

Introduction to
Anthropology:
Holistic and Applied
Research on Being
Human

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MODULE 4: EVOLUTION AND GENETICS

Basic Genetics

In this chapter, we discuss some basic genetic principles, including biological inheritance and Darwinian evolution. Then, we present several examples to demonstrate how culture shapes the human genome. Finally, we discuss the importance of ethical research and the relationship between science and religion and demonstrate that these belief systems are not mutually exclusive.

Humans have distinct cultures across time and space. These cultures dictate the way we dress, the foods we eat, the languages we speak, the way we interact with one another, and dictate what behaviors are considered appropriate. Interestingly, these cultures can also influence our genetics. In other words, the genetics of our offspring are impacted by our cultural practices. Throughout this chapter, we will study examples of this relationship.

All living organisms have **genes** that determine what traits an individual expresses. Genes come in pairs, known as **alleles**. An individual inherits one allele from each parent: they may inherit the same or a different version of a gene, and this combination of genes impacts the way an organism looks or acts. These genes determine things like hair, eye, and skin color, the shape of our ear lobes and noses, or aspects of our personality, to name a few. Since offspring inherit half of their gene pair from the mother and the other half

from the father, offspring are not carbon copies of their parents. Instead, offspring display differential expression of traits based on the combinations of genes inherited from both parents.

This relationship is at the heart of **Mendelian genetics**. Gregor Mendel was a biologist who spent years breeding pea plants in the mid-1800s. Through this work, he discovered the **Laws of Heredity**, which is broken down into three laws. First, the **Law of Segregation** states that parent sex cells contain one pair of a gene. Therefore, when a male and female sex cell unite, the offspring inherits a full pair of genes. Second, the **Law of Independent Assortment** indicates that genes are not dependent; inheriting one gene does not necessarily indicate what other genes might be inherited. Additionally, because offspring inherit different allele pairs, they can have new combinations of genes that are not present in either parent. Third, the **Law of Dominance** states that if an individual inherits two different alleles of a gene, the **dominant** gene is expressed or visible, while the **recessive** gene is not. In other words, dominant alleles mask or hide the effects of recessive alleles.

Mendel illustrated these laws through pea plants. Pea plants are self-pollinating, and their seeds are either yellow or green. Mendel wanted to understand how seed color was determined, so he divided plants into groups based on their seed color. A yellow-seed pea plant can self-pollinate and produce a yellow-seed pea plant, while a green-seed pea plant self-pollinates to produce a green-seed pea plant. However, Mendel was curious to see what would happen if he bred a yellow-seed and green-seed pea plant together. When he cross-pollinated these plants, he discovered that a plant with both types of alleles does not produce a mixed yellow-green seed. Because seed color is a **discrete** trait, the alleles do not mix like paint colors. Instead, only the dominant allele is visible. Since the yellow-seed allele is the dominant form, only the yellow-seed allele is expressed. Therefore, the hybrid plant will have yellow seeds, even though it has two different alleles, because the green-seed allele is recessive and is not expressed. A pea plant must have two green-seed alleles to produce green seeds.

Similarly, some human traits are discrete, such as freckles. People either have freckles or they don't. A person cannot have incomplete or partial

freckles. If two people, one with freckles and one without, reproduce, then their offspring will either have the trait or not. Because freckles are a dominant trait, an offspring is more likely to have freckles than not.

Because of this relationship of dominant and recessive genes, Mendel reasoned that offspring with hybrid genes will exhibit a 3:1 ratio of dominant to recessive traits. This is illustrated below in the **Punnett square** (see Table 4.1). An offspring can inherit **homozygous** alleles, such as two dominant or two recessive traits, or they inherit **heterozygous** traits with one dominant and one recessive allele. In these cases, if a dominant allele is present at all, whether homozygous or heterozygous, the individual will express the dominant trait. Recessive traits are only expressed when genes contain homozygous recessive alleles.

Table 4.1. Example of Punnett square for freckle traits

| Hybrid Offspring | Freckles Allele (A) | No Freckles Allele (a) |
|-------------------------|---------------------|------------------------|
| Freckles Allele (A) | AA | Aa |
| No Freckles Allele (a) | Aa | aa |

How do these concepts relate to anthropology when humans are more complex than pea plants? We contain both discrete and **continuous** traits. Discrete traits usually have only two alleles. For example, freckles or no freckles, presence or absence, or either/or states. Discrete human traits, represented by one gene and two alleles, includes features like male baldness, body hair, cleft chins, dimples, earlobes, eyelash length, blood Rh factor, and toe length. Continuous traits, which are polygenic (having a range of possible expressions) and controlled by multiple genes, determine characteristics like stature and hair or skin color. Another example of a continuous trait are human leukocyte antigens, which are shaped by more than 200 genes and more than 300 alleles and related to our immune responses. Unlike discrete traits, alleles for continuous traits can mix like paint colors. If someone with very dark skin reproduces with someone with very light skin, their offspring can have a skin color that may be close to one of the parents or somewhere in between. If a very tall and a very short person reproduce, their offspring will mature into an adult with a

stature that may be somewhere in between the parents' statures. Further complicating the matter, some traits are linked to biological sex, such as color blindness and certain genetic disorders. Other traits only be expressed based on external conditions, such as diet. These complex relationships help explain human variation across the globe.

