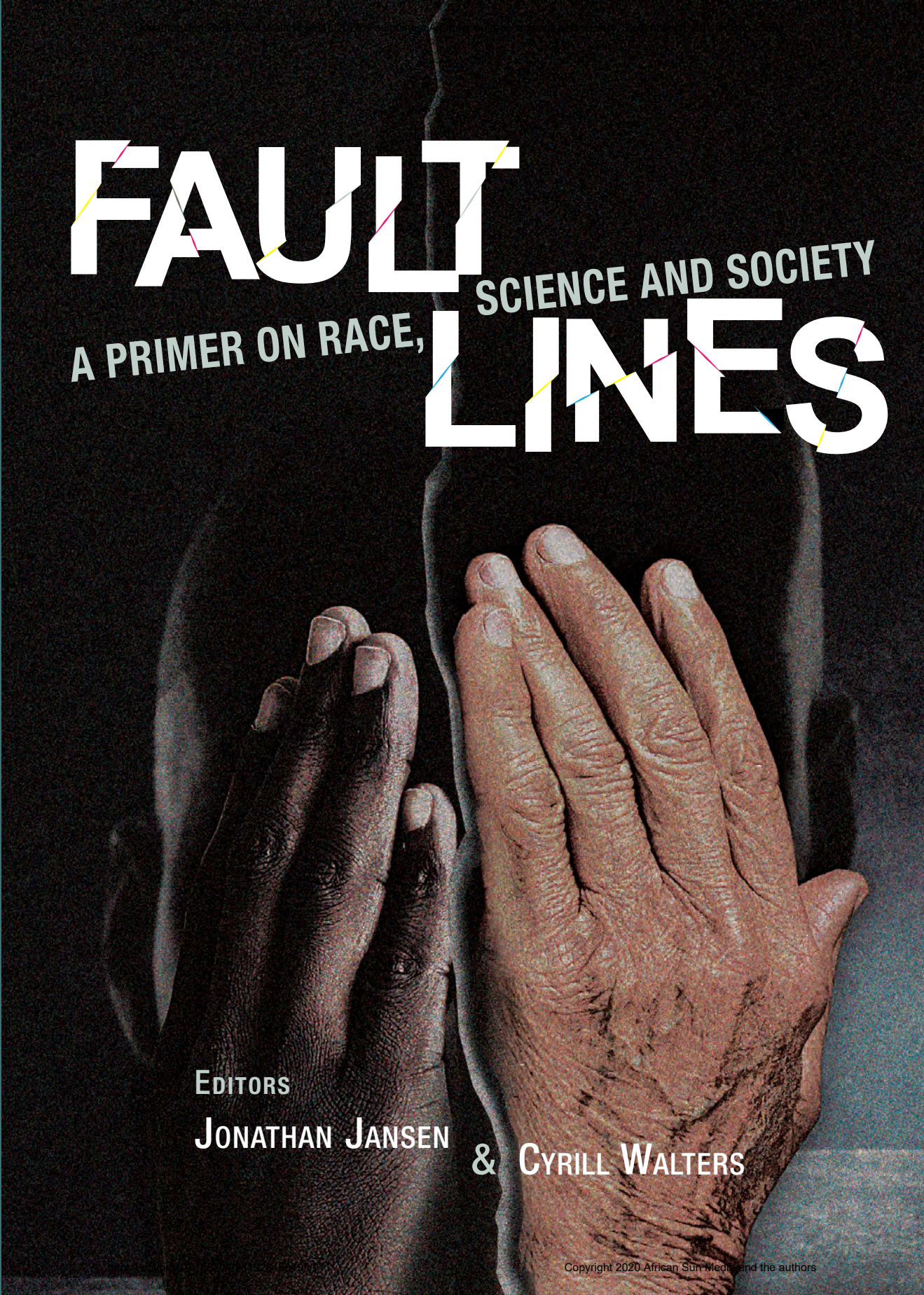
FAULT LINES

A PRIMER ON RACE, SCIENCE AND SOCIETY



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01

The Role of Genetics in Racial Categorisation of Humans

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Introduction

Only very recently in the history of modern humans have we learned how to read the stories hidden in our DNA. The ability to read and interpret DNA has revealed that many things are not as they are perceived to be. For instance, physical features between two people may be strikingly different and therefore be taken to mean that the individuals are fundamentally different, when in fact the DNA of any two humans is almost identical (99.9% the same) on a genetic level.

