Issues

• How can we describe and compare variation within and between populations?
• How is the study of population genetics related to human variation?
• Why do human populations differ biologically?
• Do human races exist?
• Is there a biological basis for the idea of race? Is biology the most accurate descriptor of race?
• Will changing biological concepts of race diminish racism? Why or why not?

Biological Concepts

• Populations and population ecology
• Population genetics (genetic variation, Hardy-Weinberg equilibrium, blood groups, genetic drift)
• Patterns of evolution (adaptation, physiology)
• Forces of evolutionary change (natural selection, environmental factors, communicable diseases, parasitism, human health)
• Gene action (molecular structure, genetic polymorphism)
• Scaling (body size and shape)

Chapter Outline

There Is Biological Variation Both Within and Between Human Populations

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The study of human variation

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Isolated populations and genetic drift
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Malaria and Other Diseases Are Agents of Natural Selection

Malaria
Sickle-cell anemia and resistance to malaria
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Population genetics of malaria resistance
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Natural Selection by Physical Factors Causes More Population Variation

Human variation in physiology and physique
Natural selection, skin color, and disease resistance
The human species is highly variable in every biological trait. Humans vary in their physiology, body proportions, skin color, and body chemicals. Many of these features influence susceptibility to disease and other forces of natural selection. Continued selection over time has produced adaptations of local populations to the environments in which they live. Much of human biological variation is geographic; that is, there are differences between population groups from different geographical areas. For example, northern European peoples differ in certain ways from those from eastern Africa, and those from Japan differ in some ways from those from the mountains of Peru. Between these populations, however, lie many other populations that fill in all degrees of variation between the populations we have named, and there is also a lot of variation within each of these groups.

Central to the study of human variation is the concept of a biological population, as defined in Chapter 5 (p. 151), and as explained again later. Both physical features and genotypes vary from one person to another within populations, but there is also a good deal of variation between human populations from different geographic areas as the result of evolutionary processes. How do populations come to differ from one another? How do alleles spread through populations? How do environmental factors such as infectious diseases influence the spread? Why are certain features more common in Arctic populations and other features more common in tropical populations? Why do we think of some of these variations as ‘races’? These are some of the questions that are explored in this chapter.

There Is Biological Variation Both Within and Between Human Populations

All genetic traits in humans and other species vary considerably from one individual to another. Some of this variation consists of different alleles at each gene locus; other variation results from the interaction of genotypes with the environment. The simplest type of variation governs traits such as those discussed in Chapter 3 (pp. 75–77), in which an enzyme may either be functional or nonfunctional. The inheritance of these traits follows the patterns described in Chapter 2, which you may want to review at this time. In particular, be sure that you understand the meaning of dominant and recessive alleles and of homozygous and heterozygous genotypes. Many other traits, as we saw in Chapter 3, have a more complex genetic basis. In this section we examine how biological variation is described.

Continuous and discontinuous variation within populations

Many human traits vary over a range of values, with all intermediate values being possible; such variation is called continuous variation.
Continuously variable traits, such as height, can often be measured in an individual and expressed as a numerical value. Other traits that vary continuously, such as hair curliness or skin color, are seldom expressed numerically, although theoretically they could be.

Continuous variation can result from the cumulative effects of multiple genes, each of which by itself contributes a small effect. Dozens of known genes, perhaps even hundreds, influence height in one direction or another. If we make the simplifying assumption that these effects are independent of one another and that they add up, we can predict that a population of individuals will show a variation in height similar to the bell-shaped curve (normal distribution) of Figure 7.1. When we measure heights in any large population, we do in fact get a curve that closely matches this predicted curve. Many other continuous traits vary in much the same way as height. For most of these traits, a strong environmental component also exists. Height, for example, is strongly influenced by childhood nutrition as well as by genes. Environmental components of traits also contribute to the formation of a bell-shaped curve.

A numerical description of continuous variation in a population requires the use of statistical concepts such as average (mean) values. The average values are characteristic of the population as a whole, not of any individual member within the group. For a particular group of people, we can calculate an average height, weight, or head breadth, but these averages are just statistical abstractions—there are perfectly normal individuals that differ from the average, perhaps even greatly, as can be seen in Figure 7.1. Thus, the group average for a continuously variable trait tells us little about any individual. Also, whereas height can actually be measured (and average height computed), concepts such as ‘tall’ are relative: a height that is average in England may be considered tall in India or the Philippines.

Your individual traits result from both the genes that you inherited from your parents and the environmental factors to which you are exposed. What you inherit from your parents is a predisposition for a range of possible future variations in phenotype. For example, when a child is born, its exact height as an adult cannot be predicted, but if the mother and father are both significantly taller than average, it will, if it receives adequate nutrition, probably also be taller than average.

**Discontinuous variation** within a population is represented by traits that are either present or absent, with no intermediate values possible. Most of these traits have a simple genetic basis, so that someone’s genotype may sometimes be deduced from their phenotypes and the phenotypes of their close relatives. Traits that vary discontinuously include blood groups and the presence or absence of conditions such as albinism or Tay–Sachs disease (see Chapter 3). A particular
phenotype for such a trait is either present or not in a particular individual and is generally not altered by environmental influences.

To describe discontinuous variation in a population, we divide the number of people who have a particular phenotype by the total size of the population; the resulting fraction is the frequency of that phenotype. From these phenotypic frequencies, scientists can calculate the frequencies of the alleles responsible. These **allele frequencies** (originally called gene frequencies) are most easily studied for traits whose patterns of inheritance are known and simple. Like the average values of continuously variable traits, allele frequencies are characteristic of entire populations, not of individuals. All individuals have genotypes, but only populations can have allele frequencies.

**Variation between populations**

The study of genetic variation both within and between populations is called **population genetics**, and it includes the study of allele frequencies for discontinuous traits. The measuring of allele frequencies requires that the different genotypes, and the alleles responsible for them, can readily be distinguished from one another. It is for this reason that population geneticists often concentrate on those genes whose phenotypic effects are easy to tell apart. Most of those genes control discontinuously variable traits that are either present or not. Differences in the average values for traits that vary continuously are also of interest to population geneticists, but the study of these traits is more difficult because the phenotypes of continuously variable traits are often altered by environmental influences such as nutrition.

One of the central tenets of modern biology is that evolution can occur only if populations are genetically varied. However, biologists did not always think in terms of evolving and variable populations. For over 2000 years, biologists believed that species were constant, unvarying entities. Plato and Aristotle had declared that each species was designed according to an ideal form that they called an *eidos*, often translated as ‘type’ or ‘archetype.’ Biologists following this view developed the **morphological species concept**. Each species was described as having certain fixed and invariant physical characteristics (morphology). The whole ‘type’ of that species was believed to be a cluster of ‘essential’ characteristics inherited as a single unit.

Biologists now recognize that species are constantly evolving, largely as the result of natural selection working on the genetic variation that is present within populations (Chapter 5). The Human Genome Project (Chapter 4) has revealed that over 99.9% of the human genome is identical in all people. However, the remaining fraction of a percent varies geographically, meaning that populations from different locations differ from one another. We must have some clear way to describe this variation and to describe population groups.

To define a population or a larger group of populations, we could sort people by some physical trait, such as distinguishing between people who are tall, short, or average in height. For any trait that we could choose, much of the variation exists within each and every population. If we chose some other physical trait, such as eye color or hair curliness, we would find that each physical characteristic results in a different...
grouping of the same people. In addition, we find that groupings based exclusively and strictly on any single trait always group together people who are quite dissimilar in many other respects (especially on a worldwide basis). For these reasons primarily, biologists prefer not to base the definition of population groups on physical characteristics.

Instead of using physical characteristics to define populations, biologists use the term **population** to refer to all members of a species who live in a given area and therefore can interbreed with one another. Membership in a population is determined by geographical location and by mating behavior, not by physical characteristics. Populations that interbreed under natural conditions belong to the same species (Chapter 5). All humans are placed in a single species, *Homo sapiens*, because all of them have the capacity to mate with one another and produce fertile offspring. However, people in different geographical locations belong to different populations. Genetic variation within any population is usually less than in the species as a whole. In past centuries, geographic isolation kept many human populations more distinct than they are now with worldwide transportation and migration. Population boundaries are not the same as national boundaries. Several different populations may live in the same geographic area, especially if cultural factors have maintained their separateness and inhibited matings between them. Sometimes, these populations are distinguishable by their derivation from geographically separate earlier populations.

Human populations in different places differ from one another in many physical traits. The average Canadian is taller than the average Southeast Asian, and the average African has darker skin than the average European. For natural selection, however, the characteristics that matter the most are those with the greatest impact on health and disease (or life and death). For example, cystic fibrosis and skin cancer are more frequent among people of European descent, but people of African descent have a higher risk of sickle-cell anemia and are more susceptible to frostbite if exposed to very cold temperatures. Most discontinuously variable traits that are examined closely show differences in allele frequency from one human population to another. For continuously variable traits, the difference between the averages of two populations is much less than the variation within either population (Figure 7.2). For example, the average height in the United States is taller than in China, but many Americans are shorter than the Chinese average and many Chinese are taller than the American average.

Although it is easy to find human populations that differ from one another in both physical features (morphology) and genetic traits, it is usually very difficult to find sharp boundary

**Figure 7.2**
Continuous variation in two populations with different mean values.

Distribution of height in two populations whose average values are 165 and 180 cm respectively. The variation within each population is greater than the difference between the average values of the two populations. Note that one of these populations is identical to the one shown in Figure 7.1.
lines dividing these populations from one another. If you were to walk from Asia to Europe and then to Africa, you would see populations differing only slightly, in most cases imperceptibly, from their neighbors, and you would meet representatives of the three largest population groups on Earth without finding any abrupt boundaries between them. Another way to say this is to say that variation between human populations is always continuous. This is so even when the trait in one individual is discontinuous. The population frequency of the allele responsible for the trait can vary continuously between zero (no one has the phenotype) and 100% (everyone has the phenotype). For discontinuous traits such as blood type, the allele frequencies of adjacent populations are generally close, just as is true for the average values of continuous traits, as seen in Figure 7.2.

Concepts of race

Humans have developed various ways of describing both themselves and the other human populations with which they have had contact. Biologists (who study all forms of life) and anthropologists (social scientists who study human populations and human cultures) have assisted in these descriptions by studying and measuring certain physical traits and allele frequencies. There are many ways in which human variation can be described, and there are many uses to which these descriptions have been put. One of the most problematic has been the attempt to separate people into different races. As we will soon see, there are various different meanings to this term, all of them different from the term ‘population.’ The term ‘population’ always describes smaller and more cohesive units than the term ‘race.’ No physical features are used in defining populations, but some race concepts have been based on physical features. In this section we describe four different concepts of race in the order in which they originated. The older concepts have not entirely died out; they have in many cases persisted side by side with the concepts that came later.

