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# RELATIONSHIP TO PHYSICAL TRAITS AND THEIR GENETIC DIFFERENCES AMONG INDIVIDUALS

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## Abstract

Human diversity is based on genetic variation, which determines a vast number of physical characteristics such as the color of eyes, hair type, pigmentation of the skin, height and disease susceptibility. This review will be looking at the existing literature regarding the genetic differences among individuals and how they relate to observable traits. It emphasizes the contribution of mutations, gene recombination and chromosome variations in the production of specific genotypes and the role of environmental and epigenetic factors in the expression of the phenotype. There are specific genes, e.g. OCA2 and HERC2 that control the color of the eye, EDAR and TCHH that control the type of hair, and several polygenic loci that control the height and the pigmentation of the skin are discussed to illustrate the genetic origin of traits. The gaps in existing research also include the lack of knowledge on polygenic interactions, underrepresentation of diverse populations, and lack of literature on the effect of genes and the environment and epigenetics. Future research recommendations include the need to adopt interdisciplinary studies, longitudinal studies, and large-scale population studies to improve insights on human diversity. The importance of knowing the genetic variation is paramount to personalized medicine, anthropology, and population health.

## 1. Introduction

Genetic variation describes the differences in the sequences of DNA among the representatives of the same species, and it is the very basis of the great diversity of physical characteristics that people and other organisms have. Even though humans are almost 99.9% related to each other in terms of genetic composition, the 0.1% of genetic variation explain the differences in the characteristics of the human species, including eye color, hair texture, height, skin pigmentation, and even the dissimilarity in the vulnerability to some illnesses (National Human Genome Research Institute, as cited in Understanding Genetics, 2026). Such variations occur in a number of biological mechanisms such as mutation, genetic recombination during meiosis, chromosomal alterations, and others that produce new combinations of genetic material that may be transmitted to the offspring (National Geographic Society, 2026; Parker, 2023).

The mutations can be described as alterations in the DNA sequence of a nucleotide and are the ultimate source of new genetic material in a population. They may be spontaneous during the process of replicating the DNA, or caused by environmental factors, such as radiation exposure or the use of the chemical mutagens (Encyclopedia Britannica, 2026). Other mutations involve one base substitution, also called single nucleotide polymorphisms (SNP), and they happen about 100 to 300 times in the length of the human genome (Understanding Genetics, 2026). Although most of the mutations do not have any distinct impact on the organism, some of the mutations may modify the functionality of the gene and cause the observable alterations in physical characteristics or even impact the health condition.

Besides mutation, genetic recombination is also observed during sexual reproduction; maternal and paternal DNA are shuffled in gametes and new allele formations are created in the offspring. It is a process that enhances the genetic diversity in populations and leads to the individuality of the

genotype of each individual (Unraveling Genetic Variation, 2023). Similarly, the larger parts of DNA may also change due to the presence of structural variations, including insertions, deletions, and duplications, and play a major role in the genetic variations among individuals ( Structural variation, 2026).

There are phenotypes that are regulated by one gene (Mendelian inheritance) with the appearance of certain alleles defining the appearance of a certain phenotype. Polygenic traits are other characteristics that are determined by numerous genes each producing a minor effect in a continuous variation like human height or skin color. Genome wide association studies (GWAS) are large scale genomic studies that assist scientists in discovering the numerous genetic loci that are related to these complex traits (Genetic Contributions to Complex Traits, 2025). In addition, the relationships between genes, including the idea of epistasis, when the ability of one gene is determined by another or a combination of others, also influence phenotypes and make the interaction between genotype and physical appearance more complicated (Epistasis, 2026).

The knowledge of genetic variations and their impact on bodily features has significant consequences to various fields of science. In health, genetic variation can be used to assist scientists in determining the risk of disease, comprehend drug sensitivity, and create tailored treatment methods. Variation in evolutionary biology offers the raw material upon which natural selection can act to ensure that populations can adapt to changing environments across generations. In anthropology, genetic diversity is used to explain the existence of the various physical differences in human populations as a result of time.

