading to incorrect conclusions. When racial constructs are mistaken for a biological reality, the research results pose limited generalizability and might be otherwise misleading [Burchard et al., 2003]. This practice may perpetuate pseudoscientific notions and distort public perceptions of genetic differences and similarities among human populations, potentially leading to deterministic and essentialist bias [Bryant et al., 2022].

Furthermore, in clinical practice, genetic predispositions based on race can lead to both over-treatment and under-treatment. For instance, equations used to determine kidney function based on serum creatinine (e.g. Cockcroft-Gault equation) take into account a racially dependent coefficient. This means that wrongfully attributing patients' race could lead to obscured calculations and clinical mistreatment [Powe, 2020]. Such excessive focus on race can obscure more pertinent factors like environment, lifestyle, and individual genetics, diverting attention from the need for personalized care and leading

to a generalized approach where nuanced differences in patient histories and conditions are overlooked.

2.2.2 Ancestry, Belonging, and Narrative Identity

The emergence of direct-to-consumer genealogical DNA tests or genetic ancestry tests (GATs) during the last 20 years has introduced a significant shift in personal narrative identity and a sense of belonging [Royal et al., 2010]. The psychological effects of receiving ancestral results in some cases call for a reevaluation of personal identity, often accompanied by internal conflict [Theunissen, 2022]. In this sense, genealogical DNA tests can act as a probe to examine and challenge one's own narrative identity, however, as a growing body of evidence suggests, GATs may also have little or nothing to do with how a person identifies ethnically [Shim et al., 2018].

Most GAT companies refrain from using the highly politicized term "race" and instead opt for a seemingly more neutral term "Ancestry". Representatives from deCodeme and 23andMe stated that ancestry tests do not determine race, they simply provide data about shared markers with reference population and it is primarily up to the individual to make a sense of these data, potentially identifying with a certain ethnoracial identity [Lee, 2013].

Narrative identity is crafted through personal experiences, cultural contexts, and social interactions. It significantly involves how people understand and internalize aspects of their ethnoracial backgrounds into their self-concepts. This integration often reflects broader societal narratives about race and ethnicity and can influence an individual's feelings of belonging and identity [Hammack, 2008]. The coherence of narrative identity, including how individuals relate their ethnoracial identities to their life stories, is linked to psychological well-being. Those who can articulate a clear and positive connection between their ethnoracial backgrounds and their identity often experience better mental health and a stronger sense of self [Adler, 2012].

The influence of GAT results has been studied primarily through the medium of qualitative interviews with participants. For example, [Theunissen, 2022] has found that the core topics of participants undergoing GATs include family belonging, genetic family connection, national pride, cultural affinity, and seeking answers. Some participants reported a sense of identity crisis elicited by unexpected results, such as a mismatch with expected biological family members. Others described feelings of emptiness, which could be described as genealogical bewilderment [Leighton, 2012] coupled with a motivation to seek answers and learn more about their new identity contribution.

Customers undergoing 23andMe GATs reported a sense of disappointment that genetic tests did not provide them with as much information about their ancestry as they hoped for. They discovered that the information was not ready-made, but rather required additional work to make sense of [Lee, 2013]. The participants of interviews conducted by Shim et. al. also reported some degree of negative emotions surrounding the tests. This study separates participants into four groups regarding their overall experience with GAT results. Out of these four groups, only one generally felt an enhanced sense of belonging, individuality, and ethnoracial identity. The others did not observe a significant impact of GAT results on their perception of narrative identity. For example, they regarded the results as an artifact from a very distant past, which is somewhere "in them", but is not directly influencing them. Many participants also felt that the GAT results did not bring any additional information, but instead, they confirmed certain narrative beliefs that the

participants already held and knew about themselves [Shim et al., 2018].

To conclude, in some cases, the confrontation with direct genealogical results may prompt a renegotiation of one's narrative identity coupled with an internal conflict, feelings of emptiness, and a motivation to seek more answers. However what most participants report suggests a strong prevalence of sociocultural constitution of their ethnoracial identification - GAT results therefore have only a miniscule effect on the narrative identity itself.

2.3 Genomic Rights and Society

In previous chapters, we have discussed how genomic advancements relate to health and sickness, as well as personal narrative identity. Now I would like to shift our attention to broader societal implications that come with the increased availability of genomic data - this chapter will explore privacy and ownership of genomics information, surveillance, personal autonomy, and liability.

2.3.1 Privacy and Ethical Problems of Genetic Information

By genomic data, we understand either a complete nucleotide sequence (e.g., autosomal, mitochondrial) or a set of specific polymorphisms of an individual. During and after the HGP the collection of such data has become increasingly more effective and cost-efficient, which resulted in a data abundance and some ethical concerns. As of this time, genomic data is usually collected by several entities with different motivations. A common scenario includes genetic testing in a clinical setting, where the data is used for diagnostic purposes and the development of personalized treatment plans. In other cases, the samples may be collected by commercial entities, such as GAT (genetic ancestry testing) or preventive healthcare-oriented companies [Naveed et al., 2015].

However, the highest amount of genomic information is gathered for the purpose of biomedical research, such as GWAS (Genome-wide Association Studies). Data from various sources may be shared and reused - for example, biological samples and genomic data gathered from patients may be further stored in databases or biobanks to be used in additional research [McEwen et al., 2013].

Genomic data, contrary to other types of health data can be reconstructed to reveal a significant portion of phenotypic information about the individual [Ury, 2013]. They are also immutable and unambiguously determining. For these reasons, an ethical discussion around privacy and ownership has emerged [Naveed et al., 2015].