Races based on cultural characteristics. In the Bantu languages of Africa, the word for ‘people’ is bantu. Likewise, the Inuit word for ‘people’ is inuit. Every group of people has a name for itself and its members, and the name often means people or human. Names that people apply to other groups of people may simply be descriptive, but value judgments are often implied as well. In some instances, the value judgment implicit in the choice of name has been used to justify widespread abuses against the negatively labeled population. Such was the case when land and labor shortages resulted from large-scale cereal agriculture, a problem that arose independently in many places. A commonly developed solution to these shortages was to conquer neighboring people (the ‘other’) and confiscate their land. Slavery and several other systems of coercion were developed to secure the labor of conquered peoples. Slavery, oppression, and conquest all call upon the victorious people to practice certain atrocities on others that they would never tolerate within their own group. To justify these atrocities to themselves, and to protect their own members from practicing similar atrocities on one another, just about every conquering group has found it expedient to distinguish themselves from the ‘other,’ and furthermore to depict the conquered
There is biological variation both within and between human populations. People as somehow inferior, subhuman, or deserving of their fate. Many of the groups that were culturally defined as races in the past are really language groups, cultural groups, or national groups that are hardly distinguishable on any biological basis from the group that traditionally oppressed them.

The imposition of social inequalities between ‘Us’ and ‘Them’ is now recognized as racism. Racism has many meanings, but all of them include the belief that some groups of people are better than others, and that it is somehow justified or proper for the more powerful group to subdue and oppress the less powerful. In most cases, the motivation to conquer and oppress others came first; the racist ideology came later.

The ‘races’ identified by the conquering group are socially constructed to serve the interests of the oppressors only. The distinctions and values of the oppressors are forcibly imposed on the oppressed, who are often taught to believe in their own inferiority. Most people now regard racism as unethical because it denies basic rights to many people and because it results in frequent crime, violence, and social conflict.

Separation based on race serves better the political and economic causes that have engendered it if the distinctions recognized are declared to be ‘natural’ and unchangeable, as opposed to characteristics that can easily be changed by education or religious conversion. Scientists belonging to racist societies have therefore sometimes attempted to ‘prove’ that the traits characteristic of another race have an inherited basis that cannot easily be changed, an assertion called biological or genetic determinism (or hereditarianism). Behind such assertions is the view that a group identity (an ‘essence’ or Platonic eidos) can be inherited, a view for which there is no basis in genetics. Anthropologist Eugenia Shanklin documents several instances in which scientists conducted ‘scientific’ studies to help ‘prove’ the values and prejudices of their own social group. In their genocidal campaigns of the 1940s, the Nazis exterminated many millions of Jews, gypsies, Slavs, and other groups, but not until they had declared each of them to be an inferior ‘race.’

Racism and hereditarianism are not synonymous, but they often go together as attitudes shared by many of the same people. The supporters of eugenics (see Chapter 3) had many followers, including Nazis in Germany and anti-immigrationists in the United States. These followers sought ways to prove the inferiority, and especially the biologically unchangeable inferiority, of other people.

The other race concepts that we discuss later differ from this earliest concept, and resemble one another, in their avoidance of language, customs, and other cultural traits in the delineation of races. However, racism is not confined to those societies that embrace the cultural concept of race. Many biologists and anthropologists have pointed out that racism is also built into the the next concept, race delineated by body features.

The morphological or typological race concept. Biologists who study plant and animal species often describe the geographical variation within a species by subdividing the larger species into smaller and more compact subgroups, each of which is less variable than the species as a whole. These subgroups are generally called subspecies, but within our own species they are called races. To bring the study of human variation more in conformity with that of other species, scientists began to restrict
their attention to characters that could be studied biologically and to exclude personality traits, languages, religions, and customs more influenced by culture than by biology.

Before the days of ocean-going vessels, most of the world’s people had only a limited awareness of human variation on a worldwide scale. Each population, of course, knew about other populations nearby, but in most cases adjacent populations differed only slightly from one another. When trade extended over great distances, it usually did so in stages, so that none of the traders ever had to go more than a few hundred miles from home. The trade routes were also in most cases traditional, meaning that traders and migrants had generally come and gone over the same routes for centuries. This contributed to a gene flow or mixing of alleles that lessened the degree of difference between populations that would be noticed along the trade routes.

When explorers began to sail directly to other continents, they found people in other lands who differed more sharply from themselves in physical features. Many scientists subsequently became curious about the origin of these physical differences. Discussions of racial origins from about 1750 to 1940 tended to dwell on the origin of physical differences. A morphological definition of each race, based on physical features (morphology), was an outgrowth of the same thinking that had earlier resulted in a morphological species concept. At least initially, the major founders of this tradition were scientists who had no interest in oppressing the newly discovered peoples, so finding an excuse for racial oppression was less of a motive than was scientific curiosity. The emphasis was no longer on distinguishing only ‘Us’ from ‘Them,’ but on distinguishing among many different racial groups.

By the 1700s, biologists were actively describing and categorizing the variation in all living species. The eighteenth century naturalist Linnaeus (Carl von Linné) divided the biological world into kingdoms, classes, orders, genera, and species (see Chapter 6). He also divided humans into four subspecies: white Europeans, yellow Asians, black Africans, and red (native) Americans. The use of physical features such as skin color and hair texture to define subspecies was common among biologists using a morphological race concept. Other scientists in this same tradition recognized more races or fewer, but each race was always described on the basis of morphological characteristics such as skin color, hair color, curly or straight hair, and the occurrence of epicanthic folds of skin over the eyes.

Under the morphological concept of race, each race was defined by listing its common physical features as though they were invariant. For example, when describing a feature such as color, only one color was given, as if this color were invariant throughout the group and throughout time. This approach, which classified races on the basis of ‘typical’ or ‘ideal’ characteristics, ignoring variation, is called typology. Morphological definitions of race were always typological. Africans, for example, were declared to have black skins and curly hair, overlooking the fact that both skin color and hair form vary considerably from place to place within Africa and even within many African populations. All of the morphological characteristics were assumed to be inherited as a whole; a person was assumed to inherit a Platonic *eidos* (a ‘type’ or ‘essence’) for whiteness or redness, not just a white or red skin. Supporters of the typological concept of races were also supporters of a typological concept of species.
There is biological variation both within and between human populations. Years after morphological races had been defined, closer scrutiny revealed both variation within the morphological races and intergradation between them across their common boundaries. A few Europeans tried to save the morphological definitions by proposing that each race had originally been ‘pure’ and invariant, and that present-day variation within any population was the result of mixture with other races. One zoologist, Johann Blumenbach (1752–1840), divided up humans into American, Ethiopian, Caucasian, Mongolian, and Malayan races. He thought that each of these races was originally homogeneous (that is, ‘pure’), and he named each after the place that he identified as its ancestral homeland. For example, white-skinned people are called ‘Caucasian’ because Blumenbach thought that this race originated in the Caucasus Mountains, east of the Black Sea.

There is no scientific support nowadays for the concept of originally pure races or for the concept of different ancestral centers of origin of different races; human populations have never been homogeneous and have always been quite variable. In some cases, however, Europeans and others who feared for the ‘purity’ of their own group sought to pass laws limiting contacts, especially sexual contacts, between the races that they recognized. Most of these laws were brutal but still ineffective in stopping what were viewed as interracial matings. There is no scientific basis for the belief that such matings are in any way harmful. On the contrary, variation within any species confers a long-term evolutionary advantage because it provides the raw material that natural selection can use to adjust to changing environmental conditions.

But hereditarian assumptions were even more strongly embedded in the morphological race concepts than they are in the culturally based race concepts. Lest one think that science has long since banished such attitudes in educated people, it is only necessary to point to the great storm of controversy that flourished over the subject of race and IQ in the 1970s. Arthur Jensen attempted to convince his readers that the mental abilities of African Americans were below those of other races and that these differences were fixed by heredity and unchangeable by educational means. A number of scientists, including Leon Kamin, Richard C. Lewontin, and Stephen Jay Gould, showed that his claims were unsupported and based on fallacies and fabricated evidence. As recently as 1994, a book by Richard Herrnstein and Charles Murray once again brought up many of the same hereditarian arguments that had earlier been debunked (Box 7.1).

One of the strange ironies of a racist past is that many attempts at remediation, such as affirmative action, continue to require, at least for a time, the identification and naming of the same groups that were used previously for racially divisive purposes. Attempts to ensure fair and nondiscriminatory treatment for members of different socially recognized racial groups (in housing, employment, schooling, and so forth) require that we first identify and study the groups that we wish to compare. In this way, societies trying to overcome a history of racism find themselves using the very racial classifications of their racist past in order to redress the injustices of past generations.

Population genetics, clines, and race. Modern studies of human variation are based in large measure on genetics. Genetic variation between
BOX 7.1 Is Intelligence Heritable?

To address a question such as this, we must first define intelligence. Intelligence is not easily defined, but it includes the ability to reason and to learn new ideas and new forms of behavior, the measurement of which is far from simple. The biological bases for these abilities are likely to be multifaceted (Chapter 13), and genetic factors are likely to be the result of the interaction of many, many genes. Most discussions on the inheritance of human intelligence deal only with a single measure of this very complex trait, the IQ score, obtained from a test. IQ is not the same thing as intelligence and is at best an imperfect measure of mental abilities.

Also, to address this question, we must define the word ‘heritable.’ Heritability is defined in statistical terms as the proportion of the population's variation in some trait associated with genetic as opposed to environmental variation. Statistical association, or correlation, does not imply causation, and it certainly cannot be used to justify the claim that ‘there is a gene for’ the trait in question. One way to determine heritability of a trait in a domesticated species is to compare the variability of that trait in the population at large with the variability of the trait among highly inbred, genetically uniform individuals. Another way to determine heritability is to compare the variability that a trait exhibits at large with the variability of that trait among individuals raised in a standardized, experimentally controlled environment. Neither of these methods can be applied to humans, and the measures that are used to study humans are all indirect, complicated, and subject to criticism on technical grounds. For these reasons, there is no agreement on the heritability of any important human ability, including ‘intelligence.’