## **Background**

Genetic variation is the difference in the DNA sequences that result in genetic variation in individuals. These differences are the reason behind the vast difference in physical traits that can be observed such as color of the eyes, hair type, height, skin colour, and even susceptibility to some diseases (National Human Genome Research Institute, 2026). Although genetics is the blueprint of those traits, the environment, including nutrition, climate, lifestyle, and exposure to outside agents, may interact with genes in determining the ultimate expression of these traits (Parker, 2023). As an example, height is not only predetermined by inherited genes but also by such factors as nutrition and general health in the childhood. The origin of genetic variation is found in mechanisms of mutation, recombination of chromosomes during meiosis, and structural alterations in DNA, which makes a person different (Encyclopedia Britannica, 2026). There are those traits that are simple to inherit through the Mendelian rules, and which are dictated by just one gene, and there are also polygenic traits that are multi-gene and require a combination of genes to work together as a result of which continuous variation occurs ( Genetic Contributions to Complex Traits, 2025). The study of these differences is central in medicine where it can be used to create an individualized treatment plan; in biology where it can be used to understand how heredity and evolution works; and in anthropology where it can be used to understand human diversity and adaptation to populations. Through genetic variation studies, one can associate particular genes or genetic markers with physical appearances, make prediction about the inheritance patterns and also the genetic basis of normal as well as diseased traits.

The aim of this study is to investigate how genetic differences among individuals relate to variations in physical traits. Specifically, it seeks to identify patterns of inheritance, the role of specific genes, and how these genetic variations contribute to observable characteristics in humans.

This research aims at improving knowledge of human genetic diversity and how the same affects physical appearance. The field of application of this knowledge is in genetics education, personalized medicine and awareness of how genes affect health and appearance. It also leads to the expansion of the general concepts of the evolution of man and the variation of populations.

## **2. Literature Review**

Genetic variation is one of the basic notions in explaining human diversity. It means variations in the sequences of genes between individuals of the same species that result in the large diversity of noticeable physical characteristics, also known as phenotypes. Such characteristics are the color of the eyes, hair, texture, and skin pigmentation, height, body structure, and predisposition to illnesses. The genetic material has the blueprint of these traits, and their ultimate expression may be affected by the environmental factors, including nutrition, climate, lifestyle, and exposure to external agents (Parker, 2023).

The study of genetic variation enables the scientists to investigate the process of inheritance, determine the probability of particular characteristics emerging in individuals, examine how genes intersect with each other, as well as with the environment, to form visible characteristics. In the last few decades, with the emergence of molecular biology, bioinformatics, and genomics, it has become possible to discover certain genes and genetic markers associated with physical appearance. The implications of these discoveries are important to the fields of medicine, anthropology, evolutionary biology and genetics education.

## **Genetic Variation in Humans**

There are various biological processes that result in genetic variation. Mutations are one of the largest sources of such changes in the DNA sequence. Mutations are either spontaneous, which is natural and happens during hereditary replication, or induced by environmental factors like radiation, chemicals or viruses (Encyclopedia Britannica, 2026). Mutations can be small scale such as the single nucleotide polymorphism (SNP) or large scale such as deletions, duplications, inversions, or translocations of complete segments of chromosomes. Such variations make up the distinct genetic composition of all human beings and they are also necessary in the evolution since they furnish the raw material on which natural selection may operate.

Genetic recombination is another source of variation and it takes place in the process of sexual reproduction. In meiosis, inheritance takes place when the mother and father exchange parts of the chromosomes in a process known as crossing over which produces new combinations of alleles in the offspring (National Human Genome Research Institute, 2026). This recombination enhances genetic diversity in a population and also they make individuals unique.

The other concept is polygenic traits. The height, weight, and skin color are some of the traits that are affected by various genes and each gene has a minor effect. These characteristics are continuously varied, that is, they do not have discrete categories, but they have a broad spectrum of potential phenotypes. Mendelian traits, on the other hand, which are regulated by one gene e.g. some eye color or blood type, exhibit discrete variation and are subject to predictable patterns of inheritance as dictated by the laws of Mendel.

It has been demonstrated through population genetics studies that populations differ in terms of allele frequencies. As an example, some of the genetic variants that cause some of the traits such as lactose intolerance, hair texture, and skin pigmentation vary between ethnic groups. Such variations are influenced by evolutionary forces like natural selection, genetic drift, migration and environmental adaptation. (Parker, 2023).

## **Physical Traits and Their Genetic Basis**

The eye color is a strongly investigated characteristic that is affected by several genes, in which OCA2 and HERC2 are the central factors. The genes control the synthesis and release of melanin in the iris. Mutations in these genes may result in brown, blue, green or intermediate eye color (Encyclopedia Britannica, 2026).