A review of this discussion, particularly around the problem of biobanks, has been conducted by [Budimir et al., 2011], highlighting the main concerns and dilemmas. One of the most discussed topics was informed consent for participation in research. Informed consent ensures that participants know exactly what can and cannot happen to their data and biological samples. It protects their privacy, articulates rights, and signifies voluntariness. Most authors agree that participants should have the right to withdraw their consent at any time and not participate in any further research [Helgesson and Johnsson, 2005], however, there is no consensus over how exactly this withdrawal should be executed [Cambon-Thomsen et al., 2007].

The specific content of the consent form is also subject to changes and discussion with some authors offering their own versions of universal standardized consent [Beskow et al., 2010].

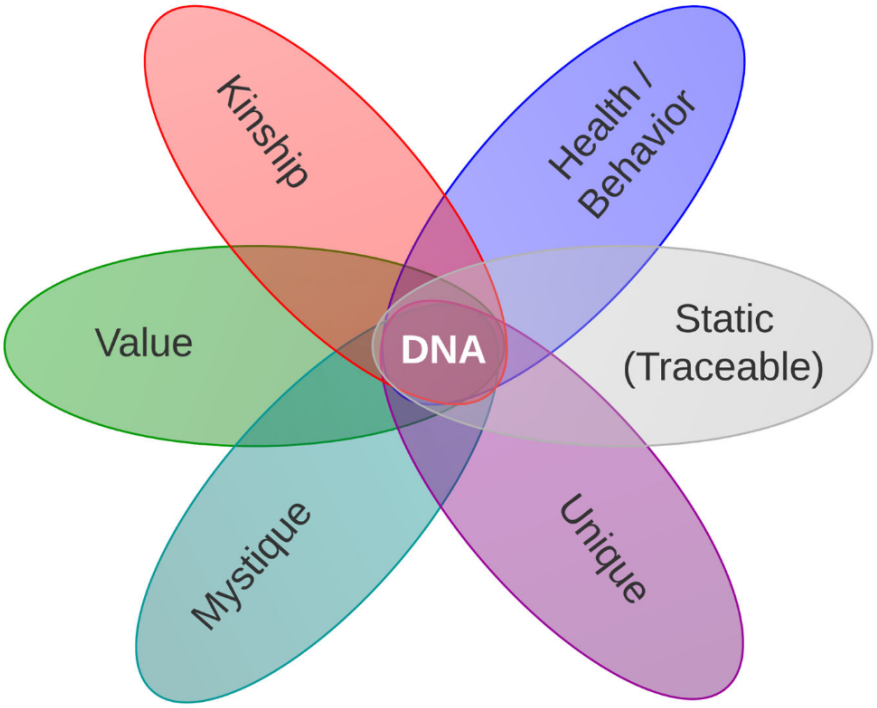

Figure 2.2: Unique properties of genomic data that distinguish it from other types of medical data. It contains information about a person's medical state (health) and their kinship relations. It is unique for each person, does not change over time (static), and is a valuable information source for others (value). Mystique refers to public perception of DNA as something mysterious [Naveed et al., 2015].

Currently, however, there is no universal version, because of varying research scenarios and different sociocultural predispositions. A broad consent is suggested by many authors to be the most applicable for participating in further research. Although broad consent does not specify every scenario down to the last detail, it lays the groundwork for undisclosed research that may or may not happen in the future. If a research would occur, it must: be "of great importance", provide maximum privacy protection, and be approved by an ethical review board (ERB) [Hansson et al., 2006].

Using anonymous or anonymized samples and data would in theory provide the best participant protection, however, this practice seriously limits research use cases, breaks the link between phenotype and genotype, and last but not least makes returning research results to individuals impossible [Eriksson and Helgesson, 2005]. Many authors therefore refuse complete anonymization and instead rely on encrypted information and gated databases to preserve the link between sample and data, albeit in a more secure form. Privacy in particular has shown to be a significant concern of potential research participants [Kaufman et al., 2009].

More ethical difficulties appear when minors or incompetent adults are involved in genomic studies. These groups are largely disadvantaged in their capacity to provide informed consent - because of this, biobanks tend not to involve them, which in turn can cause medical research to be slower in this population segment [Budimir et al., 2011]. Discussion on this topic is polarized, although many authors advocate for involvement with extra protection and risk minimization [Hens et al., 2009].

Incidental findings are another highly discussed topic regarding genomic data. Do

researchers have the obligation to report and return results to participants? As of this time, most authors agree that returning results individually is impractical, especially in large-scale population studies. Exception can be argued in a case of very high clinical importance - potential health issues, carrier status for genetic conditions, etc. Ethical obligations surrounding such findings are higher and it is therefore advocated that results should be returned and professionally communicated [Greely, 2007].

Outside of research and healthcare, genomic data is frequently used for forensic purposes - identification and exclusion of suspects by comparing SNPs (Single Nucleotide Polymorphisms) of different samples. Such a use case naturally sparks questions about data privacy and personal liberty. Should authorities be able to store an indexed database of genetic fingerprints connected directly to personal identity? What is the best way of manipulating and storing genomic data for forensic purposes without infringing on the privacy of the suspect's kin? Should sample comparing be done only suspect against crime scene or crime scene sample against a wide genomic database? [Stajano et al., 2008] These are just some of the open-ended questions to illustrate that the problem of genomic data storage and usage is not exclusive to research or healthcare and also occurs in other fields.