Darwinian Evolution

Mendel's work aligned with some of the tenets of **Darwinian Evolution**. Darwin's ideas on speciation and variation significantly impacted how people understood themselves and their place in the world. **Evolution** refers to changes in heritable traits of a population over generations. To explain how evolution occurs, Darwin forwarded ideas like **natural selection, mutations, gene flow, and genetic drift** to describe processes that organisms use to adapt to environmental challenges and perpetuate their species with successful reproduction. These principles and mechanisms of natural selection are still foundational to evolution studies today. Natural selection suggests that populations will have different frequencies of traits in a population, and only the fittest traits (traits that help individuals reproduce) will be selected and remain prominent while lesser fit traits will decline. Over time, a population changes so that the fittest traits become more common. Mutations are essentially "typos" in genetic sequences. While mutations represent a weak evolutionary force, they are essential as the ultimate source of variation on which natural selection operates.

Acclimation and adaptation are two types of natural selection. **Acclimation** refers to temporary, involuntary changes experienced by individuals that help improve survival in stressful environments. Acclimation does not indicate genetic change. However, over time, populations that acclimate to specific environmental conditions may begin to see longer-term genetic changes as specific traits are prioritized. **Adaptations** refer to genetic changes within a population that improve survival odds for individuals.

An individual acclimates through a series of physiological changes to their body that helps them survive or handle environmental stressors. For example, tanning is an example of seasonal acclimation that provides a

natural shield against ultraviolet radiation. Ultraviolet radiation can damage DNA, deplete folate nutrients necessary for neural tube development, and lead to skin cancer, but animal bodies can darken their skin with melanin pigments to reduce ultraviolet penetration. Some animals can use these pigments to absorb heat, color feathers and scales, or even sharpen their vision.

Bergmann’s rule and **Allen’s rule** both illustrate the idea of adaptation (see Table 4.2 and Figure 4.1). Together, these rules posit that organisms who live at higher latitudes or colder regions will have relatively large, thick bodies with short limbs to conserve heat. Conversely, individuals who live at lower latitudes or warmer regions will have relatively small, lean bodies with long limbs to expel heat. Bergman noticed this trend even within the same species of animals. For example, adult male white-tailed deer weigh an average of 125 lbs. in subtropical Florida but 250-275 lbs. in cooler, temperate Montana.

Bergmann observed that large animals have a relatively smaller surface area compared to their body mass because volume increases at a faster rate than surface area. This relationship allows large animals to conserve more heat. He also hypothesized that large animals were able to produce more heat. Therefore, larger body size was an advantageous adaptation to cooler regions. Examples of human adaptations are presented below.

Table 4.2. Summary of Bergmann’s and Allen’s Rules.

| Environmental Conditions | High Latitude Adaptations | Low Latitude Adaptations |
|---------------------------------|-----------------------------|----------------------------|
| Bergmann’s Rule | Large, thick-bodied animals | Small, lean-bodied animals |
| Allen’s Rule | Short limbs | Long limbs |

Other evolutionary forces include genetic drift and gene flow. Genetic drift reflects random changes in trait frequency when populations diverge, while gene flow refers to the mixing of two or more populations to become more similar. Evidence of evolution as an ongoing process can be found throughout the world. Taken together, these processes mean that evolution never stops. Several examples are provided to illustrate the

mechanisms of natural selection, genetic drift, and gene flow.



Figure 4.1. Northern red fox (left) and southern desert red fox (right), illustrating Bergmann's Rule. Image from Wikimedia Commons.

Kaibab Squirrels and Allopatric Speciation

Allopatric speciation is a specific type of genetic drift that occurs when a population diverges and becomes geographically isolated. This isolation stops gene flow and can lead to different subspecies or entirely new species with enough time. Abert's squirrel (*Sciurus aberti*) is a tassel-eared squirrel with a white underbelly, reddish stripe down the back, and a grey tail (see Figure 4.2). Abert's squirrel is found throughout the American Southwest in parts of Mexico, Colorado, Utah, and Wyoming. They are tree squirrels who rely almost exclusively on Ponderosa pine forests and build nests high in the branches of these trees, and their diet is comprised predominantly of pine products. In warm seasons, they eat seeds and buds, and in the winter, they eat the inner bark of the tree.

The Kaibab squirrel (*Sciurus aberti kaibabensis*) is one of the rarest mammals in North America. It represents a distinct subspecies of the Abert's squirrel that is only found in the Kaibab Plateau of Northern Arizona. This region is at the remote northern edge of the Grand Canyon. The Canyon acts as a geographic barrier with Kaibab squirrels at the North Rim, and Abert's squirrels at the South Rim. Kaibab squirrels evolved when they became reproductively isolated from Abert's squirrels around 10,000 years ago. The Kaibab squirrel differs in appearance and is characterized by a dark grey body and white tail.

Researchers debate whether the Kaibab squirrel represents a distinct species or subspecies of Abert's squirrel, and recent research suggests that Kaibab squirrels are highly diverged from all other Abert's squirrel subspecies. Interestingly, however, this isolation may not have been complete. There is evidence of DNA **introgression** and at least two migration events over the past 10,000 years, wherein Kaibab squirrels interbred with other types of Abert's squirrels. This suggests that a “biological” definition of species, based on reproductive isolation, is not a requirement to define speciation. Instead, we refer to the “ecological” definition of a species, where geographically isolated populations become adapted to their local environments and may not be suited to other habitats.