Numerous studies on IQ scores have shown the following:

- It is difficult to devise IQ tests that are free from cultural bias and from bias based on the language of the test, the gender and race of the test subjects, and the circumstances in which the test is administered.
- IQ scores seem to have both genetic and non-genetic components. Children’s IQ scores correlate strongly with those of their parents. The IQ scores of adopted children usually agree more closely with their adoptive parents than with their birth parents, although studies on adopted children have been criticized for a variety of reasons (see Chapter 3, pp. 71–72).
- IQ scores can be greatly improved by environmental enrichment. They can also be adversely affected by poor nutrition, poor prenatal conditions, and a number of other environmental circumstances.
- Populations historically subject to discrimination, such as African Americans in the United States, Maoris in New Zealand, and Buraku-Min in Japan, have average IQ scores about 15 points below those of the surrounding majority populations. However, these lower average scores do not always persist in people who migrate elsewhere: descendants of Buraku-Min living in the United States have, on average, IQ scores on a par with those of other people of Japanese descent.
- In the United States, IQ scores of whites and also of blacks (African Americans) vary from state to state, in some cases more than the average 15-point difference between blacks and whites. Among African Americans born in the South but now living in the North, IQ scores vary in proportion to the number of years spent in northern school systems.
- Transracial adoption studies show that African American children adopted at birth and raised by white families had IQ scores close to (in fact, slightly higher than) the white average.
- Careful studies of matched samples in schools in Philadelphia failed to show significant average differences in IQ scores between black and white schoolchildren if differences in background were controlled. ‘Matched samples’ mean that children in the study were compared only with other children of comparable age, gender, family income level, parents’ occupation, and similar variables.

Taken together, these data indicate that there is, at most, a small degree of heritability for IQ. They provide little support for the hereditarian claim that IQ is fixed and immutable, or that observed differences in scores cannot be diminished. They provide no support whatever for predicting any individual’s IQ score on the basis of their inclusion in any group.
populations is continuous, and all boundaries between groups of populations are arbitrary. Even for traits that vary discontinuously and for which an allele frequency can be calculated for each population, geographic variation in allele frequencies is continuous between populations.

A continuous increase or decrease in the average value, allele frequency or phenotypic frequency of any one trait is called a cline, after a Greek word meaning ‘slope’ (as in words like ‘incline’ or ‘recline’). Clines are an accurate (but lengthy) way of describing the geographic variation in each trait, one trait at a time, and each cline could be shown on a map. For a dozen characteristics, a dozen different maps would be needed, because the patterns of variation would in general not coincide.

The maps in Figure 7.3 show the clinal variation in the allele frequencies of three blood group alleles. Before such maps of allele frequencies can be drawn, local populations must first be identified and sampled. For example, blood groups must first be studied in many local populations; then the allele frequencies found in each geographic area can be drawn on maps such as those in Figure 7.3. From maps such as these, we learn that large continental areas usually show gradual clines. Thus, Figure 7.3A shows a gradual south-to-north increase in the frequency of allele A across North America, and Figure 7.3B shows a gradual west-to-east increase in the frequency of allele B across most of Eurasia. In geographic variation, clines of this sort are gradual, and boundaries of population groups are therefore arbitrary. Abrupt changes are uncommon, and when they do occur they generally coincide with geographic barriers that hinder both migration and gene flow. Examples of such barriers include the Sahara Desert, the Himalaya Mountains, and the Timor Sea north of Australia.

As can be seen in Figure 7.3, the frequencies of the blood group alleles A, B, and O vary greatly from one human population to another. The variations, however, do not necessarily coincide with other traits or with the groups recognized on the basis of morphology. Allele B, for example, reaches its highest frequency on mainland Asia, but is nearly absent from Native American populations or among Australian Aborigines. The frequency of allele A decreases from west to east across Asia and Europe. In Native American populations, allele A occurs mostly in Canada, and is mostly absent from indigenous Central or South American populations. The allele for blood group O has a frequency of 50% or more in most human populations, but its frequency approaches 100% in Native American populations south of the United States. African populations generally have all three of the alleles for ABO blood groups at levels close to worldwide averages.

Since the clinal variation concept was introduced in 1939, it has become customary to describe human variation by drawing maps of one cline after another. In addition to cline maps of phenotypes and allele frequencies, the techniques of molecular genetics (such as the DNA marker techniques described in Chapter 3, p. 72) are now being used to study clines at the molecular level. Clinal maps can also be drawn for continuous traits, in which case average values for the trait are calculated in each population. To describe the geographic variation in Homo sapiens or any other species, we could draw one map showing clinal variation in average body height, another showing variation in skin color or hair form, and so on. The population genetics approach encourages the scientific study and description of populations, including studies on the origins and former migrations of populations.
After the Holocaust (1933–1945), the fledgling United Nations felt the need to refute many Nazi claims about race. The result was the 1948 *Statement on Race and Racism*, written by a committee that included several prominent anthropologists and geneticists. The statement, which has been revised several times since 1948, correctly pointed out that nations,
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language groups, and religions have nothing to do with race, and that no group of people can claim any sort of superiority over another. The statement went further, however, to proclaim a new definition of race that replaced older, morphological definitions based on the inheritance of Platonist ‘ideal types’ with a new definition based on population genetics.

Under the population genetics definition, a race is a geographic subdivision of a species distinguished from others by the allele frequencies of a number of genes. A race could also be defined as a coherent group of populations possessing less genetic variation than the species as a whole. Either definition means that blood group frequencies are now considered more important than skin color in describing race, and that races are groups of similar populations whose boundaries are poorly defined. It also means that one cannot assign an individual to a race without first knowing what interbreeding population that individual belongs to. ‘Race’ is no longer a characteristic feature of any individual, because allele frequencies, like average phenotype values, characterize populations only, not individuals. Allele frequencies are consequences of population membership; they cannot be used to assign someone to a particular population or group. For this reason, racially discriminatory laws cannot and do not use population genetics; such laws rely invariably on the older morphological definitions or the still older social definitions of race.

Some writers maintain that racism is still contained in the population genetics race concept. They contend that studies that describe allele frequencies in geographic populations are merely reinscribing the racism of earlier concepts. Although far fewer people see racism in population genetics than in the earlier race concepts, some wish to go even further than the U.N. statement goes.

The ‘no races’ concept. Some scientists went still further in rejecting the heritage of the racist past: in the 1960s, led by the British anthropologist M.F. Ashley Montagu (who had earlier contributed to the U.N. definition), they declared that they would not recognize races at all. Among their arguments, one of the most compelling is that race concepts have always been misused by racists of the past and that the only way to rid the world of racism was to reject the entire concept of race. History is replete with examples of slavery, apartheid, discrimination, genocide, and warfare between racial groups. It is therefore easy to argue that the naming of races has in past generations done far more harm than good.

One stimulus to the ‘no races’ approach arises from the realization that there are no unique alleles or other genetic markers that could identify a person’s race. Races, like populations, differ in the frequency of various alleles but do not have alleles that belong exclusively to that group. Even differences in allele frequencies have become less pronounced as a result of the great increase in international travel and migration that has occurred especially since World War II. To a certain extent, human populations have always mated with one another whenever there has been geographic contact between them; this is one reason why human population groups do not differ more than they do and why neighboring populations are so often similar. Since the advent of the jet age, frequent migrations have allowed more extensive contact and more opportunities for mating between people of different genetic backgrounds than ever existed before. Such matings have always occurred and always will; they even occur in societies that have tried to outlaw them. This type of mating will slowly but inevitably diminish the differences in the mix of alleles (the
gene pools) of populations, making it progressively more difficult to identify any significant differences between populations.

The study of human variation

All studies of human variation run the risk of being misused or misinterpreted by racists. Nevertheless, there are many good reasons for studying human variation, and this study serves as the basis for the entire field of ‘human factors engineering.’ To take a simple example, the design of a passenger compartment (for automobiles, aircraft, etc.) must accommodate a certain range in the size, sitting height, arm length, and other dimensions of its possible occupants. These and other accommodations must take into account the total range of human variation, including all races and both sexes. In airline cockpits and similar enclosures, controls should be both visible and reachable by persons of different sizes. Moreover, these features are often matters of safety as well as comfort. Vehicle seat belts and airbags, sports equipment, surgical equipment, wheelchairs and similar aids, boots, helmets, kitchen counters, telephone receivers, gas masks, toilets, and doorways all need to accommodate the range of dimensions of the human body. Variation in other human characteristics (breathing rates, sweating) must also be considered in the design of space suits, diving equipment, respiratory equipment for fire fighting, or protective clothing for other situations. Most of the variation relevant for human factors engineering is found within each population group, including variation by age and sex; variation between human populations is generally minor by comparison.

A further reason for studying genetic variation among human populations is that it can help us to understand evolution. Population genetics has helped us to recognize geographic patterns of disease resulting from natural selection acting on human populations. Studies of this kind can also help us to reconstruct the past history of particular human populations, or of the human species as a whole. In succeeding sections of this chapter we examine some of these studies.

THOUGHT QUESTIONS

1. Twentieth-century approaches to the description of human variation have in large measure been revolts against the earlier approaches. Against which of these earlier approaches was the ‘no races’ approach primarily directed? Against which earlier approach was the population genetics approach directed?

2. African Americans more often have high blood pressure and more often die from their first heart attacks than do white Americans. How would you decide whether this is the result of a difference in genes, in diets, in the availability of medical care, or in the lasting effects of discrimination in U.S. society? If people in rural Africa seldom have heart attacks or high blood pressure, what possible hypotheses are falsified?

3. To produce research results of the kind referred to in Thought Question 2, one must have a way of assigning an individual to a population group. How does one determine a person’s membership in a biological population? Is it sufficient to know that they live in a particular place? Will asking people to name the racial or ethnic group in which they claim membership (self-identification) produce biologically meaningful results?
Population Genetics Can Help Us to Understand Human Variation

The geographic variation shown in Figure 7.3 deals with human blood groups. We know a lot about the genetic basis of blood groups, and a person’s blood group is easily determined, making blood groups good candidates for study by population geneticists. We now look in more detail at human blood groups and what their study has taught us about our own species.

Human blood groups and geography

In the days before reliable blood banks, blood transfusions were much riskier than they are today. Soldiers wounded in battle were generally treated in the field. If a transfusion was needed, it was done directly from the blood donor to a patient lying on an adjacent stretcher. Some transfusions were successful, but others resulted in death of the patient. Studies on the reasons for these different outcomes led to our knowledge of the existence of blood groups.

ABO blood groups. During the Crimean war (1854–1856), a British army surgeon kept careful records of which transfusions succeeded and which did not. From his notes he was able to identify several types of soldiers, including two types that he called A and B. Transfusions from type A to type A were nearly always successful, as were transfusions from type B to type B, but transfusions from A to B or B to A were always fatal. Also discovered at this time was a third blood type, O, which was initially called ‘universal donor’ because people with this blood type could give transfusions to anyone. These results were put to immediate practical use in treating battlefield injuries.