Gathering, storage, manipulation, and storage of biological samples and genomic data is a sensitive issue concerning the privacy and ethical rights of potentially entire populations. To summarize, the main concerns and dilemmas in this regard include: the scope of consent, preserving participants' privacy, reporting of incidental findings, and the involvement of minors in research. Currently established processes seem to be a constantly negotiated equilibrium balancing participants' rights and research interests.

2.3.2 Genetics and Criminal Behaviour

Attributing traits and behaviors to specific genotypes is becoming increasingly more complex, due to the influence of environment, epigenetics, and other emergent genetic properties. This is, on one hand, a research problem, however, it is in the context of ELSI (ethical, legal, and societal implications) that this unclear attribution starts to create moral dilemmas and ambiguous situations.

Behavioral genetics has seen great advances in tackling this problem under the influence of molecular approaches largely developed during and after the HGP. Since its inception in the early 20th century, behavioral genetics has been intertwined with the eugenics movement and later during World War II used predominantly to study and track the inheritance of favorable phenotypes. In what we could call a "post-eugenic shift" towards the end of the 20th century, the field of behavioral genetics is distancing itself from its controversial past, emphasizing a more nuanced (gene-environment interactions, epigenetics, etc.) and less mechanistic approach [Allen, 1997].

Together with advances in genomics, there appears to be an increasing number of criminal case defenses built on top of certain genetic predispositions for violent behaviors. Such attempts have been present since the 1970s, although generally unsuccessful. For a behavior to be considered criminal, two key elements have to take place - *mens rea* and *actus reus*, i.e. the guilty state of mind (intent) and the guilty act. Genetic evidence has been used both to mitigate *mens rea*, for instance by establishing legal insanity [Farahany and Jr, 1969], and to disprove actus reus by diminishing personal liability for one's actions [Berryessa and Cho, 2013].

One highly publicized case is that of Stephen Mobley [Supreme Court of Georgia, 1995],

convicted of murdering a 25-year-old Domino's pizza store manager John C. Collins in 1991. His defense attempted to appeal his death sentence by presenting evidence of a family history of behavioral disorders and a possible connection of mutation in the MAOA gene (Monoamine oxidase A) to increased violence predisposition. This mutation would possibly render Mobley less responsible for his violent behavior, therefore mitigating his sentence. However, the appeal was rejected by the Georgia Supreme Court and Mobley was in February 1994 sentenced to death, setting a precedent on the limitations of genetic evidence in mitigating criminal responsibility. Testing for MAOA mutation was not permitted by the court based on the lack of a proven causal link between MAOA mutation and violent behavior [Denno, 2007].

A review conducted by [Raine, 2008] has clarified the relationship of MAOA gene polymorphisms in predisposing individuals to antisocial and violent behaviors. MAOA is crucial for the breakdown of neurotransmitters such as serotonin, which are often found at lower levels in antisocial individuals. Notably, specific variants of the MAOA gene are linked to reduced volumes in critical brain regions including the amygdala, anterior cingulate, and orbitofrontal cortex - areas involved in emotional regulation. These structural impairments contribute to the heightened risk of antisocial behavior, emphasizing the profound impact of genetic and neurobiological factors in shaping such behaviors. This gene-brain-behavior pathway highlights the importance of considering genetic influences when assessing predispositions to antisocial conduct. Another case utilizing the MAOA defense is a 2009 murder trial of Bradley Waldroup [Carroll L. Ross, 2011], who had shot and killed a friend of his spouse, Leslie Bradshaw, and later attacked his wife with a machete. Waldroup's defense highlighted that he carried a polymorphism of the MAOA linked to aggressive behavior under stress. Experts testified that this genetic predisposition, exacerbated by a troubled upbringing, could impair emotional regulation and impulse control. The jury, influenced by these arguments, convicted Waldroup of voluntary manslaughter and attempted second-degree murder, rather than first-degree murder, resulting in a 32-year sentence instead of life imprisonment or the death penalty [Aiello, 2021].

The case of Bradley Waldroup represents one of the few, where the appeal on genetic predisposition was successful in mitigating the sentence. It is also important to note that genetic arguments are generally used in correspondence with other mitigating conditions, such as history of abuse, environmental factors, and so on. According to [Berryessa and Cho, 2013] it appears that judges are slowly getting more receptive toward some degree of genetic defense in the court, however, this receptivity is still relatively low. Behavioral genetics at this time is likely not disrupting the legal system, but instead, it is questioning the notion of predetermination and free will in the face of criminal actions.

Discussion and Conclusion

In this review, I have discussed how key research and development trends in new genetics are reflected in the public discourse, as well as on the individual level. The Human Genome Project has played a crucial role in establishing genomics as a kind of comparative data-driven field, much more similar to hard sciences like physics. We can argue that, unlike the strictly mechanistic and deterministic understanding of early molecular biology, the nature of this field has become a lot more systemic and probabilistic. The public discourse on genetics seems to be particularly keen on the deterministic view of things, sparking numerous concerns about the implications of such a "disruptive" field. Often medialized topics of concern include genetic engineering, eugenics, and discrimination based on ethnicity or disability. Some of these concerns are in place, however, it is notable that the actual academic discussion is preoccupied with different topics.

Despite these concerns and a relatively low level of understanding, the public support for genomic research is immense and positive. It seems like the perceived potential benefits and applications outweigh catastrophic scenarios. In healthcare, for instance, the potential for creating value is vast and the public seems to notice it. It is also a field that is intimately connected to patients' well-being and self-perception by identifying the "pathological". As it appears from the literature, the traditional binary distinction of "healthy" or "sick" is largely disrupted by the systemic nature of genomics, shifting our perception more towards a "continuum" model of health and sickness. Stepping away from the generalized into the individualized could help patients get rid of harmful stereotypes and narratives, potentially empowering their self-perception and encouraging acceptance, however, this approach also does have its setbacks notably in the practical healthcare implementation and ethical concerns.