Figure 4.2 Abert's squirrel (left) and Kaibab squirrel (right). Images courtesy of Wikimedia Commons.

Genetics and Human Culture

Modern humans (*Homo sapiens*) have existed for well over 100,000 years (see Module 7: Genus Homo and First Cultures). Today, multitudes of cultural groups exist across the globe who vary in terms of practices, genetic adaptations, and short-term acclimation, but we are all part of one human species. However, that human species has changed over time, and humans today are different from early humans in significant ways. In fact, we are even substantially different from humans that existed only 10,000 years ago. As people have responded to local challenges in the world around them (environmental conditions, disease, or other human action), there have been corresponding local changes to culture and genetics. These changes demonstrate the **biological plasticity** of humans.

High-Altitude Acclimation and Adaptation

Living at high altitudes causes physiological and health impacts that are not seen in people who live at lower elevations. In high regions, breathing is difficult, temperatures are colder, food resources are sparse, and ultraviolet radiation is more intense. While air at any altitude contains approximately 21% oxygen and 79% nitrogen, the amount of air pressure decreases as altitude increases. This decreased pressure makes breathing and physical activity more difficult and is associated with **hypoxia**. The low air pressure also impacts pregnancy, birth weight, and overall health. Lower pressure means that it is more difficult to intake adequate oxygen supplies, which results in less availability of oxygen in the blood, and a body that works harder to circulate that blood.

However, people can acclimate in the short-term and populations can adapt in the long term to deal with these challenges. This acclimation includes changes in breathing patterns, heart pressure, and red blood cell count. For example, even when only visiting high altitudes for a short time, our bodies respond by thickening the blood with more red blood cells and hemoglobin, which increases the amount of oxygen in the blood. With these types of short-term benefits in mind, some athletes choose to include altitude acclimation in their training to boost their performance in competitions at lower altitudes.

For people who stay in these regions long-term, however, population-level adaptations are observed. People in different high-altitude regions, from Peru to Tibet to Ethiopia, demonstrate various genetic changes that impact heart muscles, digestion, bone growth and development, and/or red blood cell and hemoglobin levels. These genetic variations differ from low-altitude populations, and also between other high-altitude populations. In fact, these different high-altitude populations represent an example of **convergent evolution**, where the genetics of each group differ, but reflect adaptations to similar environmental challenges (see Table 4.3 and Figure 4.3).

For example, Ethiopian groups adapted to high altitudes tend to have increased hemoglobin levels and variant mitochondrial functions that impact

energy production. Andeans have increased hemoglobin levels and barrel-shaped chests that help them intake more air. However, Ethiopians and Tibetans do not have barrel-shaped chests, and Tibetans do not have increased hemoglobin levels. Increased hemoglobin can strain the body and heart as it pumps thicker, more concentrated blood. Instead, Tibetan groups breathe at a faster rate, exhale more nitric oxide, and process vitamin D differently. This Tibetan adaptation leads to **vasodilation**, or wider blood vessels. This adaptation has been traced to Denisovan genetics (see Module 7: Genus Homo and First Cultures), indicating that the ancestors of Tibetan and Han Chinese people reproduced with Denisovans.

Table 4.3. Example of convergent evolution among high-altitude human populations.

| High-Altitude Adaptations | | | | | |
|---------------------------|------------------------------------|------------------------------------|--------------------------------|--------------|-------------------------|
| Population | Mitochondria 1 DNA functions | Increased hemoglobi n levels | Barrel- shape d chest | Vasodilation | Vitamin D processing |
| Ethiopian | X | X | | | |
| Andean | | X | X | | |
| Tibetan | | | | X | X |

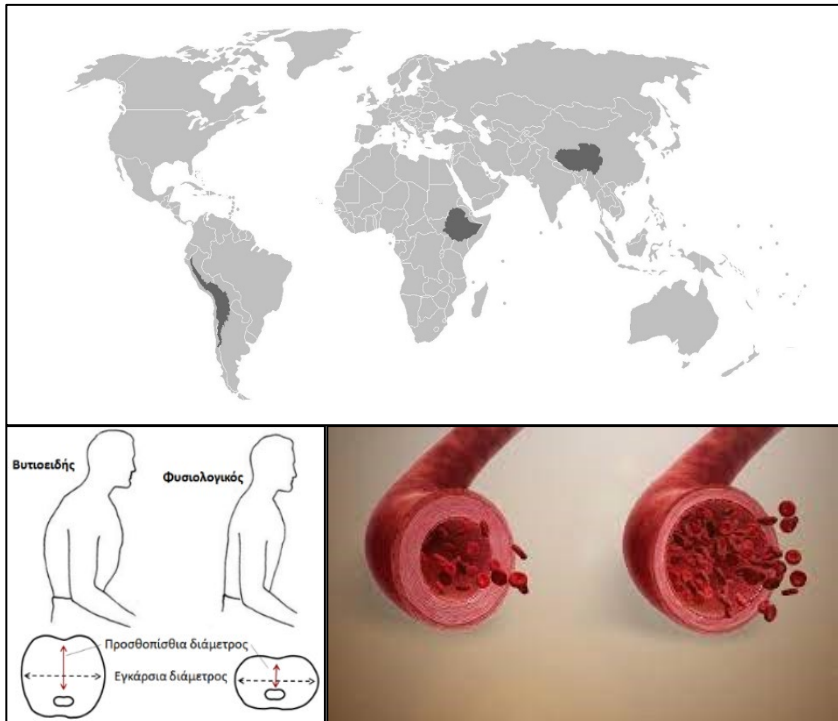


Figure 4.3. Map of high-altitude populations and examples of convergent evolution: barrel chests and vasodilation. Images from Wikimedia Commons.