Given the physical differences apparent between populations, much research has gone into studying what makes them different. This type of research, no matter how well intentioned, has led to the pseudoscientific arguments used to justify movements such as the slave trade, the eugenics movement and apartheid in South Africa. Scientists at Stellenbosch University have also played a significant role in highlighting the 'racial' differences in the South African population. One such study is the now-retracted Sport Science article.[1] In this study, the authors, albeit unwittingly, reinforce racial stereotyping by concluding that so-called 'coloured' women in South Africa have lower cognitive functioning when compared to American age-standardised norms, and that this is due to exposure to a variety of factors with known negative effects on cognitive function. In an attempt to shed some light on the inaccuracies of the assumptions on which this article is based, this chapter will provide some background to racial categorisation from a genetic perspective. It will start with basic concepts in genetics and then expand into some of the more complex concepts and theories supporting the fact that there is no genetic basis for race in humans.

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The basics of DNA

DNA stands for **d**eoxyribo**n**ucleic **a**cid. Everyone, with the exception of identical twins, has a unique set of DNA. This DNA is an instruction manual that contains the information our cells need to make proteins and other molecules essential for our development, growth and survival. All human cells, except red blood cells, contain DNA, which is stored in a part of the cell known as the nucleus.

DNA occurs in the form of a double helix, which resembles a twisted ladder-like structure. The rungs of this 'ladder' are made up of four nucleotide bases: adenine (A), cytosine (C), guanine (G) and thymine (T). Combinations of these bases form three-letter 'words' called **codons**, which the cell reads to make proteins.

Each codon specifies which protein building block, known as an **amino acid**, should be added next during the process of making proteins. Various combinations of amino acids make up different proteins. The three-letter codons are pieced together in an estimated 20 000 to 25 000 'sentences' called **genes**.[2] The genes are separated by nucleotide bases which do not code for amino acids but are still important. These stretches of jumbled-up bases are called non-coding DNA (also referred to as junk DNA) and they make up the vast majority (98.8%) of our DNA. Some of the non-coding DNA functions as 'punctuation marks', providing information as to where one gene ends and the next one starts. Other non-coding DNA regulates when and how much of the proteins are made, or control how DNA is packaged within the cell. However, there is still a lot that is not yet known about non-coding DNA and its functions.

The DNA double helix is tightly coiled around proteins to form X-like structures called **chromosomes**. Humans have a total of 23 chromosome pairs. Twenty-two of these are called autosomes and one is a pair of sex chromosomes that determines whether one is female or male. If one inherits two X sex chromosomes, one is female, whereas if one has inherited an X and a Y sex chromosome, one is male. Humans inherit one chromosome of every chromosome pair from each of their parents.

It is important to note that *all humans have the same set of genes*, but they can have *slightly different* versions of these genes. These different versions are due to variations in DNA, known as **alleles**. Two alleles of a gene could have different properties, for example, one coding for blue eyes and another coding for brown eyes. The fact that humans have the same genes, but different alleles, is what makes them incredibly similar, yet amazingly unique.

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Genetic variation in humans: How did this come about?

The differences in alleles between individuals is known as **genetic variation**, and there are several ways in which this can come about, including mutation and sexual reproduction. Importantly, genetic variation can exist not only in genes but also in non-coding DNA. Sexual reproduction is an important source of genetic variation in humans. Siblings (except identical twins) from the same parents are not identical genetically or physically. This is because sexual reproduction involves genetic shuffling and random fertilisation, which contribute to genetic variation and the resulting differences in appearance.

Genetic shuffling (i.e. crossing over of individual chromosomes from chromosome pairs) occurs during the formation of sex cells (i.e. a woman's egg cells and a man's sperm cells). When these sex cells are formed, the maternal and paternal chromosomes of an individual exchange pieces of DNA to form new combinations of alleles (Figure 1.1). Subsequently, the individual chromosomes from each newly shuffled chromosome pair are randomly separated into different cells, so that every sex cell contains only 23 individual *chromosomes* instead of 23 *chromosome pairs*. This process ensures that every sex cell formed has a unique set of chromosomes. Upon fertilisation of an egg cell with a sperm cell, the individual chromosomes from the two sex cells form a new combination of 23 chromosome pairs with a unique combination of alleles.