Bioethics in genetic research is a particularly discussed topic - much more so in academic circles rather than in the public discourse.From reviewing the literature a lot of the bioethical problems centered around genetic information stem from the fact that the genome holds a great amount of potentially highly confidential and specific information - disease predisposition, ancestry information, behavioral predisposition, and so on. On that note, there has to be a certain level of lay genomic literacy to grasp the full implications of genetic testing be it for healthcare or genealogical purposes. Modern-day consent forms seem to emphasize this kind of factual understanding, which could help mitigate misuse attempts and other unwanted consequences.

Additionally, the commercialization of genetic technologies has introduced a market-driven aspect to genetic testing, where companies offer personalized genetic insights directly to consumers. This commercialization raises ethical questions about the commodification of human genetic information and the potential exacerbation of social inequalities, as access to genetic technologies may reinforce socio-economic disparities. Luckily, actual cases of genetic discrimination since the 1990s have been rare - this could be the result of specific anti-discrimination policies reflecting on ethical and societal discussions.

These reflections reveal that the discourse surrounding molecular genetics is embedded within a larger socio-ethical context that extends beyond scientific laboratories. Public engagement with genetics is mediated through a lens colored by hope, skepticism, and the pursuit of identity, highlighting the dual promise and peril of these technologies.

Summary

The Human Genome Project was an internationally collaborative initiative funded to a large extent by the US government. It began in 1990 and the first almost complete sequence of the human genome was published in 2001. Sequencing technology progressed rapidly during the 1990s, introducing the capillary Sanger sequencing method and automated sequencers. The HGP generated vast amounts of genomic data, which prompted advancements in bioinformatics, such as genetic databases (e.g. GenBank) or assembly algorithms.

This era also saw the step towards open science, underpinned by the Bermuda Principles which advocate for data sharing and international collaboration. As a result, biology has been transitioning towards a data-driven, systems-leaning, and probabilistic field, moving away from a solely reductionistic understanding of biological processes.

The Ethical, Legal, and Social Implications (ELSI) program of the HGP underscored concerns that genomic data, while holding immense research potential, also poses significant risks related to privacy infringements, liberty limitations, discrimination, and essentialism.

There is a growing, albeit still limited, understanding of genetics among the general population, accompanied by strong support for genomic research, especially in fields like healthcare and forensics. Despite this support, there are notable concerns regarding the implications of human enhancement and genetic discrimination. Genetic determinism, the belief that solely genes determine traits and behaviors, is not prevalent in the general population, nor is it commonly linked to socioeconomic status. However, deterministic beliefs can be mitigated even further by proper education.

Genetic health assessments represent an ongoing shift towards predictive and personalized healthcare, including pharmacogenomics. This shift is built upon an individualistic understanding of health and sickness, which are not seen as distinct states, but rather as a continuum. Genetic assessments on ethical and psychological levels affect not just the individual but their entire kin. Clear distinctions between therapeutic interventions and enhancements can become blurred. Enhancements, particularly, raise ethical concerns about the commodification of genetic features and the potential societal stratification they could engender.

Ethnically and culturally, the intersection of traditional racial categories with genetic findings poses challenges. Over-generalization can obscure scientific findings and lead to medical mistreatments, whereas excessive individualization might congest research and prevent generalizable results.

The reevaluation of personal identity, influenced by genealogical test results (GATs), can lead to identity crises and estrangement, particularly when genetic information provides limited insights. Such revelations are not conclusive but are intended for further exploration. Genealogical test results have shown only a minuscule effect on one's narrative identity.

In the societal and legal arenas, genomic data, which is unique and unchanging, raises significant ethical questions about its collection, use, and storage. Healthcare institutions and for-profit ventures usually gather this data. The issues of informed consent, data anonymization, and the balance between privacy and research utility remain open-ended and are subject to negotiation. The encryption and guarded handling of genomic data offer a compromise, though the reporting of incidental findings remains a challenging ethical dilemma, especially concerning the criteria for clinical importance.

Furthermore, the interplay between genetics and criminal behavior has emerged in legal defenses, sometimes citing genetic predispositions, like mutations in the MAOA gene, to argue reduced personal liability. While such defenses have not been broadly successful, there are instances where they have influenced judicial outcomes, indicating an increasing receptivity among judges to genetic arguments as the research progresses.

Bibliography

[Adams, 2016] Adams, T. (2016). Annotated Scholarly Guide to the HGP.

[Adler, 2012] Adler, J. M. (2012). Living into the story: Agency and coherence in a longitudinal study of narrative identity development and mental health over the course of psychotherapy. *Journal of Personality and Social Psychology*, 102(2):367–389.

[Agar, 2005] Agar, N. (2005). *Liberal eugenics: in defence of human enhancement*. Blackwell, Malden, MA.

[Aiello, 2021] Aiello, T. (2021). MAOA Genetics in the Courtroom: The Last Legacy of State v. Waldroup. Publisher: [object Object].

[Allen, 1997] Allen, G. E. (1997). The social and economic origins of genetic determinism: a case history of the American Eugenics Movement, 1900–1940 and its lessons for today. *Genetica*, 99(2-3):77–88.

[Anderson, 1981] Anderson, S. (1981). Shotgun DNA sequencing using cloned DNase I-generated fragments. *Nucleic Acids Research*, 9(13):3015–3027.