Video 4.1. Check out a video about an interview with Cynthia Beall about her research on high-altitude adaptations among human groups for more details.

Lactose Tolerance – A Genetic Adaptation Linked with Animal Domestication

Other genetic adaptations are more widespread. Human infants often rely exclusively on milk for the first few months of their lives. However, eventually their caloric needs to expand beyond milk, and children begin including other types of nutrition in their diet. Ten thousand years ago,

humans could not digest milk after infancy because milk contains sugar known as lactose, and adult humans were lactose intolerant. Their bodies did not contain enough of the lactase enzyme to properly process the lactose sugars. Therefore, people became sick if they drank milk because it acted like a toxin in their bodies. Today, thanks to a genetic mutation, approximately 35% of people, predominantly of European ancestry, can drink milk with no negative side effects.

Scientists can track an increase in lactase enzymes, or **lactose tolerance**, in ancient human populations that aligns with their domestication of dairy animals, such as cows and goats starting 10,000 years ago (see Module 9: Development of Agriculture). People learned to ferment milk to make cheese and yogurt, the fermentation process reduced the amount of lactose present, and people could tolerate these products. However, this process also reduced the food calories by up to 50%. Lipid evidence suggests that people were sieving milk fat from liquid whey around 7,000 years ago, and biomolecular evidence indicates that people were eating cheese in Egypt 3,000 years ago. These dietary shifts were possible because of a **genetic mutation** that spread across Europe, the Middle East, and Africa. This mutation allowed people to produce enough lactase to process milk. As this mutation became more prevalent in human gene pools, people were able to introduce milk into their diets with all their caloric content. These genetic adaptations boosted human rates of survival and provided another avenue of resources in case of agricultural crop failure. Because it was an advantageous trait, it increased and spread throughout these populations.



Video 4.2. Check out a Sci Show video discussing how lactose tolerance changed human history for more details.

Gene Flow and Genetic Drift Among Oceania Seafarers

Today, anthropologists use genetics to identify different hominin species, understand processes of domestication, trace disease origins, and track migration patterns. However, the genetic processes are not always clear cut. For example, thousands of years ago, early Oceanic settlers spread from

east Asia to colonize dozens of small islands spread across the Pacific Ocean in one of the major events of human history. These voyages were often long and difficult with only the stars, seabirds, or wind currents to help seafarers navigate entirely new areas and discover tiny islands in the great expanse of the ocean.

Understanding this massive exploration and settlement pattern requires interdisciplinary approaches. Anthropologists from all four fields, folklorists, historians, oceanographers, and other scientists are working to unravel the mysteries. While researchers are interested in the migrations and island settlement patterns, including who first undertook these journeys and where they began, the nature of the archaeological and genetic records limit interpretations. Today, people debate where the Austronesian expansion began, but many researchers suggest that it started in Taiwan around 5,000 years ago. The nature of these trans-Oceanic movements and cultural interactions through time complicate our understanding of this major world event.

The first Oceania seafarers were brilliant navigators that were able to decipher subtle clues on the horizon to find land amid the endless waves. Laden with domestic animals and plants—chickens, pigs, dogs, taro, bananas, yams, breadfruit, and sugar cane—ceramic pottery, bone and stone tools, and some incidental species like rats, people ventured into this unknown expanse. Over several hundred years and spanning thousands of kilometers, people made their way east. By 1,000 years ago, numerous communities and cultures had been established across islands throughout the western Pacific region.

Oceania is comprised of three distinct geographical regions: Melanesia, Micronesia, and Polynesia. Melanesia is the western-most of these regions and was settled first. From there, seafarers moved north into the islands of Micronesia, then they edged into western Polynesia. These movements took around 500 years. This exploratory process represents a specific type of genetic drift known as the **Founder's Effect**. As people ventured into the unknown ocean, they discovered and settled new islands to develop a community. However, eventually a small subset of the island's population would break away to continue venturing across the ocean. These smaller subset populations had less genetic diversity than their parent population and,

thus, different allele frequencies than their parent populations. This process was further compounded with each subsequent migration.

Upon reaching western Polynesia, eastward migration stopped for a few hundred years before continuing into the rest of Polynesia. Surprisingly, people maintained contact between these three geographic regions. They were not traveling in one unilinear direction. There is evidence of back migration in linguistic connections, oral traditions, and genetic data. Additionally, parts of Melanesia and Micronesia were already inhabited prior to the arrival of the Oceania seafarers, and these communities interacted. These back migrations and interactions illustrate the concept of gene flow, which would have partially negated the impacts of the Founder's Effect. These complicated patterns of habitation and migration are part of the reason it is difficult to trace genetics of people across Oceania.