Karl Landsteiner, an Austrian pathologist who migrated to the United States, discovered the reason for these distinctions. Persons with blood type A make a carbohydrate of type A, which appears on the surfaces of their blood cells. Persons with blood type B make a carbohydrate of type B; persons with type AB make both type A and B carbohydrates; and persons with blood type O make neither of these carbohydrates. The A and B carbohydrates are also called antigens because they are capable of being recognized by the immune system (see Chapter 15). The immune system of each individual also makes antibodies against the blood group antigens that their own body does not make. In a person receiving a transfusion with incorrectly matched blood, these antibodies bind to the type A or B antigens, causing the blood cells to clump together within the blood vessels (Figure 7.4), often with fatal results. For explaining these immune reactions, Landsteiner received the Nobel Prize in 1930.

The A and B antigens allow all people to be classified into the four blood groups A, B, AB, and O. These blood groups are controlled by a gene that has three alleles: allele A is dominant and it contains information for producing antigen A (its phenotype); allele B is dominant and it contains information for producing antigen B (its phenotype); allele o is recessive and it functions as a ‘place-holder’ on the DNA but produces neither functional antigen. The AA and Ao genotypes both produce antigen A
and are therefore assigned to blood group A. Likewise, both $BB$ and $Bo$ genotypes produce antigen B and result in the B blood type. Genotype $oo$ produces neither A nor B antigens, which results in the O blood type (universal donor). Finally, genotype $AB$ allows both alleles $A$ and $B$ to produce their respective antigens, resulting in the AB blood type. When they occur together, the $A$ and $B$ alleles are said to be codominant because the heterozygote shows both phenotypes.

For the purpose of matching blood donors and recipients, any person who shares your blood type is a good donor. It is therefore possible to collect blood in advance from many donors, sort the blood by blood type, and store it under refrigeration for use in an emergency. It is ironic that the doctor who developed this concept, an African American named Charles Drew (1904–1950), was denied its full benefits because many hospitals at the time kept separate blood banks for whites and nonwhite patients, a practice that has no biological foundation. Because the chemical composition of the allele products does not vary, type A antigen from an African American is identical to type A antigen from a Native American or from anyone else. A person with blood type A is therefore a good donor for almost any other person with blood type A.

Other human blood groups. Karl Landsteiner also discovered several other blood group systems that are totally independent of ABO. One such system, called the Rh system, actually has three genes located very close together on the same chromosome: the first gene has alleles $C$ and $c$, the second has alleles $D$ and $d$, and the third has alleles $E$ and $e$. Unlike the ABO system, in which alleles are codominant, $c$, $d$, and $e$ are recessive to $C$, $D$, and $E$. In all, there are eight phenotypic possibilities, of which phenotype cde (genotype $ccddee$, homozygous recessive for all three genes) is sometimes called Rh-negative and the others Rh-positive. The CDe phenotype is the most frequent phenotype in most populations, except in Africa south of the Sahara, where cDe predominates. The Rh-negative

---

**Figure 7.4**
The human ABO blood groups. If a person of blood type $A$, who makes antibodies against blood type $B$, receives a transfusion of type $B$ or $AB$ blood, those antibodies cause the donated blood cells to clump together. Transfusion with matched blood or with blood type $O$ (no A or B antigens) does not cause clumping.

<table>
<thead>
<tr>
<th>blood type</th>
<th>genotype</th>
<th>antigen</th>
<th>antibodies made</th>
<th>recipient</th>
<th>donor</th>
</tr>
</thead>
<tbody>
<tr>
<td>$A$</td>
<td>$AA$ or $Ao$</td>
<td>$A$</td>
<td>anti-$B$</td>
<td>$A$</td>
<td>$A$</td>
</tr>
<tr>
<td>$B$</td>
<td>$BB$ or $Bo$</td>
<td>$B$</td>
<td>anti-$A$</td>
<td>$B$</td>
<td>$B$</td>
</tr>
<tr>
<td>$AB$</td>
<td>$AB$</td>
<td>$A + B$</td>
<td>neither anti-$A$ nor anti-$B$</td>
<td>$AB$</td>
<td>$AB$</td>
</tr>
<tr>
<td>$O$</td>
<td>$oo$</td>
<td>neither</td>
<td>both anti-$A$ and anti-$B$</td>
<td>$O$</td>
<td>$O$</td>
</tr>
</tbody>
</table>

The table above shows the compatibility of blood types for transfusions. The $A$ and $B$ columns represent the donor, while the $A$, $B$, $AB$, and $O$ columns represent the recipient. The universal donor ($O$) can receive any type of blood, while the universal recipient ($AB$) can donate to any type of blood.
phenotype cde is the second most common Rh phenotype in Europe and Africa, but is rare elsewhere.

Problems arise when a mother with the cde Rh-negative phenotype is pregnant with a baby who has a dominant \( C \) or \( D \) or \( E \) allele and is therefore Rh-positive. In this case, the mother makes antibodies against the \( C \), \( D \), or \( E \) antigens on the baby’s blood cells, especially in response to the tearing of blood vessels during the process of birth. Because these antibodies are made at the end of pregnancy, they usually don’t affect the first Rh-positive fetus that the mother carries. However, once these antibodies have been made, the mother’s immune system attacks any subsequent pregnancy with an Rh-positive fetus, destroying many of the fetus’ immature red blood cells, which can cause the death of the fetus (Figure 7.5). This problem can now be prevented by giving the Rh-negative mother gamma globulin (e.g., RhoGAM) at the time of the birth of any Rh-positive child; the globulin inhibits the formation of antibodies against Rh antigens, thereby protecting future pregnancies.

Separate from the ABO and Rh blood group systems are an MN system (with \( M \) most frequent among Native Americans and \( N \) among Australian Aborigines), a Duffy blood group system (with alleles \( Fy^a \), \( Fy^b \), and \( Fy^c \)), and many others.

**Geographic variation in blood group frequencies.** We saw earlier that the alleles for the ABO blood groups vary in frequency in different geographic locations (see Figure 7.3). Table 7.1 shows how the major geographic subgroups of *Homo sapiens* differ in the frequencies of various blood groups and other genetic traits. It is important to remember that

---

**Figure 7.5**

Rh incompatibility arising in an Rh-negative mother pregnant with an Rh-positive child.

---

FIRST Rh⁺ PREGNANCY

When an Rh-negative mother has her first Rh-positive pregnancy, \( C \), \( D \), or \( E \) antigens from the baby enter the mother’s circulation during the detachment of the placenta following birth.

SOON AFTER BIRTH

The mother’s immune system soon makes antibodies against Rh antigens \( C \), \( D \), or \( E \).

SECOND Rh⁺ PREGNANCY

Antibodies made after the first pregnancy can endanger any subsequent Rh-positive fetus unless protective measures are taken.
allele frequencies characterize populations only, not individuals. No blood group is unique to any population, so a person’s blood type cannot identify them as a member of any population.

Frequencies of blood group alleles also vary on a smaller geographic scale. This is especially true among rural people who remain in their native villages or districts all their lives. The geneticist Luigi Cavalli-Sforza has documented variation in the ABO, MN, and Rh blood group frequencies from one locality to another across rural Italy. Similar results have been observed in rural populations in the valleys of Wales, in African Americans from city to city across the United States, and among the castes and tribes of a single province in India. These studies emphasize the hazards of assigning all people in a single country to a single population, especially when cultural barriers discourage random mating. However, populations that have become more mobile experience less of this microgeographic variation. As stated earlier, these are variations in allele frequencies and therefore can not be used to establish clear-cut boundaries between populations.

### Isolated populations and genetic drift

In large, randomly mating populations in which selection and migration are not operating, the frequencies of the genotypes in the population tend to remain the same. This principle, which operates in all sexually reproducing species, is called the **Hardy–Weinberg principle**, and the predicted equilibrium is called the **Hardy–Weinberg equilibrium** (Box 7.2).

One of the criteria for a Hardy–Weinberg equilibrium is that the population be large. In small populations, allele frequencies tend to vary erratically, in unpredictable directions, from the expectations of the Hardy–Weinberg equilibrium. This phenomenon, called **genetic drift**, is

### Table 7.1

<table>
<thead>
<tr>
<th>Allele frequencies in the major geographic population groups.</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>AFRICAN POPULATIONS</strong></td>
</tr>
<tr>
<td>Frequencies of blood group alleles A, B, and O in the ABO system and M and N in the MN system close to world averages; Rh blood group system with allelic combination CDe most frequent and cde second; allele Fy most common in the Duffy blood group system; allele P1 more common than P2 in the P blood group system; hemoglobin alleles HbS and HbC more frequent than in most other populations.</td>
</tr>
<tr>
<td><strong>CAUCASIAN (EUROPEAN AND WEST ASIAN) POPULATIONS</strong></td>
</tr>
<tr>
<td>Allele A in the ABO blood group system somewhat more frequent than in African or Asian populations; frequencies of M and N in the MN system close to world averages; Rh blood group system with allelic combination CDe most frequent and cde second; Duffy blood group system with allelle Fya most frequent and Fyb second; alleles P1 and P2 both common in P blood group system; alleles for G6PD deficiency and thalassemia more frequent than in most other population groups.</td>
</tr>
<tr>
<td><strong>ASIAN POPULATIONS</strong></td>
</tr>
<tr>
<td>High frequencies of allele B (and correspondingly less of allele A) in the ABO blood group system; M and N alleles at frequencies close to world averages; Rh blood group system with allelic combination CDe most frequent and cde rare or absent; Fya especially common in the Duffy blood group system; allele P2 more common than P1 in the P blood group system; some populations with high frequencies of alleles for thalassemia.</td>
</tr>
<tr>
<td><strong>NATIVE AMERICAN (AMERINDIAN) POPULATIONS</strong></td>
</tr>
<tr>
<td>Very high frequencies of allele O and virtually no B in the ABO blood group system; very high frequencies of allele M in the MN system; Rh blood groups with allelic combination CDe most frequent and cde rare or absent; allele P1 more common than P2 in the P blood group system; high frequencies of Dr in the Diego blood group system.</td>
</tr>
<tr>
<td><strong>AUSTRALIAN AND PACIFIC ISLAND POPULATIONS</strong></td>
</tr>
<tr>
<td>Frequencies of blood group alleles A, B, and O in the ABO system close to world averages; high frequencies of allele N in the MN blood group system; Rh blood groups with allelic combination CDe most frequent and cde rare or absent.</td>
</tr>
</tbody>
</table>
Population Genetics Can Help Us to Understand Human Variation

defined as changes in allele frequencies in small to medium-sized populations due to chance alone.

The original model of genetic drift dealt with populations that remained small all the time, but other types of genetic drift were found to apply in particular situations. For example, if a large population became temporarily small and then large again, the random changes in allele frequencies that occurred when the population was small—the bottleneck—would be reflected in the allele frequencies of subsequent generations. This bottleneck effect is shown in Figure 7.6. Another type of genetic drift occurs if a small number of individuals become the founders of a new population. The allele frequencies in such a new population—whatever its subsequent size—will reflect the allele composition of this small group of founders, an influence known as the founder effect.