[Arias et al., 2015] Arias, J. J., Pham-Kanter, G., and Campbell, E. G. (2015). The growth and gaps of genetic data sharing policies in the United States. *Journal of Law and the Biosciences*, 2(1):56–68.

[Bennett, 2020] Bennett, G., editor (2020). *The ethics of biotechnology*. Routledge, London, 1st edition. OCLC: 1295179399.

[Berryessa and Cho, 2013] Berryessa, C. M. and Cho, M. K. (2013). Ethical, Legal, Social, and Policy Implications of Behavioral Genetics. *Annual Review of Genomics and Human Genetics*, 14(1):515–534.

[Beskow et al., 2010] Beskow, L. M., Friedman, J. Y., Hardy, N. C., Lin, L., and Weinfurt, K. P. (2010). Developing a Simplified Consent Form for Biobanking. *PLoS ONE*, 5(10):e13302.

[Bondy and Mastromarino, 1997] Bondy, M. and Mastromarino, C. (1997). Ethical issues of genetic testing and their implications in epidemiologic studies. *Annals of Epidemiology*, 7(5):363–366.

[Brey, 2009] Brey, P. (2009). Human Enhancement and Personal Identity. In Olsen, J. K. B., Selinger, E., and Riis, S., editors, *New Waves in Philosophy of Technology*, pages 169–185. Palgrave Macmillan UK, London.

[Bryant et al., 2022] Bryant, B. E., Jordan, A., and Clark, U. S. (2022). Race as a Social Construct in Psychiatry Research and Practice. *JAMA Psychiatry*, 79(2):93.

[Buchanan, 2001] Buchanan, A. E. (2001). *From chance to choice: genetics and justice*. Cambridge University Press, Cambridge. OCLC: 1181611727.

[Budimir et al., 2011] Budimir, D., Polašek, O., Marušić, A., Kolčić, I., Zemunik, T., Boraska, V., Jerončić, A., Boban, M., Campbell, H., and Rudan, I. (2011). Ethical aspects of human biobanks: a systematic review. *Croatian Medical Journal*, 52(3):262–279.

[Burchard et al., 2003] Burchard, E. G., Ziv, E., Coyle, N., Gomez, S. L., Tang, H., Karter, A. J., Mountain, J. L., Pérez-Stable, E. J., Sheppard, D., and Risch, N. (2003). The Importance of Race and Ethnic Background in Biomedical Research and Clinical Practice. *New England Journal of Medicine*, 348(12):1170–1175.

[Cambon-Thomsen et al., 2007] Cambon-Thomsen, A., Rial-Sebbag, E., and Knoppers, B. M. (2007). Trends in ethical and legal frameworks for the use of human biobanks. *European Respiratory Journal*, 30(2):373–382.

[Canguilhem, 1989] Canguilhem, G. (1989). *The normal and the pathological*. Zone Books, New York.

[Carroll L. Ross, 2011] Carroll L. Ross (2011). STATE OF TENNESSEE v. DAVIS BRADLEY WALDROUP, JR.

[Collins, 1999] Collins, F. S. (1999). Medical and Societal Consequences of the Human Genome Project. *New England Journal of Medicine*, 341(1):28–37.

[Collins et al., 2003] Collins, F. S., Morgan, M., and Patrinos, A. (2003). The Human Genome Project: Lessons from Large-Scale Biology. *Science*, 300(5617):286–290.

[Cook-Deegan and McGuire, 2017] Cook-Deegan, R. and McGuire, A. L. (2017). Moving beyond Bermuda: sharing data to build a medical information commons. *Genome Research*, 27(6):897–901.

[Cooper et al., 2003] Cooper, R. S., Kaufman, J. S., and Ward, R. (2003). Race and Genomics. *New England Journal of Medicine*, 348(12):1166–1170.

[D. Rodriguez-Rincon, 2022] D. Rodriguez-Rincon (2022). *Assessing the impact of developments in genetic testing on insurers' risk exposure*. RAND Corporation.

[Das et al., 2001] Das, M., Burge, C. B., Park, E., Colinas, J., and Pelletier, J. (2001). Assessment of the Total Number of Human Transcription Units. *Genomics*, 77(1-2):71–78.

[De Wert, 1998] De Wert, G. (1998). Ethics of predictive DNA-testing for hereditary breast and ovarian cancer. *Patient Education and Counseling*, 35(1):43–52.

[Denno, 2007] Denno, D. W. (2007). Legal Implications of Genetics and Crime Research. In Bock, G. R. and Goode, J. A., editors, *Novartis Foundation Symposia*, pages 248–264. Wiley, 1 edition.

[Di Pietro et al., 2004] Di Pietro, M., Giuli, A., and Spagnolo, A. (2004). Ethical implications of predictive DNA testing for hereditary breast cancer. *Annals of Oncology*, 15:i65–i70.

[Donovan et al., 2021] Donovan, B. M., Weindling, M., Salazar, B., Duncan, A., Stuhlsatz, M., and Keck, P. (2021). Genomics literacy matters: Supporting the development of genomics literacy through genetics education could reduce the prevalence of genetic essentialism. *Journal of Research in Science Teaching*, 58(4):520–550.

[Dressler et al., 2014] Dressler, L. G., Jones, S. S., Markey, J. M., Byerly, K. W., and Roberts, M. C. (2014). Genomics Education for the Public: Perspectives of Genomic Researchers and ELSI Advisors. *Genetic Testing and Molecular Biomarkers*, 18(3):131–140.

[Durant, 1993] Durant, J. (1993). The Human Genome Project and the British public. 1993.