The issue of migration and settlement patterns becomes even more complicated when European colonization is considered. By 400 A.D., the Hawaiian Islands were occupied, and by 800 A.D., Rapa Nui was settled. These two locations represent the furthest extent and last island settlements established in Polynesia. Polynesia was occupied for nearly 1,000 years before European contact. It wasn't until the 1700s that the first Europeans made their way to this region.

European groups were surprised to find dozens of islands occupied across the Pacific Ocean. As they sailed between islands, they met distinct cultural groups, speaking different types of Austronesian languages and with their own cultural histories, yet surprisingly connected despite the distances. However, European groups inadvertently brought diseases with them as well, which contributed to the massive loss of life and culture across Oceania. In some cases, more than 90% of an island's population was wiped out from disease, and much of these cultures—people, customs, and histories—were lost.

These losses represent another type of genetic drift known as the **Bottleneck Effect**. With most of a population dying suddenly from disease and other contact-related causes, entire generations were severely diminished. Only a handful of people remained on these islands, and their

cultural and genetic diversity were greatly reduced compared to the original population. In other words, a daughter population that had already been significantly reduced through the Founder's Effect. Even today, these groups grapple with the uncompromising impacts of colonialization. Migrations of small populations over large distances can significantly impact a population's genotype and phenotype, even without substantial influences from natural selection. The Austronesian expansion into the Pacific is a good example of evolution whereby evolutionary forces of genetic drift and gene flow play an important role with natural selection.

Neanderthal Genetic Mutations

We mentioned above that high-altitude Tibetan communities may have inherited DNA from Denisovan ancestors. That is just one example of mutations and inherited genes from early human ancestors. Neanderthals (*Homo neanderthalensis*) are one of our closest extinct human relatives (see Module 7: Genus Homo and First Cultures). The complete Neanderthal genome was sequenced in 2010. Evidence suggests that humans and Neanderthals were able to interbreed, and those interactions left lasting impacts on our genetics today. People with European and Asian ancestry share between 1-4% of our genetic makeup with Neanderthals. Individuals with sub-Saharan African ancestry share about 0.3% of their genomes with Neanderthals. Evidence suggests that Neanderthal DNA migrated back to Africa in the last 20,000 years through populations who carried the DNA from previous Neanderthal matings.

Why does this 1-4% shared genomes matter? Some of the corresponding traits may be viewed as a double-edged sword that advantaged and helped early humans survive in the past, but today, those same traits may cause more harm because of differences in culture, lifestyle, and environment. For example, genes shared between Neanderthals and modern humans have been associated with tougher skin, hair, and nails, and thicker insulation, which would have helped people adapt to colder environments. Other beneficial traits include increased blood clotting, which would have been helpful in the past when dealing with injuries and trauma without access to modern medical technologies. However, on the downside, these same traits are today linked to increased heart attack risks, obesity, addictive behaviors, and depression.

Other shared genetics are related to disease and immune system functioning. Recently, researchers have demonstrated that different shared traits between humans and Neanderthals can influence the impacts of major illnesses, such as COVID-19. Some people inherited traits that may increase the severity of COVID-19, while other people have inherited traits that reduce their risk for severe infection. These studies demonstrate the unintended aftermath when once-adaptive traits are presented with new challenges.

Vestigial Traits

While humans around the world exhibit genetic differences that allow some people to drink milk or live at high altitudes, not all of the changes serve a particular purpose. Humans display **vestigial traits** that no longer serve biological functions for our survival or reproductive fitness. In other words, these traits illustrate natural selection without causing death. These traits may have been inherited from far-distant ancestors in our primate or mammalian lineage and initially served important roles in our ancestors' survival, such as the *palmaris longus* tendon in our arms. While we retain these traits today, they are no longer necessary. Vestigial traits do not harm or disadvantage people. However, they have never been actively selected against, and so they persist in human populations.



Video 4.3. Check out a video on Vox presenting a discussion of vestigial structures

Genetic Anthropology and Ethics

As the examples above illustrate, genetic data is not often easy to interpret. There are so many complicating factors, including gene flow, genetic drift, vestigial traits, and convergent evolution. However, biological anthropologists have used genetic data to understand the peopling of the world, how people adapt to various environmental and cultural forces, and relationships among groups of people. We must also consider ethical

concerns related to factors such as conflicting belief systems, consultation, and consent. Considering anthropology's insensitive origins of studying human differences (see Module 2: A Brief History of Anthropology), it is imperative that we ensure that study subjects are treated with respect and compassion and that we avoid interpretations that may cause harm to them and their culture.

One ethical concern involves when genetic data does not align with a population's beliefs about their own origins and history. Consider the public's response to Darwin's ideas about evolution in the 19th century. Darwinian evolution was disputed and rejected in parts of Europe and America in part because it conflicted with Christianity's origin stories or was associated with rejected scientific ideas. Even today, roughly 20% of Americans reject the concept of evolution, often citing it as irreconcilable with their religious or other cultural belief systems. In many cases, people's religious and cultural beliefs are so foundational to their identities that divergence from these beliefs may be perceived as a threat to a culture's way of life.