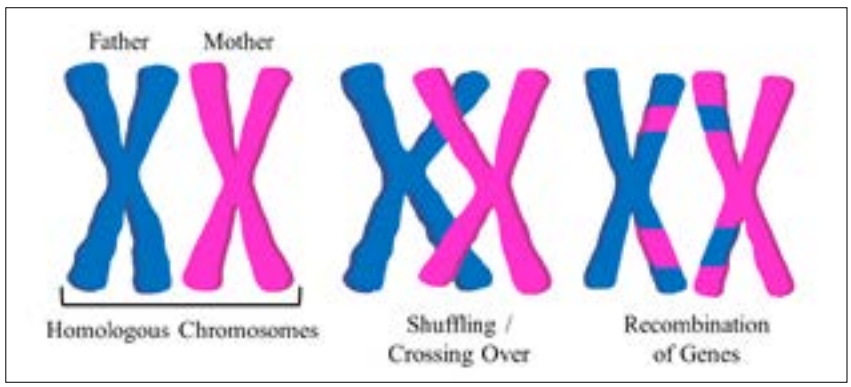

FIGURE 1.1: Genetic shuffling to produce new combinations of alleles in offspring [*Illustration by Caitlin McCaffrey*]

Another way in which new alleles can come about is through mutation – a random change in the nucleotide sequence of DNA. This can be caused by environmental factors, such as chemicals and radiation, but also by errors made by the cell when copying DNA into new cells. In humans and other multicellular organisms, only mutations in cell lines that give rise to sex cells will be passed on to the offspring.

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Genetic variation in populations

By biological definition, populations consist of members of the same species that interbreed. High genetic variation is evident in a population when there are many different alleles and many different combinations of these alleles. Collectively, the different alleles within a population are known as the population's **gene pool**.

This gene pool can change over time. Different allelic forms of a single gene can appear and disappear. Some alleles may also become 'fixed' in a population, which means that a population only has one version of that allele, and all others have been removed or lost. Changes in a population's gene pool and allele fixation can be due to environmental factors that favour certain traits over others, the death of a large number of individuals within the population or the migration of individuals into or out of the population.

Out-of-Africa hypothesis

Ancestors of humans originated in Africa and subsequently populated the rest of the world.[3] Groups moved away from the population nested in Africa and expanded in all directions of the globe about 60 000 years ago (Figure 1.2).[4] The smaller groups took with them only a subset of the alleles and genetic variation found in the ancestral African gene pool. This means that the *greatest genetic diversity is found in African populations*. The smaller populations that moved away from Africa settled, grew in size and gave rise to new populations, which 'budded off' and repeated this process.[5] However, it is now known that these populations moved in all directions all the time, with genetic exchange (gene flow) occurring between populations, thus blurring the genetic lines between them.[6] For instance, Eurasian populations that had left Africa later (~3 000 years ago) again exchanged genetic variation with ancestral African populations.[7] Moreover, analysis of several genomes has indicated ancient **admixture** (more than one genetic ancestry) amongst populations that expanded from Africa.[8] This means that some populations that left Africa also exchanged genetic variation (mated) with now-extinct groups of hominids such as Neanderthals and Denisovans.

To study genetic variation, gene flow and migration patterns amongst humans, geneticists often investigate **single nucleotide polymorphisms (SNPs)** in DNA. These are individual nucleotide bases within the genome that vary widely between people and are in fact the most common type of genetic differences between individuals, accounting for ~95% of all known sequence variation.[9] SNPs can be located within genes or in non-coding DNA. Given that most human DNA is non-coding, most SNPs (and therefore most genetic variation) are also located in the non-coding DNA. Some SNPs have been conserved over thousands of generations in human populations and therefore enable one to infer the genetic ancestry of an individual.[10]

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FIGURE 1.2: A depiction of the Out-of-Africa hypothesis indicating that modern humans originated in Africa and then migrated to other regions of the world. The greatest genetic diversity is seen in Africa when compared to the rest of the world. [*Illustration by Caitlin McCaffrey*]

Many are shared by populations across the globe,[11] but their frequencies can differ widely between geographic populations[12] due to the different ancestral gene pools of populations and the different factors that may have shaped these gene pools over time.