Several cases of genetic drift have been studied in isolated human populations. One well-studied example concerns the German Baptist Brethren, or Dunkers, a religious sect that originated in Germany during the Protestant Reformation. Forced to flee their native Germany, a few dozen Dunkers came to Pennsylvania in 1719 and started a colony that grew to several thousands and spread to Ohio, Indiana, and elsewhere. Because their strict religious code forbids marriage outside the group, they have remained a genetically distinct population.

Allele frequencies among the Dunkers have been influenced by genetic drift, particularly by the founder effect. If the Dunkers were a representative sample of seventeenth-century German populations, we would expect similar allele frequencies to those of present-day German populations derived from the same source. If, however, natural selection had changed the Dunker populations as the result of adaptations to their new location, then we would expect their allele frequencies to come closer to those of neighboring populations of rural Pennsylvania. Neither of these predictions is correct. Allele frequencies among the Dunkers differ from populations of both western Germany and rural Pennsylvania in a number of traits that have been studied. Blood group B, for example, hardly occurs at all among the Dunkers, although the frequency of the B allele is around 6–8% in most European-derived populations, including those of both Germany and Pennsylvania. Other genetically determined traits show similar patterns, including the nearly total absence of the \( Fy^a \) allele (from the Duffy blood group system) among Dunkers. The explanation that best agrees with the data is that the original founder population, known to have been made up of only a few dozen individuals, happened not to include anyone carrying \( Fy^a \) or...
the allele for blood group B. Additional alleles may have been lost by genetic drift while the population remained small. The result was a population that derived its allele frequencies from the assortment of alleles that happened to be present in the founders. We can test this assumption by looking for the rare Dunkers who do possess an allele such as Fya. In every case that has been investigated, the occurrence of such an allele among the Dunkers can be traced to a person who joined the group as a religious convert within the last few generations.

Because they are genetically isolated, except for occasional religious conversions, the Dunkers have kept a unique combination of unusual allele frequencies. In the absence of blood group B, they resemble Native American populations; in the absence of Fya, they resemble

---

**BOX 7.2 The Hardy–Weinberg Equilibrium**

The Hardy–Weinberg principle can be stated as follows:

In a large, randomly mating population characterized by no immigration, no emigration, no unbalanced mutation, and no differential survival or reproduction (that is, no selection), the frequencies of the alleles (genotypes) tend to remain the same.

Allele frequencies are fractions of the total number of alleles present. If a population of 500 individuals (or 1000 alleles at a single genetic locus) contains 400 alleles of type A and 600 alleles of type a, then we say that the frequencies of the two alleles are 0.40 and 0.60 respectively, or 40% and 60% of the total number of alleles in the gene pool. At a given locus, the allele frequencies always add up to 1, or 100% of the population's gene pool.

Under the conditions specified in the Hardy–Weinberg principle, as stated above, there is a simple equilibrium of unchanging allele frequencies. Let us consider the case of a gene locus that contains two alleles, A and a. If the frequency of allele A is called p and the frequency of allele a is called q (where p + q = 1), then the equilibrium frequencies of all three diploid genotypes is given by the Hardy–Weinberg formula:

<table>
<thead>
<tr>
<th>Genotypes</th>
<th>AA</th>
<th>Aa</th>
<th>aa</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequencies</td>
<td>p²</td>
<td>2pq</td>
<td>q²</td>
</tr>
</tbody>
</table>

This formula predicts that the frequency of the homozygous dominant genotype AA will be p², the frequency of the heterozygous genotype Aa will be 2pq, and the frequency of the homozygous recessive genotype aa will be q².

To show that these equilibrium frequencies remain stable over successive generations and do not tend to change in either direction, consider the production of gametes in a population already at equilibrium. All of the gametes produced by the dominant homozygotes AA carry allele A, so the frequency of A gametes from AA homozygotes is p². Half of the gametes produced by the heterozygotes Aa also carry allele A, so the frequency of A gametes from heterozygotes is half of 2pq, which equals pq. The total proportion of A gametes is thus p² + pq. We can now use simple algebra, separating out the common factor and then applying the equation p + q = 1 to calculate the frequency of A gametes:

Frequency of A gametes:

\[
p² + pq = p(p+q) = p(1) = p
\]

In similar fashion, the proportion of gametes carrying allele a is equal to pq (the other half of 2pq) from the heterozygotes plus q² from the recessive homozygotes aa.

Frequency of a gametes:

\[
pq + q² = (p + q)q = (1)q = q
\]

So the frequency of A and a gametes corresponds to the frequency of A and a alleles.
African populations. In most traits, however, their derivation from a European source population is evident. These findings show that population resemblances based on a single blood group or gene system may often be misleading, and that distinctions among human populations, if used at all, should be based on a multiplicity of genetic traits.

The bottleneck effect has been used as a hypothesis to explain the near-total absence of blood group B among Native Americans and of cde (in the Rh blood groups) among Pacific Islanders. When the ancestors of these people first migrated from Asia, the random changes in allele frequency that occurred when the groups were small gave rise to distinct, isolated populations whose allele frequencies differed from those of the ancestral populations. Genetic drift of this kind would apply primarily to

Combining the gametes in all possible combinations (to simulate random mating) produces the following results:

<table>
<thead>
<tr>
<th>Female gametes</th>
<th>Aa</th>
<th>Aa</th>
<th>Aa</th>
</tr>
</thead>
<tbody>
<tr>
<td>p</td>
<td>q</td>
<td>q</td>
<td>q</td>
</tr>
</tbody>
</table>

Genotypes

<table>
<thead>
<tr>
<th>Male gametes</th>
<th>Aa</th>
<th>Aa</th>
</tr>
</thead>
<tbody>
<tr>
<td>p</td>
<td>q</td>
<td>q</td>
</tr>
</tbody>
</table>

Frequencies

<table>
<thead>
<tr>
<th>Genotypes</th>
<th>AA</th>
<th>Aa</th>
<th>aa</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequencies</td>
<td>p²</td>
<td>2pq</td>
<td>q²</td>
</tr>
</tbody>
</table>

Taking the resulting genotypes from the chart above (and adding the two heterozygous combinations together), we obtain:

\[
p^2 + 2pq + q^2 = 1
\]

This is the same equation that we started with, which shows that the frequencies have not changed. It can also be shown that a population that does not start out at equilibrium will establish an equilibrium in a single generation of random mating.

Notice all the assumptions of the model: the population must be closed to both emigration and immigration, and there must be no unbalanced mutation and no selection. The population must be large enough to permit accurate statistical predictions, and the population members must mate at random. In reality, most natural populations are subject to mutation, selection, and nonrandom mating (including inbreeding), and most usually experience emigration and immigration as well.

The Hardy–Weinberg model, in other words, describes an idealized situation that is seldom realized in practice. The Hardy–Weinberg equilibrium is important to population genetics as an ideal situation with which real situations can be compared; if a population is not in Hardy–Weinberg equilibrium, one can ask why and then seek to measure the extent of the deviation from equilibrium. The same procedure is followed in other sciences as well. For example, ‘freely falling bodies without air resistance’ are an ideal situation in physics, and air resistance can be measured as a deviation from this ideal.

The Hardy–Weinberg equation is useful in estimating allele frequencies for traits controlled by a single gene. For example, if a population of 1000 has 960 individuals showing the dominant phenotype (such as normal pigmentation) and 40 displaying the recessive phenotype (such as albinism), then \(q^2\), the proportion of homozygous recessive individuals, is equal to 40/1000, or 0.04. From this, we can calculate \(q = \sqrt{0.04} = 0.2\).

From the fact that \(p + q = 1\), we can calculate \(p = 1 - q\). Substituting the value of 0.2 that we found for \(q\) gives us \(p = 1 - 0.2\) or \(p = 0.8\). Then the proportion of homozygous dominant individuals in the population is \(p^2 = (0.8)^2 = 0.64\) and the proportion of heterozygous individuals is \(2pq = 2(0.8)(0.2) = 0.32\).
groups of people, like the Polynesians or Native Americans, whose founder populations were initially small. The effects of genetic drift are minimal in the larger and more widespread population groups of Africa, Europe, and mainland Asia.

Reconstructing the history of human populations

Allele frequencies and DNA sequences in modern populations can be used as clues to their evolutionary origins. For example, American molecular biologist Rebecca Cann and her co-workers studied mitochondrial DNA sequences in samples from over 100 human populations. Mitochondria are organelles in the cytoplasm of eucaryotic cells (Chapter 6, p. 170) that produce much of the cell’s energy and that also contain small strands of DNA independent of the DNA in the nucleus. Mitochondrial DNA is transmitted only maternally, from mother to both male and female offspring. Sperm from the father contain almost no cytoplasm and do not transmit mitochondrial DNA. Because mitochondrial DNA is smaller than chromosomal DNA in the nucleus, it is easier to sequence and is thus ideal for tracing evolutionary patterns. On the basis of these DNA sequences, Cann and her colleagues proposed a family tree of human populations using a maximum-parsimony computer model: of all possible family trees, the one shown in Figure 7.7 requires fewer mutational changes to have occurred than for any other tree. Another research team, headed by Luigi Cavalli-Sforza, used alleles of 120 genes to study the genetic similarities among 42 populations representing all the world’s major population groups and many small ones as well. The findings of these two studies (and others) support the hypothesis of a divergence in the distant past between African and non-African populations, with the non-African populations later splitting into North Eurasian and Southeast Asian subgroups (see Figure 7.7). Australian Aborigines and Pacific Islanders are descended from the Southeast Asian subgroup, whereas Caucasians (Europeans, West Asians) and Native Americans (Amerind) are both descended from the North Eurasian group, which also includes Arctic peoples. The groups suggested by this study are geographically coherent and confirm certain well-documented patterns of migration. Existing linguistic evidence also matches these groupings, except for a few
cases of cultural borrowing, which can be documented historically. Cavalli-Sforza’s group estimates, largely on the basis of archaeological evidence, that the split between African and non-African populations took place 92,000 or more years ago. Other estimates have placed this split much earlier, back to the time of *Homo erectus*. The spread of human genes outward from Africa was either a very early event, or perhaps there were several such diffusions.

Studies such as those we have just described have sometimes been criticized for not being politically correct or for ‘reinscribing racism.’ A related criticism of the methodology is that geneticists with no training in anthropology are often tempted to lump together people who live close together even if there is good evidence that they have been historically and culturally separate. In other cases, people may maintain contact across considerable distances with other people who are culturally similar and speak the same language, and may consider themselves as belonging to the same group, even if population geneticists list them as separate because of the geographical distance between them. Although a good deal of interbreeding between groups always takes place, people more often choose their mates from what they consider as their own group. In order to assess what population groups actually exist (or existed historically), population geneticists need to cooperate with anthropologists familiar with the people being studied.