Additionally, genetic data has not always been collected honestly and stored with consent. For example, in 2003, a small mummified, human-like skeleton was discovered in the Atacama Desert of Chile. The mummified skeleton was only 6-in. long and had large eye sockets and a long, pointed skull. News outlets likened the remains to otherworldly aliens. However, a team of researchers analyzed the remains and determined that the skeleton belonged to a young child (6-8 yrs.) of Chilean descent with numerous congenital disorders arising from genetic mutations.

The Atacama skeleton research was received with backlash from the biological anthropological community. They argued that the research was flawed, defied scientific research conventions, provided gross misdiagnoses, and misrepresented the materials at hand. Based on the data provided in the original research, Halcrow and colleagues determined that the skeletal remains likely represented a miscarriage or stillbirth around 15 weeks gestational age and not an older child. The elongated skull was attributed to birth deformation and deterioration from the postmortem environment. Halcrow and colleagues were unable to find any skeletal evidence for genetic mutations in the remains, and they argued that the original research did not

follow ethical and legal standards in anthropology. No local groups were consulted, no ethical consent or archaeological permits were acquired, and the human remains were sold for financial gain.

In other cases, study participants were provided incomplete information or misled about the purposes of biological data collections. People cannot give informed consent to participate in a study without appropriate information. However, there are numerous examples where requisite information was withheld. The Tuskegee Syphilis Project represents one example of these (see Module 16: The Issue with Race).

In another instance, the Nuu-chah-nulth people of British Columbia, Canada, were told they submitted genetic data for a rheumatoid arthritis study. Rheumatoid arthritis, an autoimmune and inflammatory disease, impacted these peoples more frequently and severely than other groups, and they hoped to discover the cause and potential cures. However, they later discovered that their genetic data—more than 800 blood samples—were being used to study human migration and viruses, but not to help them understand the prevalence of rheumatoid arthritis in their communities. The data was not used for the reported purposes, and the Nuu-chah-nulth did not benefit from the study. Furthermore, the researchers never reported any pathological results to the Nuu-chah-nulth people as promised. The group learned how their genetic data was used when they viewed a documentary that interpreted their ancestors' migration patterns. As a result, the Nuu-chah-nulth formed a research ethics committee and worked with the Canadian Institutes for Health Research to ensure indigenous people received proper respect and information when working with researchers.

It is important to understand the process of evolution to understand humans' past, present, and future. However, appropriate protocols must be enacted to ensure that research is conducted soundly and ethically. People must be properly informed about the purpose of a study, permissions must be acquired, and interpretations must be rigorous and reliable. Anything less undermines the very institutions of science and research.

Science and Faith Based Belief Systems

Because of the public debate that persists in the United States today,

science and faith are briefly compared here. Science and religion are often presented as polar opposites, and schoolboards and teachers clash about how to present human origins and evolution to students. Some proponents of these belief systems mistakenly designate them as mutually exclusive, and legal battles continue as schools grapple with whether evolution or **creationism** should be taught in American public schools. Students may not even recognize this contentious aspect of their education until years later.

Science and religion are complex topics, but they do not need to exist in opposition. People who stoke the fires of this debate may present a false dichotomy between the two, which misses the fact that science and faith study very different things using different methods and criteria that may not overlap. While both concepts are difficult to define, science emphasizes only the natural world, while religion explains both the natural and the supernatural, including forces that cannot be directly observed, such as higher-level god entities, angels, miracles, karma, or souls (see Module 18: Religion). Supernatural variables cannot be tested or falsified in the natural world, and so these topics lay outside the purview of science. This perspective represents an **independence model** on the relationship between science and religion.

Proponents of the independence model suggest that religious leaders should not confuse faith and irrefutable fact. Religious perspectives cannot be used to make factual claims about evolution and science because they rely on indirect associations or intermediate variables to link the supernatural to the natural. Scientific perspectives cannot explain supernatural, moral, or ethical questions, and they rely only on observable variables. Others argue that the independence model is flawed, and science and religion overlap and must occasionally respond to questions where they intersect. The **dialogue model** presents a mutualistic relationship between religion and science. While faith or religion perpetuates the idea that the world and all creation was designed by a higher-level entity, this creator is often depicted as intelligible and orderly. In this regard, science can untangle aspects of creation and identify laws put into place by the creator.



Video 4.4. Check out a video on Brian Enderle discussing the relationship between science and faith in this *TEDx* talk

Because these topics are concerned with different basic questions and methodological approaches, they can both exist without inconveniencing the other. Furthermore, they are not mutually exclusive because they are trying to explain different (though arguable related) phenomena (e.g., natural only vs natural and supernatural). These approaches to understanding the world should not be presented as if they address the same questions or use the same methods to somehow provide conflicting results. These perspectives rest on completely different foundations.

Review Questions

- **T/F.** Discrete traits are polygenic and controlled by multiple genes.
- **T/F.** Today, approximately 35% of the human population can drink milk thanks to a genetic mutation that occurred thousands of years ago.
- **T/F.** Different high-altitude populations display different adaptive traits because of convergent evolution.
- **T/F.** The Founder's Effect and Bottleneck effect are types of gene flow.
- **T/F.** Natural selection is a process that individuals use to adapt to cultural challenges and establish hierarchical societies.