Link between genetic variation and human disease

Why do some genetic diseases and traits appear to be more common in some populations than in others? In addition to genetic variation, which can help infer the ancestry of individuals, disease-causing alleles and SNPs can also be found at different frequencies amongst different populations around the world. Again, this is due to the different ancestral gene pools from which the populations originated and the different ways these genes pools have changed over time.

Selective advantage

One way in which an allele can become more frequent in a gene pool is if it codes for a trait that is favourable for a given environment. As such, some alleles have been passed on through generations because they confer some form of selective advantage to the individuals in the population. Sometimes an allele that causes a genetic disease is actually passed on to subsequent generations because it helps individuals survive

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in certain climates or protects them against a deadly disease. For instance, an allele in the **HbS** gene, which is protective against malaria infection, but causes sickle cell disease, has been passed on through generations in populations living in areas with a high incidence of malaria (e.g. West Africa).[13] In this case, the sickle cell allele was advantageous because it protected individuals from malaria infection (a common cause of death), and therefore it became more common in the population's gene pool over time.

Another example is an allele in the **HFE** gene that causes hemochromatosis (an iron overload disorder), which is found at high frequencies in individuals of Northern European ancestry.[14] Given that iron is an essential micronutrient needed to effectively regulate body temperature, the allele is thought to have helped individuals survive the cold and wet climate in European countries.[15] It was therefore selected for in individuals living in Northern Europe.

In addition to diseases, some differences in physical traits between populations, such as skin pigmentation, are also due to selective advantage. In humans, skin pigmentation is an adaptation to differing levels of ultraviolet (UV) radiation. Regions close to the equator receive more UV radiation than temperate regions. Therefore, alleles for darker pigmentation became more common in populations living in these equatorial regions, as they protected individuals from harmful diseases such as skin cancer. In contrast, alleles for lighter skin pigmentation were favoured in the temperate regions, where the UV radiation was less severe. It should be noted, however, that skin pigmentation is a complex trait influenced by alleles at a number of different genes, and it has been under continuous evolution throughout hominid history.[16]

Founder effects

Sometimes populations have an increased frequency of a disease-causing allele not because it was particularly useful, but simply because the allele was present in one or a few of the individuals who originally founded the population (i.e. the individuals who moved away from their ancestral population and gave rise to the new population). As the new population increased in size, the allele increased in frequency over generations and became more common in the gene pool. This is known as the **founder effect** and it is thought to account for an increased frequency of genetic disorders in some populations. The increased frequency of Bardet-Biedl syndrome (BBS) in the island population of Newfoundland[17] is a fairly recent example of how the founder effect has increased the frequency of a disease-causing allele in a population. BBS affects multiple body systems, and features of the disease include obesity and intellectual impairment. The founding population of Newfoundland was small (~20 000 settlers in 1760) and consisted of individuals from England and Ireland, some of whom carried alleles for BBS.[18]

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Another example of a population with a founder effect can be found in South Africa. The Afrikaner population is based on the Dutch, German and French immigrants who settled in the Cape in the 1600s and founded a new population here. It has been estimated that in the period between 1637 and 1806, the total number of progenitors for this population was approximately 4000 individuals.[19] Founder effects for a number of diseases have been observed in this population, possibly due to the fact that the disease alleles were present in the original progenitors and were later amplified through exponential population expansion.[20]

However, it is important to note that the fact that a disease is more common in some populations than in others does not mean it is exclusive to a particular population. The same genetic diseases can be found across all populations in the world. The fact that modern-day humans move across the globe with ease and intermarry also means that frequencies of these disease-causing alleles and other genetic variations are likely to change again over time. Moreover, it should be stressed that differences in allele frequencies between populations do not solely account for health disparities between populations. Health disparities apparent between different geographic regions are also due to differences in access to healthcare, diets, lifestyles and socioeconomic factors, all of which influence disease prevalence and incidence.

Concept of race: On what is the label based?

Different populations around the world can have different frequencies of alleles based on which ancestral gene pool they originated from. However, this does not mean that humans from populations around the world are different enough from one another to justify being separated into distinct groups. In this section, it will be explained why this is the case.