Paleontological and anthropological studies show that *Homo sapiens* has always been geographically widespread, with early populations spread across three continents, from Indonesia to Zambia and Western Europe. The earlier species *Homo erectus* was also geographically widespread. Despite this geographic spread, however, neighboring populations have always maintained genetic contact. Adaptation to local environments has caused populations to evolve geographic differences from one another, while matings between populations has maintained enough gene flow to prevent populations from becoming even more different. These two opposing tendencies form the basis for what American anthropologist Milford Wolpoff has called the multiregional model of the human species, which asserts that human populations have always maintained genetic contact with one another despite the differences resulting from local adaptation. The genetic contact maintains all human populations as one species, while the local adaptations have prevented geographic uniformity.

The study of allele frequencies has also been used to determine the origins of particular groups of people. One such study, for example, showed that Koreans are derived from a group that includes the Mongolians and Japanese but not the Chinese. Also, several studies have provided evidence for a Middle Eastern contribution (perhaps via Phoenecian sailors) to the populations of both Sicily and Sardinia. Studies of the Native Americans have shown that a minimum of three separate migrations were responsible for populating the Western Hemisphere, and more recent studies show that the situation is far more complex than this.

How did the adaptations come about that led to the various population differences in allele frequencies? The next two sections attempt to provide some of the answers to this question.
Malaria and Other Diseases Are Agents of Natural Selection

As any species evolves, biological differences among its populations arise largely through natural selection. Diseases are among the selective forces that can result in genetic differences among populations. In this section we consider some genetic traits that confer partial resistance to malaria. In malaria-ridden areas, natural selection acts to increase the frequency of alleles that confer partial resistance to malaria while decreasing the frequency of alleles that leave people susceptible to malaria. Many other selective forces have also operated over the course of human history, but resistance to malaria provides a series of well-studied examples.

New traits are produced by mutation (see Chapter 3, pp. 67–69) and are then subjected to natural selection, a process in which many traits die out in populations. The traits that survive natural selection are adaptive traits, or adaptations (Chapter 5), that is, traits that increase a population’s ability to persist successfully in a particular environment. A good deal of human variation consists of adaptations that have resulted from natural selection operating over time, disease being a significant agent of that selective process.

Malaria

On a worldwide basis, malaria causes over 110 million cases of illness each year and causes close to 2 million deaths, more than most other diseases. (Only malnutrition and tuberculosis cause more deaths each year, and measles causes about the same number.) Malaria also has a greater impact than most other diseases on the average human life expectancy because most of its victims are young, so that many more years of life are lost for each death that occurs. Malaria is more prevalent in tropical and subtropical regions than in temperate climates. The threat of malaria has largely been eliminated in the industrially developed countries through
mosquito eradication programs and the draining of swamps, but as late as the first half of the twentieth century, malaria claimed many thousands of victims in Florida, Louisiana, Mississippi, and Virginia.

Historical and anthropological evidence confirms that malaria was rare (and therefore not a significant selective force) before the invention of agriculture. Even today, the disease is rare in undisturbed forests or in hunting-and-gathering societies. The clearing of forests for agricultural use opens up more swampy areas, and the building of irrigation canals or drainage ditches creates additional pools of stagnant water. The mosquitoes that carry malaria breed best in stagnant water open to direct sunlight. Agriculture therefore did much to change, in unintended directions, the agents of death (and thus the selective pressures) that act on human populations.

**Life cycle of Plasmodium.** Malaria is caused by one-celled protozoan parasites belonging to the genus *Plasmodium* (kingdom Protista, phylum Sporozoa), which live in human blood and liver cells. Of the four species of *Plasmodium* that cause malaria, *Plasmodium falciparum* is the most virulent. All species of *Plasmodium* have a complex life cycle, spending different parts of their life cycle in two different host species, mosquitoes and humans. The *Plasmodium* sexual stages (male and female gametocytes) are intracellular parasites that inhabit human red blood cells. When a female mosquito of the genus *Anopheles* is ready to lay her eggs, she first takes a blood meal from a person during which she ingests large numbers of red blood cells. (Mosquitoes rarely bite otherwise.) If the red blood cells contain *Plasmodium*, the male and female gametocytes combine in the mosquito’s gut to form zygotes (fertilized eggs). The zygotes develop asexually through several stages within the mosquito, culminating in the infective forms (sporozoites), which migrate into the mosquito’s salivary glands (Figure 7.8).

The mosquito’s thin mouthparts function like a tiny soda straw or hypodermic needle. Shortly before consuming a blood meal, the female mosquito injects her saliva into her victim. The saliva contains anticoagulants that prevent the human blood from clotting inside the mosquito’s mouthparts. When the mosquito injects saliva into a new human host, any sporozoites present in her salivary glands are injected along with it. These sporozoites enter the human bloodstream and are taken up by the liver. Each parasite then develops into thousands more, which may remain in the liver for years. Some parasites periodically escape from the liver into the bloodstream and invade the red blood cells. The parasites reproduce asexually within the red blood cells, producing the disease symptoms. The parasites digest the cell’s oxygen-carrying hemoglobin molecules, and one stage also ruptures the red blood cells. Any impairment of the ability of the blood to carry oxygen to the body’s tissues is called an anemia; all anemias leave their victims run-down and weakened. In malaria, the anemia is caused by destruction of both the hemoglobin and the red blood cells. Cell rupture also brings on fevers, headache, muscular pains, and liver and kidney damage. Within a given host, the asexual cycle of *Plasmodium* continues again and again until the patient either recovers or dies. In the red cells, the parasites can also develop into the sexually reproducing gametocytes, which may be picked up by another mosquito in its next blood meal, spreading the disease.
Sickle-cell anemia and resistance to malaria

One of the symptoms of malaria is anemia. There are many other types of anemia. A very serious type was first discovered in 1910 by a Chicago physician named Charles Herrick. This strange and usually fatal disease also produced abnormally shaped red blood cells that sometimes resembled sickles. For this reason, Herrick called the disease **sickle-cell anemia**.

Figure 7.8
Life cycle of the malaria parasite *Plasmodium*.
A simple blood test was soon devised to test for the condition: a glass slide containing a bowl-shaped depression is used, and a drop of the patient’s blood is placed inside the depression. A ring of petroleum jelly is placed around the margins of the depression and a cover glass is then applied, forming an airtight seal. As the red blood cells use up the available oxygen in the depression, the oxygen level decreases. Under these conditions, the red blood cells of a person with sickle-cell anemia assume their characteristic sickle-like shape, while normal red blood cells retain a circular biconcave shape (Figure 7.9). This blood test also allows the recognition of heterozygous carriers, a small percentage of whose blood cells sickle while the rest remain round.

**Normal and abnormal hemoglobins.** Sickle-cell anemia is caused by an abnormality in the molecules (called hemoglobin) that carry oxygen within the red blood cells. The hemoglobin molecule consists of four protein chains (two each of two different proteins) surrounding a ringlike ‘heme’ portion. Suspended in the middle of this ring is an iron atom that can bind one oxygen molecule (O₂), giving the hemoglobin its ability to transport oxygen and also its red color.

A change in a single amino acid, number 6 in one of the protein chains, is responsible for sickle-cell anemia. Normal adult hemoglobin (hemoglobin A) has glutamic acid in this position in the chain, while sickle-cell hemoglobin (hemoglobin S) has valine instead. This minute change alters the shape of the hemoglobin S molecules, straining the ringlike heme part of the molecule so that hemoglobin S does not carry oxygen as well as hemoglobin A. When they are not carrying oxygen, hemoglobin S molecules are stickier than normal hemoglobin. When the oxygen concentration in the blood is low, such as during physical exertion, hemoglobin S molecules adhere to one another and also to the inside of the red blood cell membrane, deforming the cells into the characteristic sickled shape. The difference in the proteins is hereditary and is caused by an altered codon in the hemoglobin gene on the DNA.

![Figure 7.9](image)

Normal red blood cells and red blood cells from a patient with sickle-cell anemia.
The genetics of sickle-cell hemoglobin. Sickle-cell anemia is inherited as a simple Mendelian trait. People who die from sickle-cell anemia are always homozygous and their parents are almost always heterozygous, as are a certain number of siblings and other relatives. The gene for hemoglobin is designated Hb and the different alleles are designated by superscripts: \( Hb^A \) is the allele for normal hemoglobin and \( Hb^S \) is the allele for sickle-cell hemoglobin.

In U.S. and Caribbean populations, the vast majority of people carrying the \( Hb^S \) allele for sickle-cell hemoglobin are blacks of African ancestry. Tests of African populations also show high frequencies of the sickling allele, up to 25% in certain populations. In homozygous individuals (\( Hb^S Hb^S \)), all the red blood cells are sickled at low oxygen concentrations, as commonly occurs during heavy exertion. Heterozygous individuals (\( Hb^A Hb^S \)) have both types of hemoglobin and about one percent of their red blood cells can become sickled while the rest are normal in shape. Because both alleles produce a phenotypic result in heterozygotes, they are codominant, as we described earlier in connection with the AB blood type.

Symptoms of sickle-cell anemia. Most of the debilitating symptoms of the disease are consequences of the deformed, sickle-shaped cells brought on by exertion. The smallest blood vessels, capillaries, have a diameter only slightly larger than the diameter of blood cells. Because of their sickle shape and changed diameter, sickled cells cause flow resistance in the capillaries and thus impair microcirculation. In most of the body’s organs, impaired microcirculation further reduces oxygen levels (hypoxia), which results immediately in a severely painful sickle-cell crisis. These crises begin in infancy. Damaged cells collect in the capillaries of the joints and result in painful swelling. The sickled cells are also more easily disrupted and destroyed than the normal-shaped round ones, resulting in a decreased oxygen-carrying capacity (anemia). The anemia and impaired circulation results in tissue damage to many organs, eventually resulting in death (Figure 7.10). In African populations, the death of homozygous \( Hb^S Hb^S \) individuals often occurs before adulthood, but in the United States and the Caribbean, survival to reproductive age is now increasingly common. The reduction in red blood cell number and the sickle-cell crises also occur among heterozygotes, but not as severely.

Population genetics of sickle-cell anemia. When geneticists realized that sickle-cell anemia in the United States and Jamaica was largely confined to people of African descent, they began to investigate other populations. Using the blood test described earlier in this chapter, researchers investigated the frequency of the allele for hemoglobin S in many African and Eurasian populations. Over large parts of tropical Africa, researchers found remarkably high frequencies of the \( Hb^S \) allele, up to 25% or more. At first this appeared puzzling, because sickle-cell anemia was nearly always fatal before reproductive age. An allele whose effects are fatal in homozygous form should long ago have been eliminated by natural selection because people having sickle-cell children would have fewer children surviving to reproductive age.