Discussion

- There is often great variation among individuals of animal species. For example, look around your classroom and list the wide range of characteristics that vary among humans such as height, skin color, body stature, etc. Why is such variation important? What role does physical variation play in evolution?
- How can genetic studies help us understand human origins and cultural diversity?
- How do concepts like gene flow and genetic drift complicate our ability to understand human migration?
- What is convergent evolution, and how can it help us understand adaptation?

Activities

1. Place a bunch of store-bought broccoli in a vase half filled with water for several days and watch what happens. Broccoli is a good example of a common vegetable that owes its existence to evolution by artificial selection. Along with brussels sprouts, cauliflower, collard greens, kale, and a few other vegetables, broccoli has evolved from a single plant species: *Brassica oleracea*. About 2,500 years ago, *B. oleracea* grew wild along the coast of Britain, France, and the Mediterranean. That wild form, what we call wild mustard, still exists today. Over millennia, beginning in ancient Greece and Rome, farmers preferentially planted seeds from plants that grew more leaves, and after many generations this kind of [artificial selection](#) produced a leafy version of wild mustard that looked something like modern-day kale. Sometime after 1600 A.D., farmers selected variants of the plant that produced enlarged leaf buds, which ultimately gave us cabbage. Elsewhere, farmers selected enlarged flowering structures, eventually creating broccoli and cauliflower. This it's still going on: in 1993, a Japanese seed company bred broccolini, a hybrid between broccoli and kai-lan, yet another form of this plant species, known as Chinese broccoli. When you let the broccoli flower, you are seeing the plant part that was preferentially selected centuries ago.
2. In a large bowl, prepare a variety of candies for a large group gathering (e.g., your classroom). Be sure to include popular ones (e.g., Hershey Kisses, Starburst) and unpopular ones (e.g., Circus Peanuts, black licorice). You'll need at least two candies per person plus plenty of unpopular ones. Try to include as many candies with different colors, sizes, brand names, etc. as possible (avoid candies with nuts if you are unsure of allergies). Prepare a list of the candies and their original abundance, then pass the bowl around the room a couple of times.
 - Once more than half of the candy is gone, gather together. Count what candies remain and compare the new list to the original list.
 - List the candy "traits" of the candies selected (e.g., chocolate flavor, large size, favorite brand, etc). These are the traits that led to the removal of certain candies.
 - List the candy "traits" of the ones NOT selected (e.g., bad flavor, small size, aftertaste). These are the traits that allowed the candies to survive being passed around the room.

The fact that there were many different candies with different traits resulted in some candies being eaten and others surviving. This is what natural selection does with individuals in a population. Each individual has unique traits; some traits help an individual survive and some traits do not. Thus,

an important reason for variation is that variation allows for differential survival of individuals. [This activity is modified from Carol Tang, California Academy of Sciences

(https://ucmp.berkeley.edu/education/dynamic/session3/sess3_act1.htm]

3. If you're an instructor, consider using a bag of M&Ms to simulate the founder effect and bottleneck effect, two examples of genetic drift. There are several variations of this activity available online. One we recommend is available here, or alternatively, another variation of this activity is available at <https://academic.oup.com/bioscience/article/52/4/373/238712>.
4. Watch the Vox video *Proof of Evolution* at <https://www.vox.com/2016/3/17/11250962/proof-evolution-vestigial>. Do you have a *palmaris longus* in one or both wrists? Do you get goosebumps? Can you wiggle your ears? What is the purpose of these traits in humans versus other animals? Why do you think we get goosebumps not just when we are physically cold, but also when we are moved emotionally?

Key Terms

Acclimation: Temporary, involuntary, physiological processes experienced by individuals that help improve survival in stressful environments but do not consist of genetic changes.

Adaptation: The process in which an organism makes successful genetic adjustments to a specific environment to improve their survival odds.

Alleles: One of two or more alternate forms of a gene.

Allen's Rule: A continuation of Bergman's rule that also posits that warm-bodied organisms living at higher latitudes or colder regions tend to have shorter limbs in order to lose less heat to the surrounding environment.

Bergman's Rule: A principle that posits that among warm-bodied organisms, a pattern exists between living at higher latitudes or colder regions and having larger and thicker bodies to conserve heat.

Biological plasticity: The ability for living organisms to alter their phenotypes to adapt to the environment they are in.

Bottleneck Effect: A type of genetic drift, when the majority of a population dies suddenly from contact-related causes such as disease, environmental events, or other outside causes, leading to significant reductions in population.

Continuous traits: Polygenic traits controlled by multiple genes, with ranges of expressions such as hair color, weight, or skin color.

Convergent evolution: Where distantly related groups with different genetics independently evolve similar traits to adapt to similar environmental challenges.

Creationism: A doctrine or theory that believes everything in the universe was created by God out of nothing.

Dialogue Model: A model that presents a mutualistic relationship between science and religion, that they can intersect and overlap to help explain aspects of each other.

Discrete traits: Traits controlled by a small number of genes, generally one, and two alleles. For example, dimples or no dimples.

Dominant alleles: The expressed or visible allele that as a variation of a gene will produce a certain phenotype when an individual inherits two different alleles in a gene.

Evolution: The process of change in heritable traits of a population over generations.

Founder's Effect: A type of genetic drift, when a subset of the population breaks away to establish a new community, creating less genetic diversity and variation within the new community compared to their parent population, as well as different allele frequencies.