Taxonomic classification and race

Taxonomy is the branch of science involving classification and naming of organisms in an ordered system to indicate the relationship between them. In taxonomy, species were traditionally and primarily distinguished based on their physical appearances. However, appearance cannot tell how genetically different or similar two organisms are. Many organisms that look almost identical are actually quite different to one another at the genetic level and vice versa. For instance, some organisms previously classified as a single species based on appearance (e.g. populations of the popular lab worm, *Lumbriculus variegatus*) have now been found to be genetically different enough to be classified as separate species.[21] In contrast, the African elephant and the dassie, which look nothing alike, are in fact evolutionarily related to one another, albeit distantly.[22] This highlights an important notion, i.e. what is visible to the naked eye does not tell one much about genetics.

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This also holds true for humans, who have historically been grouped into races based primarily on their geographic location and physical appearance, particularly their skin colour. Analysis of the complete DNA sequences of two American so-called 'white' geneticists of European origin (James Watson and Craig Venter) revealed that they were more dissimilar to each other than they were to a scientist of Asian descent, Seong-Jin Kim.[23] Although physical features may appear to be strikingly different across the world, they are only determined by a tiny percentage of our DNA. The *observable* traits that differ between populations are therefore superficial and few. Importantly, most human genetic variation (~90–95%) is due to variation amongst individuals within a population, whereas only about 5–10% is attributable to variation between populations.[24] This 5–10% of genetic variation falls well below the 25% threshold that taxonomists use to divide organisms into subspecies. In other words, this means that human populations are not genetically distinct enough to be divided into subspecies or, in this case, races. Consequently, there is *no genetic basis for race in humans*.

Race as a variable in genetic studies

If race has no genetic basis, why does one read about it in genetics studies? Although race and ethnicity are related concepts, and are often used interchangeably, they are quite different. Race is based mainly on observable differences in physical appearance (e.g. skin colour and eye colour). In contrast, ethnicity is a complex concept that reflects biological factors and refers to communality in cultural heritage, language, social practice, religion and many other factors. Often geneticists use broad racial terminology to define their study group when in fact they could define it more clearly in other ways, including the group's ethnicity. The language that is used in scientific research matters, and assigning racial or even ethnic labels to study participants is often unnecessary, depending on the research context and question. This is important to note because scientific reports (mis)using racial terminology can easily be misinterpreted by the public, and they have been used to justify racism and eugenics, as will be discussed later.

If geneticists should not use racial or even ethnic groups in their studies, why should they try to group together individuals at all? In some instances, grouping individuals together based on common *genetic ancestry* is important in order to answer key research questions. It was stated previously that populations across the world have different frequencies of genetic variation. In studies on human population genetics, individuals who share ethnolinguistic backgrounds are grouped together to study human history and migration patterns. Also, geneticists study genetic variation that could be associated with, or may cause, a disease. Very often, a group of individuals

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who have a similar genetic ancestry are selected for genetics studies so that SNPs (or other genetic variants) that are found in people affected by a disease and are absent in people without the disease can be identified. If individuals from different genetic ancestral backgrounds are studied together, it becomes difficult to pinpoint SNPs that actually cause disease, since there will be many more SNPs that differ between the individuals, given their genetic ancestry (and not their disease status). Moreover, if the disease group consists mostly of individuals from ancestry X, but the healthy control group consists mostly of individuals of ancestry Y, then a SNP found in individuals of ancestry X may be incorrectly associated with the disease simply because the SNP is overrepresented in the disease group.

To illustrate this point, two famous geneticists, Lander and Schork, provided a humorous example of a hypothetical study in San Francisco to investigate alleles in the immune system gene complex (HLA) and the ability to eat with chopsticks. An association between a particular HLA allele (HLA-A1) would be found, not because there is a biological link between the allele and eating with chopsticks, but because this allele is more common in individuals of Asian ancestry than those of European ancestry.[25] For this reason, it is necessary for geneticists to group individuals together for research based on their *genetic* ancestry to avoid making such false connections between a SNP and a disease or a trait of interest. It is important to note, however, that the knowledge gained from studies in one population in which disease-causing variants are identified can then be applied to help identify susceptible individuals from other populations. This is due to the fact that the same alleles are usually found in all populations, **but they just occur at different frequencies**.