Some genes such as TCHH (trichohyalin) and EDAR (ectodysplasin A receptor) also control the hair type and texture. As an illustration, the EDAR variations are associated with straight and curly hair among different people. The color of hair is influenced by genes that control the production of melanin, including MC1R, that also helps determine the difference between red, brown, and black hair (National Human Genome Research Institute, 2026).

Height is a conventional polygenic trait that is affected by hundreds of genetic loci in the genome. Various genes having a minor effect have been found through genome-wide association studies (GWAS) to add height. These genes are interacted with the environment to define the ultimate adult height including nutrition, health, and early childhood development (Genetic Contributions to Complex Traits, 2025).

The pigmentation of the skin is highly diverse among human beings and it is regulated by numerous genes such as SLC24A5, SLC45A2 and TYR. These genes influence the quantity and nature of melanin that is produced by melanocytes on the skin. According to evolutionary research, the lightness of the skin developed in those communities that lived at a high latitude to enable the synthesis of vitamin D, and the darkness of the skin in equatorial areas offers protection against ultraviolet radiation (Parker, 2023).

The other field of genetic variability is in disease susceptibility. An example is the sickle cell anemia caused by alleles of the HBB gene, and cystic fibrosis by variations of CFTR. The knowledge of such variations can be used to predict the risks of health and develop tailored medicine strategies (National Human Genome Research Institute, 2026).

## **Studies on Genetic Variation and Trait Correlation**

The relationship between genetic differences and physical traits is a subject that has been examined by many researchers. Some of the major findings include:

**Genome-Wide Association Studies (GWAS):** GWAS have uncovered many genetic markers that are linked to complex phenotypes, including height, weight and pigmentation of the skin. These studies examine the genomes of people with different phenotypes with the aim of identifying statistically significant associations ( Genetic Contributions to Complex Traits, 2025). **Twin and Family Studies:** The study of monozygotic (identical) and dizygotic (fraternal) twins is helpful in separating the influence of genetics and the environment. As an example, the same twins often share the similarities in height and eye color regardless of upbringing, but the fraternal twins can have more variations that can be explained by genetic variation (Parker, 2023). **Population Studies:** The study of different human populations has shed light on the differences in the frequency of alleles in different populations. As an example, the genes of lactose intolerance are found in a significant proportion in a group of people whose ancestors lived in the dairy economy, and genes of pigmentation of the skin are distributed in geographic clusters in relation to exposure to UV rays (National Human Genome Research Institute, 2026). **Epistatic Interactions:** Genes may also interact in such a way that the expression of one gene affects the expression of another. This type of interaction makes the patterns of inheritance of polygenic factors complicated and clarifies why some phenotypes are not determined by simple Mendelian patterns of inheritance (Epistasis, 2026).

## **3. Discussion**

### **Summary of Literature Review**

The literature proves that the main factor of human physical appearance is genetic variation. Mendelian (single-gene) traits are rather easy to research, and polygenic traits are associated with intricate interactions among several genes and environmental factors. Population research reveals the contribution of evolution, adaptation and migration to the apparent variations within human groups. In general, the knowledge of the genetic variation would offer an insight on human diversity, inform medical and anthropological studies and future studies on the relationship between the genotype and the phenotype. This review shows the relevance of considering genetic, environmental and population-level viewpoints to explain human physical characteristics fully.

The literature used in the study has shown that genetic variation is the dominant factor that determines physical diversity of humans. Human characteristics like eye color, hair type and texture, skin pigmentation, height, body shape and disease susceptibility are all dependent on the variation in the DNA sequences, including single nucleotide polymorphisms (SNPs), structural variations and recombination (National Human Genome Research Institute, 2026).

The traits that are governed by a single gene are known as the Mendelian traits and these are discrete in their inheritance patterns and are more or less predictable. The typical examples are some of the eye colors, earlobe attachment and blood types. Polygenic traits on the other hand entail the cumulative impact of a number of genes, each with a minimal impact which leads to continuous variation. Examples of polygenic traits include height, pigmentation of the skin, hair texture, and so on, and they depend on both a genetic component and environmental factors, including nutrition, lifestyle, and climate (Parker, 2023).

Modern bioinformatics and genome-wide association studies (GWAS) have detected certain genes and genetic markers that are linked with physical traits. For instance: Eye color depends on the OCA2 and HERC2 genes as it is the genes that control the production of melanin in the iris (Encyclopedia Britannica, 2026).