Maps were made of the frequency of the sickle-cell allele. From these maps and from other evidence, it was noticed that the areas where the sickle-cell allele was frequent were also areas with a high incidence...
of malaria, particularly the variety caused by *Plasmodium falciparum* (Figure 7.11A and B).

Subsequent research confirmed the basic fact that the $Hb^S$ allele, even in heterozygous form, confers important resistance to the most virulent form of malaria. Tests in which volunteers were exposed to *Anopheles* mosquitoes showed that the mosquitoes are far less likely to bite heterozygous $Hb^A/Hb^S$ individuals than homozygous $Hb^A/Hb^A$ individuals. Tests with the *Plasmodium falciparum* parasites showed that they thrive on the red blood cells of $Hb^A/Hb^A$ individuals, who nearly always come down with a serious case of malaria after infection. However, when $Hb^A/Hb^S$ heterozygotes or $Hb^S/Hb^S$ individuals with sickle-cell anemia are infected with *Plasmodium falciparum*, their malaria symptoms are mild and they recover quickly because the parasite cannot complete its asexual cycle in their sickled blood cells. The protection that the $Hb^S$ allele affords against malaria is sufficient to explain its persistence in those populations in which the incidence of malaria is high.

Hemoglobin S thus decreases the fitness of homozygotes by causing sickle-cell disease, but it increases the fitness of heterozygotes in areas where malaria occurs. In this way, malaria acts as an instrument of natural selection and has a dramatic influence on the allele frequencies of populations.

In addition to hemoglobin A and hemoglobin S, several other genetic variants of hemoglobin have been discovered. Some of these, such as $Hb^C$, also occur principally in areas where malaria is present and are thought to confer some resistance to malaria.

*Figure 7.10* Development of the consequences of the $Hb^S$ mutation in the hemoglobin gene. A small change in a gene can have many phenotypic consequences.
Other genetic traits that protect against malaria

Sickle-cell anemia is not the only heterozygous condition that protects against malaria. Two others are thalassemia and G6PD deficiency.

**Thalassemia.** In many countries bordering the Mediterranean Sea (including Spain, Italy, Greece, North Africa, Turkey, Lebanon, Israel, and Cyprus), many people have suffered from a different debilitating type of anemia known as thalassemia, (meaning ‘sea blood’ in Greek). The disease also occurs further east, especially in Southeast Asian countries such as Laos and Thailand (Figure 7.11C). Thalassemia is marked by a reduced amount of one or more of the protein chains in the hemoglobin molecule. The disease exists in a more serious, often fatal, homozygous
form called thalassemia major and a less severe heterozygous form called thalassemia minor. Red blood cells containing nonfunctional hemoglobin are destroyed in the spleen, producing anemia.

The symptoms of thalassemia vary, but all forms result in some decrease in oxygen transport in the blood. The bone marrow compensates by overproducing red blood cells, and this overproduction robs the body of much-needed protein and results in stunted growth and smaller stature.

Populations in which thalassemia occurs can now be screened for the genotypes that cause the disease, and genetic counseling can be provided to those found to carry the trait. Screening programs and newer methods of treatment have greatly reduced the problems caused by this disease in Italy, Greece, and elsewhere in the Mediterranean.

The geographical distribution of thalassemia follows closely the distribution of malaria in countries where sickle-cell anemia is infrequent or absent. For this reason, it has long been suspected that thalassemia confers a protective resistance to malaria, similar to that caused by sickle-cell anemia. The evidence is indirect: if heterozygous individuals (those with thalassemia minor) did not have some selective advantage such as malaria resistance, then the deaths caused by thalassemia major would have caused the genes for this trait to die out long ago.

G6PD deficiency. Blood sugar (glucose) is normally broken down within each cell in a series of reactions that begin with the formation of glucose 6-phosphate. Most of the glucose 6-phosphate is broken down into pyruvate (see Chapter 10, pp. 349–350) in a series of energy-producing reactions, but some is also used to make ribose (the sugar used in RNA) and to make reducing agents such as NADPH and glutathione. The removal of two hydrogen atoms from the glucose 6-phosphate molecule requires the enzyme glucose 6-phosphate dehydrogenase (G6PD). There are many people who have too little of this enzyme, a condition known as G6PD deficiency, or favism. G6PD deficiency results from a mutation in the gene that encodes the G6PD enzyme.

Under many or most conditions, people with G6PD deficiency remain perfectly healthy, but they occasionally suffer from an anemia in which the red blood cells rupture, spilling their hemoglobin, which then becomes physiologically useless but easy to detect by simple lab tests. This type of anemia, which is potentially fatal, can occur in G6PD-deficient people as a reaction to certain drugs (aspirin, quinine, quinidine, chloroquine, chloramphenicol, sulfanilamide, and others), in response to certain illnesses, or after eating fava beans (Vicia faba), a common legume of the Eastern Mediterranean and Middle East. The anemia may also exist chronically in a nonfatal form in people with G6PD deficiency. G6PD deficiency has been shown to offer protection against P. falciparum malaria. It affects some 10 million people, and is thus the most common disorder offering protection against malaria. Most importantly, heterozygous carriers of the deficiency are also malaria-resistant, but the exact mechanism of the resistance has yet to be worked out.

G6PD deficiency occurs mostly in Mediterranean populations from Greece to Turkey and from Tunisia to the Middle East, and among Sephardic Jews. It also occurs south of this area into Africa and eastward across Iran and Pakistan to Southeast Asia and southern China (Figure 7.11D). The Greek mathematician Pythagoras may have suffered from
this disorder, for his aversion to beans (one of the triggers of anemia in G6PD-deficient people) has become legendary. Pythagoras founded a religious cult in which the avoidance of beans was an important belief. Opponents of his cult once captured Pythagoras by chasing him toward a bean field, which they knew he would not cross.

**Population genetics of malaria resistance**

**Polymorphism** is the term used to describe a condition in which two or more alleles of the same gene are known in a given population at frequencies higher than the mutation rate. (This last restriction means that the alleles were inherited and could not all simply be the result of new mutation.) Polymorphism is a characteristic of the population, not of individuals; an individual may bear only one, or at most two, of the many alleles present in the population. Some alleles of polymorphic genes have harmful effects when homozygous, but they persist in populations because the same alleles also confer some important benefit (such as malaria resistance) when heterozygous. If the polymorphism persists for many generations, it is likely to be a balanced polymorphism. Balanced polymorphism arises when the homozygous genotypes suffer from some selective disadvantage or reduction in fitness, while the heterozygotes have the maximum fitness. For example, in a country in which malaria is present, $Hb^A Hb^A$ homozygotes have lower fitness because they are susceptible to malaria, and most $Hb^S Hb^S$ homozygotes die young from sickle-cell anemia. The $Hb^A Hb^S$ heterozygotes have maximal fitness because they are malaria-resistant and because they have enough normal red blood cells for them not to suffer from fatal sickle-cell anemia. Under conditions like these, natural selection brings about and perpetuates a situation in which both alleles persist.

The selection by malaria for genetic traits that offer resistance to it is at least as old as the open, swampy conditions (ideal for the breeding of mosquitoes) brought about by agriculture in warm climates. Evidence for this exists in the form of human bones found at a Neolithic archaeological site along the coast of Israel. Cultural remains found at this site show that it was an early farming community, one of the first in the area. Pollen analysis shows the presence of many plants characteristic of swampy areas. Some of the bones show characteristic increases in porosity (due to the increased production of red blood cells in the bone marrow) indicative of thalassemia.

**Other diseases as selective factors**

Hereditary diseases that confer some advantage in the heterozygous state are not confined to those that protect against malaria. In European populations of past centuries, tuberculosis, an infection caused by a bacterium called *Mycobacterium tuberculosis*, was an important force of selection, especially in crowded cities from the Middle Ages to the early twentieth century. One scientist has proposed that people heterozygous for the alleles that cause cystic fibrosis (an inherited lung disorder discussed in Chapter 3, p. 77) were protected against tuberculosis; they therefore survived tuberculosis epidemics in greater numbers than did people without cystic fibrosis alleles. As the heterozygotes
increased in number, some of them married one another, and, on average, one out of four of their children became afflicted with cystic fibrosis.

What about the geographic variation in blood groups and other genetic traits? There is evidence that at least some of this variation may also result from the natural selection brought about by various medical conditions. In a smallpox epidemic in Bihar province, India, researchers found that those who died were more often of blood group A, while survivors were more often of blood group B. In similar fashion, cholera selects against blood group O and favors blood group B. (Note that these studies demonstrated a difference in fitness, but did not explain the mechanism.) Other studies have shown statistical correlation of various blood types with other diseases: blood group O is correlated with an increased risk of duodenal ulcers and ovarian cancers, and blood group A with a slightly increased risk of stomach cancer. Associations of particular blood groups with cancers of the duodenum and the colon have also been postulated. Such statistical associations do not necessarily indicate a cause-and-effect relationship between the associated factors.

Fatal diseases are among the most striking agents of natural selection, but there are many other selective forces. We examine some of these other forces of natural selection in the next section.

THOUGHT QUESTIONS

1. How is an average life expectancy measured? Why is the average life expectancy of a population more affected by the deaths of children (e.g., from malaria) than by the deaths of elderly people?

2. All heterozygous carriers of the allele for G6PD deficiency are female. What does this tell you about the location of the G6PD gene? (You may need to review Chapter 3 to answer this question.)

Natural Selection by Physical Factors Causes More Population Variation

There are other agents of natural selection in addition to diseases. Among them are climatic factors such as temperature or sunlight, as well as climatic variation that makes food more scarce at some times of year or from one year to another. Like the genetically based traits that confer protection against disease, other genetic variation between populations has arisen in response to these other selective factors. In this section we look at some of these other factors and how they have selected in different geographic regions for differences in the genetically regulated aspects of physiology and of body shape and size.
Human variation in physiology and physique

During part of the Korean War (1950–1953), American soldiers were exposed to the fierce, frigid conditions of the Manchurian winter. Many soldiers were treated for frostbite. Most of the Euro-American (Caucasian) soldiers responded well to the medical treatment that was given, but a disproportionate number of African American soldiers did not and many of them lost fingers and toes as a result. Disturbed by these findings, the U.S. Army ordered tests on resistance to environmental extremes in soldiers of different racial backgrounds.