However, recruiting individuals with the same genetic ancestry for such studies is difficult, given that genetic ancestry is not an observable trait; nor is it quick and inexpensive to test for. Thus, past studies have resorted to recruiting individuals based on historically defined racial categories, despite their being poor proxies for genetic ancestry. Very often, the racial category assigned to an individual based on their physical appearance disagrees with their genetic ancestry. For instance, a person with a so-called 'black' and a 'white' parent in the United States of America (USA) is socially classified as a 'black' or 'African American'. This is due to the 'one-drop rule', a historical social and legal classification in the USA that held that an individual with even one ancestor of sub-Saharan African ancestry would be considered to be 'black'.[26] Interestingly, the proportions of African and European ancestry in self-identified 'African Americans' has been shown to vary widely,[27] highlighting that race is a poor indicator of genetic ancestry.

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The 'African American' group is not the only one in the USA varying in terms of their admixture (i.e. having more than one genetic ancestry). Another example is the 'Hispanic' or 'Latino' group. Mexican Americans have a higher proportion of Native American ancestry (between 35% and 64%), and a lower proportion of African ancestry (between 3 and 5%), than Puerto Ricans, whose African ancestry is higher (between 18 and 25%) than their Native American ancestry (between 12 and 15%).[28] Therefore, for these individuals, the use of a single 'Hispanic' or 'Latino' category is a poor description of their genetic ancestry.

Taken together, these observations reveal that characterising races simply as 'white', 'black', 'Asian' or 'Latino/Hispanic' is an inaccurate way to predict human genetic diversity or similarity. Although racial categories may be helpful in studying sociocultural and socioeconomic factors, such as income and housing, given the history of racial discrimination around the world, they are not always useful in revealing the genetic ancestry or the extent of genetic admixture in an individual. Importantly, as multiethnic marriages and intermarriage between different global population groups become more common, it is increasingly difficult (and will become more so) to assign a single ethnicity to an individual. Here it is important to highlight the fact that humans are *one continuously variable, interbreeding species*. This does not mean humans are all the same, or that there are no observable differences within our species. It just means that trying to separate the human species into distinct groups, based on physical differences, has little genetic meaning.

Despite this knowledge, the genetics community has long debated the use of racial and ethnic terminology in research, but it has failed to reach a global consensus on this question. Perhaps, many researchers fail to see the consequences of using such terminology in modern-day society. Recently, the use of such terminology, combined with the misinterpretation of study findings, has led to much debate in the media about *possible genetic superiority* of certain racial groups. This prompted the American Society of Human Genetics (ASHG) to put out a statement in November 2018 denouncing attempts to link genetics and racial supremacy.[29] The statement declares that the Society is "alarmed to see a societal resurgence of groups rejecting the value of genetic diversity and using discredited or distorted genetic concepts to bolster bogus claims of 'white supremacy'. It goes on to say, "Any attempt to use genetics to rank populations demonstrates a fundamental misunderstanding of genetics." Such misconstrued ideas about genetics being able to justify supremacy of any kind threaten the re-emergence of eugenics.

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Eugenics

Eugenics can be defined as a set of views and practices that aim to 'improve' the genetic make-up of the human population or to increase the occurrence of desirable characteristics. The term was first used by Francis Galton in 1883 and was thought to be based on the work of Charles Darwin.

Eugenics principles can be divided into two categories: *Positive eugenics* is aimed at encouraging reproduction of groups thought to be superior, for example, individuals who are thought to be intelligent, healthy, and successful. *Negative eugenics* is about eradicating, through forced sterilisation, abortions, segregation or marriage prohibitions of individuals with 'undesirable' traits such as physical or mental disorders, criminality, homosexuality and members of certain population groups. The movie *Gattaca* provides a chilling account of a future world in which eugenics is used to decide what people are capable of and their place in society. In the movie, children are conceived through genetic selection to ensure that they have the best genetic characteristics of their parents and are considered superior to individuals who are conceived outside of the eugenics programme.

In modern times, eugenics is seen as being linked to 'white' supremacism. The contemporary history of eugenics began in the late 19th and the beginning of the 20th century, when the eugenics movement was started in the United Kingdom, and later spread to other countries throughout Europe as well as the USA. These countries adopted eugenics policies with a goal to improve the quality of their populations' genetics. Later, during World War II, the eugenics movement became associated with the genocidal programmes of Nazi Germany and the Holocaust. During the Nuremberg trials, the defendants tried to justify their human rights abuses by claiming there was little difference between the Nazi and American eugenics programmes.[30] After World War II, with the institution of new human rights laws and regulations, many countries started to reject eugenics policies, although some, such as the USA, continued with involuntary or forced sterilisations.[31] A major criticism of eugenics policies is that they will permanently and artificially disrupt millions of years of evolution, and that attempting to create genetic lines devoid of 'defects' can have far-reaching resulting negative effects on immunity and species resilience.