[Elliott, 2004] Elliott, C. (2004). *Better than well: American medicine meets the American dream*. Norton, New York, 1. publ. in paperback edition.

[ElSayed et al., 2021] ElSayed, I. A., ElDahshan, K., Hefny, H., and ElSayed, E. K. (2021). Big Data and its Future in Computational Biology: A Literature Review. *Journal of Computer Science*, 17(12):1222–1228.

[Eriksson and Helgesson, 2005] Eriksson, S. and Helgesson, G. (2005). Potential harms, anonymization, and the right to withdraw consent to biobank research. *European Journal of Human Genetics*, 13(9):1071–1076.

[Eum et al., 2018] Eum, H., Lee, M., Yoon, J., Cho, J., Lee, E. S., Choi, K. S., Lee, S., Jung, S.-Y., Lim, M. C., Kong, S.-Y., and Chang, Y. J. (2018). Differences in attitudes toward genetic testing among the public, patients, and health-care professionals in Korea. *European Journal of Human Genetics*, 26(10):1432–1440.

[Farahany and Jr, 1969] Farahany, N. A. and Jr, J. E. C. (1969). Genetics and Responsibility: To Know the Criminal From the Crime. *LAW AND CONTEMPORARY PROBLEMS*, 69.

[Foster and Sharp, 2002] Foster, M. W. and Sharp, R. R. (2002). Race, Ethnicity, and Genomics: Social Classifications as Proxies of Biological Heterogeneity. *Genome Research*, 12(6):844–850.

[Friedland, 1997] Friedland, S. I. (1997). The criminal law implications of the Human Genome Project: reimagining a genetically oriented criminal justice system. *Kentucky Law Journal (Lexington, Ky.)*, 86(2):303–366.

[Gericke et al., 2017] Gericke, N., Carver, R., Castéra, J., Evangelista, N. A. M., Marre, C. C., and El-Hani, C. N. (2017). Exploring Relationships Among Belief in Genetic Determinism, Genetics Knowledge, and Social Factors. *Science & Education*, 26(10):1223–1259.

[Gibson et al., 2010] Gibson, D. G., Glass, J. I., Lartigue, C., Noskov, V. N., Chuang, R.-Y., Algire, M. A., Benders, G. A., Montague, M. G., Ma, L., Moodie, M. M., Merryman, C., Vashee, S., Krishnakumar, R., Assad-Garcia, N., Andrews-Pfannkoch, C., Denisova, E. A., Young, L., Qi, Z.-Q., Segall-Shapiro, T. H., Calvey, C. H., Parmar, P. P., Hutchison, C. A., Smith, H. O., and Venter, J. C. (2010). Creation of a Bacterial Cell Controlled by a Chemically Synthesized Genome. *Science*, 329(5987):52–56.

[Greely, 1998] Greely, H. T. (1998). LEGAL, ETHICAL, AND SOCIAL ISSUES IN HUMAN GENOME RESEARCH. *Annual Review of Anthropology*, 27(1):473–502.

[Greely, 2007] Greely, H. T. (2007). The Uneasy Ethical and Legal Underpinnings of Large-Scale Genomic Biobanks. *Annual Review of Genomics and Human Genetics*, 8(1):343–364.

[Green, 1997] Green, P. (1997). Against a Whole-Genome Shotgun. *Genome Research*, 7(5):410–417.

[Habermas, 2008] Habermas, J. (2008). *The future of human nature*. Polity, Cambridge.

[Haga et al., 2013] Haga, S. B., Barry, W. T., Mills, R., Ginsburg, G. S., Svetkey, L., Sullivan, J., and Willard, H. F. (2013). Public Knowledge of and Attitudes Toward Genetics and Genetic Testing. *Genetic Testing and Molecular Biomarkers*, 17(4):327–335.

[Hamdoun and Ehsan, 2017] Hamdoun, Z. and Ehsan, H. (2017). Aftermath of the Human Genome Project: an era of struggle and discovery. *TURKISH JOURNAL OF BIOLOGY*, 41:403–418.

[Hammack, 2008] Hammack, P. L. (2008). Narrative and the Cultural Psychology of Identity. *Personality and Social Psychology Review*, 12(3):222–247.

[Hansson et al., 2006] Hansson, M. G., Dillner, J., Bartram, C. R., Carlson, J. A., and Helgesson, G. (2006). Should donors be allowed to give broad consent to future biobank research? *The Lancet Oncology*, 7(3):266–269.

[Harden, 2023] Harden, K. P. (2023). Genetic determinism, essentialism and reductionism: semantic clarity for contested science. *Nature Reviews Genetics*, 24(3):197–204.

[Heather and Chain, 2016] Heather, J. M. and Chain, B. (2016). The sequence of sequencers: The history of sequencing DNA. *Genomics*, 107(1):1–8.

[Helgesson and Johnsson, 2005] Helgesson, G. and Johnsson, L. (2005). The Right to Withdraw Consent to Research on Biobank Samples. *Medicine, Health Care and Philosophy*, 8(3):315–321.

[Hens et al., 2009] Hens, K., Nys, H., Cassiman, J., and Dierickx, K. (2009). Genetic research on stored tissue samples from minors: A systematic review of the ethical literature. *American Journal of Medical Genetics Part A*, 149A(10):2346–2358.

[Hub Zwart, 2007] Hub Zwart, N. (2007). Genomics and self-knowledge: implications for societal research and debate. *New Genetics and Society*, 26(2):181–202.

[IHGSC, 2001] IHGSC (2001). Initial sequencing and analysis of the human genome. *Nature*, 409(6822):860–921.