Gene flow: The exchange or mixing of genes between two or more populations to become more similar.

Genes: Discrete units of hereditary information that determine specific physical characteristics or traits of organisms.

Genetic drift: Random changes in allele or trait frequencies within a population because of random processes of selection when populations diverge.

Heterozygous: Having two different alleles in a gene pair.

Homozygous: Having the same alleles in a gene pair.

Hypoxia: A state resulting from low air pressure, commonly at higher altitudes where the lower pressure makes it more difficult to intake adequate oxygen supplies, which results in less oxygen available in our blood and makes the body work harder to circulate that blood.

Independence model: A model that presents a compatible but separate view of science and religion, that they do not overlap and therefore cannot be used to make factual claims about each other.

Introgression: The transfer of genetic material from one species to another because of hybridization and backcrossing.

Lactose tolerance: A genetic adaptation of increased levels of lactase enzymes that allows for the ability to digest lactose, a sugar found in dairy products.

Law of Dominance: The third Law of Heredity that states that if an individual inherits two different alleles of a gene, the dominant gene is expressed or visible, while the recessive gene is not.

Laws of Heredity: After his work with breeding peas plants Austrian biologist, mathematician, and monk, Gregor Mendel, also known as the “Father of Genetics,” created the three Laws of Heredity to explain the transmission and inheritance of genetic traits.

Law of Independent Assortment: The second Law of Heredity that indicates that genes are not dependent; inheriting one gene does not necessarily indicate what other genes might be inherited. Additionally, because offspring inherit different allele pairs, they can have new combinations of genes that are not present in either parent.

Law of Segregation: The first Law of Heredity that states that parent sex cells contain one pair of a gene. Therefore, when a male and female sex cell unite, the offspring inherits a full pair of genes.

Mendelian genetics: The study of offspring that display differential expression of traits based on combinations of genes inherited from both parents, founded by Gregor Mendel.

Natural selection: A theory presented by Charles Darwin that nature or environmental circumstances determine which characteristics are essential for survival and that individuals better adapted to their environments with these characteristics are more likely to survive and pass them on to their offspring.

Punnett Squares: A square table of four quadrants used to illustrate the possible percentage of phenotype outcomes for an offspring of a genetic cross between two individuals with known genotypes.

Recessive alleles: The masked allele that as a variation of a gene will not produce a phenotype if a dominant allele is present when an individual inherits two different alleles in a gene.

Vasodilation: The dilatation or widening of blood vessels.

Vestigial traits: Traits present in human populations that have never been actively selected against and may have initially served important roles in our ancestors' survival, but no longer serve biological functions for our survival or have a positive impact on reproductive fitness, which illustrates natural selection without causing death.

Suggested Readings

Barras C. 2016. "Our Neanderthal genes linked to risk of depression and addiction." *NewScientist*. Electronic document, <https://www.newscientist.com/article/2077269-our-neanderthal-genes-linked-to-risk-of-depression-and-addiction/>, accessed 17 Feb. 2021.

Bickle P. 2018. "Why can't most humans drink milk?" *Sapiens*. Electronic document, <https://www.sapiens.org/biology/lactose-intolerance-cheese-evolution/>, accessed 10 Nov. 2020.

Choi CQ. 2016. "Neandertal-human trysts may be linked to modern depression, heart disease." *Scientific American*. Electronic document, <https://www.scientificamerican.com/article/neandertal-human-trysts-may-be-linked-to-modern-depression-heart-disease/>, accessed 17 Feb. 2021.

Curry A. 2013. "The milk revolution." *Nature*, vol. 500, no. 7460, pp. 20-22. doi:10.1038/500020a. <https://www.nature.com/news/archaeology-the-milk-revolution-1.13471>

De Cruz H. 2017. "Religion and science." *Stanford Encyclopedia of Philosophy*. Electronic document, <https://plato.stanford.edu/entries/religion-science/>, accessed 5 Nov. 2020.

Goldman JG. 2014. "How human culture influences our genetics." *BBC Future*. Electronic document, <https://www.bbc.com/future/article/20140410-can-we-drive-our-own-evolution>, accessed 9 Oct. 2020.

O'Neil D. 2012. *Human Biological Adaptability: An Introduction to Human Responses to Common Environmental Stresses*. Palomar College, Electronic document, <https://www2.palomar.edu/anthro/adapt/Default.htm>, accessed 13 Dec. 2021.

Smithsonian Magazine. 2009. "Lactose tolerance and human evolution." *Smithsonian Magazine*. Electronic document, <https://www.smithsonianmag.com/arts-culture/lactose-tolerance-and-human-evolution-56187902/>, accessed 10 Nov. 2020.

Videos

Humans and High Altitude

<https://www.youtube.com/watch?v=dQTbTuxxSSw>

SciShow: *Milk and the Mutants that Love it* <https://youtu.be/ecZbhf96W9k>

TED Talks: *Science and Faith* <https://www.youtube.com/watch?v=aF-ptUJKli4>

Cosmos Season 01, Episode 02

TEDEd: *Five Fingers of Evolution*. <https://ed.ted.com/lessons/five-fingers-of-evolution>

TEDEd: *Myths and misconceptions about evolution*

<https://www.youtube.com/watch?v=mZt1Gn0R22Q>