Since the 1980s, with the development of new assisted reproductive technology, such as *in vitro* fertilisation, preimplantation genetic testing, surrogacy and mitochondrial replacement therapy, fears of eugenics are re-emerging. Today, we have technologies, such as DNA editing, that make it possible to alter the genetic composition of an individual. Scientists are already using DNA editing to treat individuals with blood disorders and cancers in ongoing clinical trials and they plan to treat a range of

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additional disorders, including inherited blindness, in the near future.[32] Also, DNA editing of human embryos that inactivated a gene involved in HIV infection has recently resulted in the birth of potentially HIV-resistant humans in China.[33] These experiments were widely criticised as being ethically questionable and technically flawed, but it is anticipated that such applications will continue to become more widespread. The fear is that these approaches will not stop at disease prevention, but will also be used to intentionally 'improve' individuals, i.e. genetic enhancement of physical and intellectual traits. This will bring with it its own set of ethical, moral and legal dilemmas.

Eugenics in South Africa

As the scholar Linda Naicker points out, "the scientific theory of eugenics laid the foundation for South Africa's race policies and continued to be a key driver of racial segregation throughout the formative years of apartheid and should, therefore, be a concomitant consideration when analysing issues of racial formation in South Africa".[34] The concepts of 'inferior types' and European superiority were widely propagated by South African eugenicists. They and others were concerned that the mixing of racial groups was a social crime that would cause great damage to 'white civilisation'.

Historian Susanne Klausen has examined the eugenic beliefs of members of the English-speaking medical profession in South Africa during the first three decades of the last century. She found that South African eugenicists, concerned about the future and health of the 'white' race, believed it was their duty to interfere in people's social relations. They thought that there was a link between the health of the 'white' population, the role of 'white' women as 'mothers of the nation' and the health of the South African state. The growth of urban slums due to the migration of landless 'Afrikaners' and 'black Africans' to urban areas to find employment was a major concern in the country.[35]

The medical profession in South Africa was particularly concerned about the escalation of 'feeblemindedness' amongst 'white' people due to racial mixing in these slums. 'Feeblemindedness' was a flexible category that could include individuals with putative mental, and often physical, moral and other deficiencies, depending on the context. The medical profession believed that, if not stopped, this 'degeneration' would result in social, cultural and economic devastation of the 'white' population in this country. In her article, Naicker states:

> Even though racist practices were commonplace in the first three decades of colonialism in South Africa and loomed large throughout its history, the theoretical orientation of the concept was a British construct which was expanded by the South African colonial medical profession early in the 20th century.[36]

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Given what is now known about the negative genetic consequences and human suffering caused by the practice of eugenics, it is important not to take modern eugenic ideas (such as genome editing in the name of disease prevention) a step too far. It should also be noted that in the past, eugenics mainly involved the sterilisation of, and the enforcement of reproduction laws against, adults. Now, in the post-genomic era, it is embryos that are being artificially genetically manipulated. A major ethical concern is that the unborn foetus, upon whom these eugenic procedures are being practised, has no voice to express their opinion about what is being done to them. With the possibility of DNA editing becoming more frequent in the future, in the quest to create 'perfect' disease-resistant humans, it should be remembered that genetic diversity and genetic 'flaws' have enabled the human population to survive and thrive over thousands of years.

Concluding remarks

The purpose of this chapter is to provide an understanding of some key concepts in human genetics that explain why race has no genetic basis. It is known that DNA codes for the same set of genes in all humans, but each person can have different alleles. The differences that occur in the frequency of alleles between populations are due to the different gene pools that the populations originated from, and the factors that shaped these gene pools over time. These genetic differences between populations are too small to justify grouping humans into *distinct* categories such as 'races'. In other words, *humans are more genetically similar than they are dissimilar*. Broad racial categorisation, e.g. 'black', is a poor indicator of genetic diversity and of genetic ancestry. The use of racial terminology in scientific writing can lead to the misunderstanding of genetic studies as providing support for or fuelling racism. Finally, the discriminatory eugenics practises of the past are threatening to make a comeback, and we should be prepared for this with thorough consideration and understanding of all of the concomitant ethical, moral and legal implications.

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