Gene TCHH (trichohyalin) and EDAR (ectodysplasin A receptor) are known to influence hair texture and type, and result in straight or curly hair. Genes such as MC1R also determine the pigmentation of the hair and determine whether red, brown, or black hair would be produced (National Human Genome Research Institute, 2026).

Height is affected by hundreds of polygenic loci which have an incremental effect. These genes are influenced by environmental factors like diet, childhood health, general nutrition, among others to dictate the final adult height (Genetic Contributions to Complex Traits, 2025).

Skin pigmentation also has several genes such as SLC24A5, SLC45A2, and TYR. These genes influence the production of melanin in the skin where the evolutionary adaptations may follow the geographical and environmental conditions. As an example, the darker color of the skin was developed in the populations of the equator to ensure protection against the UV radiations, and light color in the northern latitudes helps in producing vitamin D (Parker, 2023).

Research on disease predisposition also demonstrates that genetic variation is a risk factors of such diseases as sickle cell anemia (HBB gene), cystic fibrosis (CFTR gene), and diabetes. These genetic variations are essential to understand to be able to do personalized medicine, early diagnosis, and treat better.

## Research Gaps

1. Although there have been great advances in explaining the relationship between genes and physical traits there are still some gaping holes:
2. Poorly understood polygenic interactions: Despite the many genetic markers of complex traits that have been discovered, polygenic interactions, such as epistasis (where the effect of one gene on another) have poorly been understood. This weakness limits our ability to make accurate predictions of the expression of polygenic traits (Epistasis, 2026).
3. Under researched environmental influences: Although genes are critical in the expression of traits, there are several environmental influences e.g. nutrition, environmental exposure to toxins, and lifestyle decisions which have significant impacts on most traits. Few studies have managed to incorporate these environmental factors in elaborate trait determination models.
4. Bias in population diversity: Most genetic studies have been focused mostly on the populations in Europe. This emphasis creates a knowledge gap in the genetic makeup of traits in African, Asian, and Indigenous people. These gaps limit the generalizability of the findings and knowledge about human evolution in the world (Parker, 2023). Epigenetic effects: Epigenetics which refers to the presence of heritable changes in the expression of genes but not in the DNA sequence is a field that is still emerging but underexplored. Epigenetic processes such as DNA methylation, histone modification and non-coding RNA regulation may have a large effect on the expression of physical traits, but few studies have attempted to consider these processes with reference to observable characteristics.
5. Minimal longitudinal information: There is a significant amount of cross-sectional studies that assess genetic variation at one point in time. The longitudinal studies that track changes in traits and gene expression over the lifespan are very few yet necessary in understanding age-specific trait variation.

## Recommendations for Future Research

1. Considering the current literature and the gaps that have been determined, a number of strategic recommendations are proposed:
2. Expand population research: Start an inclusive study that involves a wide variety of populations in the world to clarify how the genetic variation affects physical traits of different ethnic and geographic groups. This effort will increase the representation of the world in genetic databases and make the results of the research more relevant.
3. Inclusiveness of environmental and lifestyle determinants: Future studies ought to combine genetic, nutritional, environmental and lifestyle information in order to develop holistic models that explain interaction of genetic factors and environmental effects on expression of traits.
4. Stress polygenic and epistatic interactions: It is urgent that more research is carried out to investigate the dynamics of how several genes interact to produce complex traits. These complex interactions can be identified with the help of the use of various sophisticated computational models and bioinformatics approaches.
5. Study epigenetics: Conduct studies on the relationship between epigenetic changes and phenotype expression and how these processes interact with genetic variation that is

inherited. This is one of the areas where the consideration of characteristics that are shaped by the environment and genetic factors is of great importance.

6. Longitudinal studies: The longitudinal studies can provide useful information about how genetic traits are expressed across the lifespan, how the environmental exposures influence the expression of genes, and the development of polygenic traits.
7. Promote genetic education: Educate more about human genetic diversity in schools and colleges and through community education. This information will be utilized in order to dispel the myths about genetics and human variation.

#### 4. Conclusion

The genetic variation is the basis of the variation of physical traits that can be as simple as Mendelian traits, or as complex as polygenic traits of humans. It has been found that important genes and markers are related to easily visible characteristics, and interaction between genes, environmental factors, and epigenetics are not completely comprehended. Future studies would need to take an integrative approach by using a combination of genetics, bioinformatics, environmental studies and epigenetics to make a more comprehensive account of human diversity. These studies will be of great implication to the fields of personalized medicine, the health of the people, anthropology, and education and will assist in clarifying how the interplay between biology and environment makes each individual unique.