[Jamieson and Radick, 2017] Jamieson, A. and Radick, G. (2017). Genetic Determinism in the Genetics Curriculum: An Exploratory Study of the Effects of Mendelian and Weldonian Emphases. *Science & Education*, 26(10):1261–1290.

[Joly et al., 2010] Joly, Y., Braker, M., and Le Huynh, M. (2010). Genetic discrimination in private insurance: global perspectives. *New Genetics and Society*, 29(4):351–368.

[Joly et al., 2013] Joly, Y., Ngueng Feze, I., and Simard, J. (2013). Genetic discrimination and life insurance: a systematic review of the evidence. *BMC Medicine*, 11(1):25.

[Kathi E. Hanna, 1995] Kathi E. Hanna (1995). The Ethical, Legal, and Social Implications Program of the National Center for Human Genome Research: A Missed Opportunity? In *Society's Choices: Social and Ethical Decision Making in Biomedicine*, volume 1995. National Academies Press (US), Washington, D.C.

[Kaufman et al., 2009] Kaufman, D. J., Murphy-Bollinger, J., Scott, J., and Hudson, K. L. (2009). Public Opinion about the Importance of Privacy in Biobank Research. *The American Journal of Human Genetics*, 85(5):643–654.

[Keogh et al., 2009] Keogh, L. A., Vliet, C. M., Studdert, D. M., Maskiell, J. A., Macrae, F. A., St John, D. J., Gaff, C. L., Young, M. A., Southey, M. C., Giles, G. G., Rosenthal, D. A., Hopper, J. L., and Jenkins, M. A. (2009). Is uptake of genetic testing for colorectal cancer influenced by knowledge of insurance implications? *Medical Journal of Australia*, 191(5):255–258.

[Kevles, 2004] Kevles, D. J. (2004). *In the name of eugenics: genetics and the uses of human heredity*. Harvard Univ. Press, Cambridge, Mass., 5. print edition.

[Ku and Roukos, 2013] Ku, C.-S. and Roukos, D. H. (2013). From next-generation sequencing to nanopore sequencing technology: paving the way to personalized genomic medicine. *Expert Review of Medical Devices*, 10(1):1–6.

[Lee, 1993] Lee, C. (1993). Creating a genetic underclass: the potential for genetic discrimination by the health insurance industry. *Pace Law Review*, 13(1):189–228.

[Lee, 2013] Lee, S. S. (2013). Race, Risk, and Recreation in Personal Genomics: The Limits of Play. *Medical Anthropology Quarterly*, 27(4):550–569.

[Leighton, 2012] Leighton, K. (2012). Addressing The Harms of Not Knowing One's Heredity: Lessons From Genealogical Bewilderment. *Adoption & Culture*, 3(1):63–107.

[Lippman, 1991] Lippman, A. (1991). Research studies in applied human genetics: A quantitative analysis and critical review of recent literature. *American Journal of Medical Genetics*, 41(1):105–111.

[Little et al., 2022] Little, I. D., Koehly, L. M., and Gunter, C. (2022). Understanding changes in genetic literacy over time and in genetic research participants. *The American Journal of Human Genetics*, 109(12):2141–2151.

[López-Rubio and Ratti, 2021] López-Rubio, E. and Ratti, E. (2021). Data science and molecular biology: prediction and mechanistic explanation. *Synthese*, 198(4):3131–3156.

[Macer, 1992] Macer, D. R. J. (1992). Public Acceptance of Human Gene Therapy and Perceptions of Human Genetic Manipulation. *Human Gene Therapy*, 3(5):511–518.

[Marteau, 2000] Marteau, T., editor (2000). *The troubled helix: social and psychological implications of the new human genetics*. Cambridge Univ. Press, Cambridge, 1. paperback ed., reprint edition.

[Marteau and Croyle, 1998] Marteau, T. M. and Croyle, R. T. (1998). The new genetics. Psychological responses to genetic testing. *BMJ (Clinical research ed.)*, 316(7132):693–696.

[McEwen et al., 2013] McEwen, J. E., Boyer, J. T., and Sun, K. Y. (2013). Evolving approaches to the ethical management of genomic data. *Trends in Genetics*, 29(6):375–382.

[Miller and Pifer, 1993] Miller, J. and Pifer, L. (1993). The Public Understanding of Biomedical Science in the United States, 1993: A report to the National Institutes of Health. *Chicago: Chicago Academy of Sciences*, 1995.

[Morrison, 2005] Morrison, P. J. (2005). Insurance, unfair discrimination, and genetic testing. *The Lancet*, 366(9489):877–880.

[Muflih et al., 2019] Muflih, S., Bleidt, B. A., Lafferty, L., Shawaqfeh, M. S., and Alvarez, G. (2019). Measuring knowledge and attitudes towards the utilization of pharmacogenetic testing among physicians. *Journal of Pharmaceutical Health Services Research*, 10(2):227–234.

[Nagel, 1994] Nagel, J. (1994). Constructing Ethnicity: Creating and Recreating Ethnic Identity and Culture. *Social Problems*, 41(1):152–176.

[Naveed et al., 2015] Naveed, M., Ayday, E., Clayton, E. W., Fellay, J., Gunter, C. A., Hubaux, J.-P., Malin, B. A., and Wang, X. (2015). Privacy in the Genomic Era. *ACM Computing Surveys*, 48(1):1–44.

[Nicholls, 2016] Nicholls, S. G. (2016). Attitudes to incorporating genomic risk assessments into population screening programs: the importance of purpose, context and deliberation. *BMC Medical Genomics*, 9(1):25.