In this review, genetic variation has been pointed out as the most important cause of human physical diversity. Variations in the DNA sequences, such as single nucleotide polymorphisms (SNPs), variations in chromosomal structure, and crossover and recombination of genes result in the noticeable features of eye color, hair type, skin pigmentation, height, and predisposition to some diseases. Whereas the Mendelian traits are simple and predictable in terms of their inheritance, the polygenic traits are multiple in nature and tend to be influenced by environment including nutrition, lifestyle and climate.

Research articles discussed in this paper indicate that certain genes are linked to certain physical characteristics. Indicatively, OCA2 and HERC2 are the genes that affect eye color, EDAR, and TCHH are the genes that affect hair texture, and hundreds of polygenic loci influence height and skin pigmentation. In spite of these results, the intricate interactions between the genes (epistasis), environmental factors and the epigenetic processes are yet to be clearly comprehended. Moreover, most genetic studies have been done on populations of European origins and this has created a gap in the knowledge of the genetic structure of traits in other populations.

In general, this review highlights the role of genetics in forming human attributes and the need to combine genetic, environmental, and epigenetic approaches to get a comprehensive idea of human diversity. Such knowledge is vital not only in academic research but also in personalized medicine, as well as in the field of public health and anthropology.

#### References

- Allied Academies Review. (2025). Genetic contributions to complex traits: Genome research and polygenic traits.
- Annual Review of Genomics and Human Genetics. (2024). Human genetic variation, 25, 1–30. <https://www.annualreviews.org>
- Encyclopedia Britannica. (2026). Genetic variation. <https://www.britannica.com>
- Frazer, K. A., Murray, S. S., Schork, N. J., & Topol, E. J. (2009). Human genetic variation and its contribution to complex traits. *Nature Reviews Genetics*, 10(4), 241–251. <https://doi.org/10.1038/nrg2554>
- Genetic Science Review. (2026). Epistasis: Overview of gene interaction effects on phenotype.
- Karczewski, K. J., & Snyder, M. P. (2018). Integrative omics for health and disease. *Nature Reviews Genetics*, 19(5), 299–310. <https://doi.org/10.1038/s41576-018-0008-9>
- Lander, E. S., & Schork, N. J. (1994). Genetic dissection of complex traits. *Science*, 265(5181), 2037–2048. <https://doi.org/10.1126/science.8085176>
- Manolio, T. A., Collins, F. S., Cox, N. J., Goldstein, D. B., Hindorf, L. A., Hunter, D. J., McCarthy, M. I., Ramos, E. M., Cardon, L. R., Chakravarti, A., Cho, J. H., Guttmacher, A. E., Kong, A., Kruglyak, L., Mardis, E., Rotimi, C. N., Slatkin, M., Valle, D., Whittemore, A. S., ... Boerwinkle, E. (2009). Finding the missing heritability of complex diseases. *Nature*, 461(7265), 747–753. <https://doi.org/10.1038/nature08494>
- McGraw Hill Education. (2023). Human heredity: Principles and issues.
- National Human Genome Research Institute. (2026). Understanding genetics: DNA, genes, and human variation. <https://www.genome.gov>

- Nielsen, R., Hellmann, I., Hubisz, M., Bustamante, C., & Clark, A. G. (2007). Recent and ongoing selection in the human genome. *Nature Reviews Genetics*, 8(11), 857–868. <https://doi.org/10.1038/nrg2187>
- Parker, R. (2023). Unraveling genetic variation: Understanding human diversity. *Journal of Genetics and Molecular Biology*, 15(2), 45–58.
- Stranger, B. E., Stahl, E. A., & Raj, T. (2011). Progress and promise of genome-wide association studies for human complex trait genetics. *Genetics*, 187(2), 367–383. <https://doi.org/10.1534/genetics.110.120907>
- Visscher, P. M., Wray, N. R., Zhang, Q., Sklar, P., McCarthy, M. I., Brown, M. A., & Yang, J. (2017). 10 years of GWAS discovery: Biology, function, and translation. *American Journal of Human Genetics*, 101(1), 5–22. <https://doi.org/10.1016/j.ajhg.2017.06.005>