[of Management and Budget, 1977] of Management, T. O. and Budget (1977). OMB Directive 15.

[Oppenheimer, 2001] Oppenheimer, G. (2001). Paradigm lost: race, ethnicity, and the search for a new population taxonomy. *American Journal of Public Health*, 91(7):1049–1055.

[Parens, 2007] Parens, E., editor (2007). *Enhancing human traits: ethical and social implications*. Hastings Center studies in ethics. Georgetown Univ. Press, Washington, DC.

[Pearce et al., 2024] Pearce, A., Mitchell, L. A., Best, S., Young, M.-A., and Terrill, B. (2024). Publics' knowledge of, attitude to and motivation towards health-related genomics: a scoping review. *European Journal of Human Genetics*.

[Powe, 2020] Powe, N. R. (2020). Black Kidney Function Matters: Use or Misuse of Race? *JAMA*, 324(8):737.

[Raine, 2008] Raine, A. (2008). From Genes to Brain to Antisocial Behavior. *Current Directions in Psychological Science*, 17(5):323–328.

[Reardon et al., 2016] Reardon, J., Ankeny, R. A., Bangham, J., W Darling, K., Hilgartner, S., Jones, K. M., Shapiro, B., Stevens, H., and The Genomic Open workshop group (2016). Bermuda 2.0: reflections from Santa Cruz. *GigaScience*, 5(1).

[Riggs et al., 2019] Riggs, E. R., Azzariti, D. R., Niehaus, A., Goehringer, S. R., Ramos, E. M., Rodriguez, L. L., Knoppers, B., Rehm, H. L., and Martin, C. L. (2019). Development of a consent resource for genomic data sharing in the clinical setting. *Genetics in Medicine*, 21(1):81–88.

[Royal et al., 2010] Royal, C. D., Novembre, J., Fullerton, S. M., Goldstein, D. B., Long, J. C., Bamshad, M. J., and Clark, A. G. (2010). Inferring Genetic Ancestry: Opportunities, Challenges, and Implications. *The American Journal of Human Genetics*, 86(5):661–673.

[Sandel, 2009] Sandel, M. J. (2009). *The case against perfection: ethics in the age of genetic engineering*. Belknap Press of Harvard University Press, Cambridge, Mass., paperback ed edition.

[Sanger et al., 1977] Sanger, F., Nicklen, S., and Coulson, A. R. (1977). DNA sequencing with chain-terminating inhibitors. *Proceedings of the National Academy of Sciences of the United States of America*, 74(12):5463–5467.

[Scully and Rehmann-Sutter, 2001] Scully, J. L. and Rehmann-Sutter, C. (2001). When Norms Normalize: The Case of Genetic "Enhancement". *Human Gene Therapy*, 12(1):87–95.

[Shim et al., 2018] Shim, J. K., Rab Alam, S., and Aouizerat, B. E. (2018). Knowing something versus feeling different: the effects and non-effects of genetic ancestry on racial identity. *New Genetics and Society*, 37(1):44–66.

[Shostak et al., 2009] Shostak, S., Freese, J., Link, B. G., and Phelan, J. C. (2009). The Politics of the Gene: Social Status and Beliefs about Genetics for Individual Outcomes. *Social Psychology Quarterly*, 72(1):77–93.

[Smedley, 2003] Smedley, B. D. (2003). *Unequal Treatment: Confronting Racial and Ethnic Disparities in Health Care*. National Academies Press (US), Washington (DC).

[Stajano et al., 2008] Stajano, F., Bianchi, L., Liò, P., and Korff, D. (2008). Forensic genomics: kin privacy, driftnets and other open questions. In *Proceedings of the 7th ACM Workshop on Privacy in the Electronic Society*, WPES '08, page 15–22, New York, NY, USA. Association for Computing Machinery.

[Supreme Court of Georgia, 1995] Supreme Court of Georgia (1995). Mobley v. State.

[Theunissen, 2022] Theunissen, C. A. (2022). The Effects of DNA Test Results on Biological and Family Identities. *Genealogy*, 6(1):17.

[Thomas, 2012] Thomas, R. G. (2012). Genetics and insurance in the United Kingdom 1995–2010: the rise and fall of "scientific" discrimination. *New Genetics and Society*, 31(2):203–222.

[Tishkoff and Kidd, 2004] Tishkoff, S. A. and Kidd, K. K. (2004). Implications of biogeography of human populations for 'race' and medicine. *Nature Genetics*, 36(S11):S21–S27.

[Ury, 2013] Ury, A. G. (2013). Storing and interpreting genomic information in widely deployed electronic health record systems. *Genetics in Medicine*, 15(10):779–785.

[Van Hoyweghen et al., 2005] Van Hoyweghen, I., Horstman, K., and Schepers, R. (2005). 'Genetics is not the issue': Insurers on genetics and life insurance. *New Genetics and Society*, 24(1):79–98.

[Van Ommen, 2002] Van Ommen, G. B. (2002). The Human Genome Project and the future of diagnostics, treatment and prevention. *Journal of Inherited Metabolic Disease*, 25(3):183–188.

[Venter, 2001] Venter, J. C. e. a. (2001). The Sequence of the Human Genome. *Science*, 291(5507):1304–1351.

[Vicedo, 1992] Vicedo, M. (1992). The human genome project: Towards an analysis of the empirical, ethical, and conceptual issues involved. *Biology & Philosophy*, 7(3):255–278.

[Wynne, 1992] Wynne, B. (1992). Public understanding of science research: new horizons or hall of mirrors? *Public Understanding of Science*, 1(1):37–43.