In one series of tests, army recruits were required to perform strenuous tasks (such as chopping wood) under a variety of climatic conditions. In a hot, humid climate, the African American soldiers were able to continue working the longest and performed the best as a group; Asian American and Native American soldiers performed nearly as well as the African Americans, and Euro-American recruits lost excessive fluids through sweating and became easily fatigued and dehydrated. Under dry, desert conditions, the Asian American and Native American soldiers did best, the African Americans were second best, and again the Euro-American soldiers became dehydrated. Under extremes of cold, it was the Euro-American soldiers who did best, followed closely by the Native American and Asian American soldiers; the African Americans shivered the most and some became too cold to continue. These tests demonstrated definite differences between groups in bodily resistance to physiological stress under a variety of environmental extremes. The significance of these differences was enhanced by the fact that, in other respects, the recruits represented a fairly homogeneous population: 18- to 25-year-old males who had all been screened by the army as being physically fit and free from disease and who had passed the same army physical and mental exams.

Other physiologists outside the Army conducted tests in which adult male volunteers immersed their arms in ice water almost to the shoulders. African Americans in general shivered the most and suffered the most rapid loss of body heat, as measured by a decline in body temperature. Euro-Americans and Asian Americans lasted longer without shivering, but they, too, eventually suffered loss of body heat. Only the Inuit (Eskimo) volunteers were able to keep their arms immersed indefinitely without any discomfort and without shivering. Subsequent studies that replicated these results made the additional finding that diet is also a factor: Inuit volunteers who ate high-protein, high-fat diets (traditional for the Inuit) did far better than other Inuit who had become acculturated to American dietary habits. It would be a mistake, however, to extrapolate findings from studies such as these beyond the groups used for the tests (adult males in good health) without further investigation. Many traits vary with age or sex or both.

Bergmann’s rule. Genetically based differences in physiology that correlate with climate are the basis for a number of ecogeographic rules. Adaptations can also work indirectly, through variables such as body physique. Biologists have long noticed certain general patterns of geographic variation among mammals and birds. In one such pattern, called Bergmann’s rule, body sizes tend to be larger in cold parts of the range
and smaller in warm parts. This can be explained by the relationship of body size to mechanisms of heat generation and heat loss. For example, an animal twice as long in all directions as another animal has eight times the volume of muscle tissue generating heat \((2 \times 2 \times 2 = 8)\) as the smaller animal but only four times the surface area over which heat is lost \((2 \times 2 = 4)\). Thus, the larger animal is twice as efficient as the smaller \((8/4 = 2)\) in conserving heat under cold conditions. A survey of human variation confirms that the largest average body masses are found among people living in cold places (like Siberia), while most tropical peoples within all racial groups are of small body mass, even when their limbs are long. These relationships of body size to climate result from natural selection acting on genetic variation within populations over long periods. It does not mean that a person of a certain genotype will grow larger if they move to a cold climate.

**Allen’s rule.** Another broad, general phenotypic pattern in most geographically variable species of mammals and birds is Allen’s rule: protruding parts like arms, legs, ears, and tails are longer and thinner in the warm parts of the range and shorter and thicker in cold regions. This rule is usually explained as an adaptation that conserves heat in cold places by reducing surface area and dissipates heat more effectively in warm places by increasing surface area. Human populations generally follow this rule: Inuit people have shorter, thicker limbs, while most tropical Africans have longer, thinner limbs (Figure 7.12). There are exceptions, however: a number of forest-dwelling populations along the Equator are much smaller than Allen’s rule would predict, although they are usually thin-legged. Also, the tallest (Tutsi) and shortest (Mbuti) people on Earth live near one another in the Democratic Republic of Congo (formerly called Zaire), showing that climate is not the only instrument of natural selection influencing limb length or overall height within populations.

**Diabetes and thrifty genes.** Diabetes, a potentially life-threatening illness in many populations, may be an indirect result of one or more of the so-called ‘thrifty genes’ that protected certain people from starvation in past centuries. Ancestral Polynesians, for example, endured uncertain journeys over vast stretches of Pacific Ocean waters. Uncertain food supplies during such voyages selected for people who could withstand longer and longer periods of starvation and still remain active. The postulated ‘thrifty genes’ may have caused excess food, when it was available, to be converted into body fat that could be used for energy in times of famine. The result was a population that was stocky in build and resistant to starvation in periods when food supplies were low but that was also more susceptible to diabetes under modern conditions, when physical exhaustion is rare and food is always available. Diabetics fed on ‘ordinary’ diets have excess sugar in their blood, much of which is converted to fat and stored. Although diabetes is itself an unhealthy condition, the storage of fat may have been, under conditions like those described for the early Polynesians, an adaptive trait. Perhaps diabetes is an unfortunate modern consequence of having one or more alleles originally selected for their ability to convert sugar to body fat.
A similar history of selection for ‘thrifty genes’ (not necessarily the same ones) might also explain the late twentieth-century upsurge of diabetes in certain Native American populations, notably the Navajo and Pima of the southwestern United States. The risks that selected for ‘thrifty genes’ in the past were more significant in barren environments than in places in which the food supply was more assured. However, the commercial introduction of sugar-rich foods and a change from an active to a sedentary lifestyle have both raised the risks of diabetes, which are higher for sedentary people eating carbohydrate-rich diets. Because of these environmental changes, the genes that were once advantageous have in some cases turned into a liability, putting people of these genotypes at greater risk of diabetes. The Navajo and Pima have discovered that a return to frequent long-distance foot racing (a traditional activity they had nearly abandoned) has kept their populations healthier and has significantly lowered the incidence of diabetes in the runners. Not enough time has yet elapsed for the allele frequencies of the ‘thrifty genes’ to have again changed in this population, but the partial return to an earlier lifestyle has changed the environmental stresses and decreased the incidence of diabetes.

Natural selection, skin color, and disease resistance

The skin is the largest organ of the body and a major surface across which the body makes contact with the forces of natural selection in its environment. Human populations vary widely in skin color. Could these differences in skin color be adaptive?

Geographic variation in skin color. Skin color is one of the most visible human characteristics, and the one to which Americans have always paid the most attention when identifying race. Long-standing patterns of geographic variation are easier to understand if we ignore the population movements of the years since A.D. 1500 and consider only those populations still living where they did before that time.

Europe has for centuries been inhabited by light-skinned peoples, Africa and tropical southern Asia by dark-skinned peoples, and the drier, desert regions of Asia and the Americas by people with reddish or yellowish complexions. What is even more remarkable is that we find geographic variation along the same pattern within most continents, and in fact greater variation within the larger population groups than between such groups. For example, among the group of populations spread continuously from Europe across Western Asia to India, we find the lightest skin colors (also eye and hair colors) in Scandinavia and Scotland, progressively darker average colors (and darker hair) closer to the Mediterranean Sea, further darkening as we move through the Middle East and across Iran to Pakistan and India, and the darkest at the southern tip of India and on the island of Sri Lanka. A similar gradient (a cline) for skin color can be found among Asians, from northern Japan south through China into the Philippines and Indonesia.

Why would it be adaptive for people to be light-skinned in Europe but dark in Africa, Sri Lanka, and New Guinea? Notice that there are some very dark-skinned people outside Africa, and they generally have few other physical or genetic characteristics in common with Africans other than their dark skin colors. The natives of Sri Lanka, for example,
have very straight hair and blood group frequencies totally different from those of Africa. One clue to this puzzle is that all very dark-skinned peoples have lived for millennia in tropical latitudes.

**Sunlight as an agent of selection.** Tropical regions receive on a year-round basis more direct sunlight than do temperate regions. In fact, the amount of sunlight received at ground level decreases with increases in latitude and corresponds more closely to belts of latitude than to variations in temperature. This is especially true for light in the ultraviolet region of the sun's spectrum.

If we exclude places where few people live, Europe receives the least sunlight of all the inhabited regions of the world. This is first and foremost a function of latitude. Europe includes populated regions of higher latitudes than on any other continent: London and fourteen other European capitals are located north of latitude 50°, while North America and Asia above this latitude contain few large cities and a great deal of sparsely inhabited land. Europe also has a frequent cloud cover that screens out even more of the Sun's rays. As a result of both high latitude and cloud cover, people in Europe receive much less exposure to ultraviolet light than most other people.

That sunlight levels select for body coloration is described by a third ecogeographic rule, **Gloger's rule.** While Bergmann's and Allen's rules, described earlier, take only temperature into account, Gloger's rule takes into account sunlight and humidity as well. Under Gloger's rule, most geographically variable species of birds and mammals have pale-colored or white populations in cold, moist regions, dark-colored or black populations in warm, moist regions, and reddish and yellowish colors in arid regions. We do not know all the reasons for this variation. Camouflage has been suggested as a cause, but vitamin D synthesis also has an important role.

Vitamin D is needed for the proper formation of bone. Children who do not receive adequate vitamin D during growth suffer from a condition called rickets, a disease of bone formation that may result in weakness and curvature of the bones (especially those of the legs) and in crippling bone deformities if left untreated. Sunlight is necessary for vitamin D synthesis. Many foods are rich in vitamin D, such as egg yolks and whole milk, but most vitamin D found in foods is in a biologically inactive form. The final step of vitamin D biosynthesis takes place just beneath the skin, with the aid of the ultraviolet rays of natural sunlight. This is why vitamin D is sometimes called the ‘sunshine vitamin.’ To get adequate amounts of vitamin D, a population must have both adequate intake of the vitamin in the diet and adequate exposure to sunlight. European populations have the lightest skin colors (and they get lighter the farther north you go) as an adaptation that allows maximum sunlight penetration into the skin. Europeans also have many cultural adaptations related to vitamin D intake, such as the eating of cheeses and other fat-rich milk products containing vitamin D. Northern Europeans place great value on outdoor activity at all times of the year, including such occasional extremes as nude dashes into the snow after the traditional sauna.

In northern Europe, people with dark skins could be at a very high risk of vitamin D deficiency because melanin pigment blocks out a large proportion of the Sun's ultraviolet rays. Very few dark-skinned people
lived in northern Europe even as immigrants. This has changed since World War II, when synthetic vitamin D became widely available. Because this prepared vitamin D is already in its active form, sunlight is no longer needed for its activation. Dark-skinned people can now live and remain healthy in northern latitudes without developing deficiency diseases.

At latitudes closer to the Equator, other problems exist. The same wavelengths of ultraviolet that are needed in the final step of vitamin D synthesis are also cancer-causing. Skin cancer (see Chapter 12) is generally a disease of those white-skinned people who are overexposed to the Sun’s direct rays. Recently, it has also been found that ultraviolet radiation from overexposure to the Sun destroys up to half of the body’s store of folate, an important vitamin that protects women against giving birth to children with spina bifida and other neural tube defects. Melanin pigment screens out ultraviolet radiation and thus protects against both cancer and folate deficiency. Populations of all racial groups living closer to the Equator have been selected over the millennia to have darker skins. Those individuals who had lighter skins were less fit in this Equatorial environment; that is, they more often got skin cancer and died earlier, or more often had deformed babies or miscarriages. Melanin pigment absorbs much of the ultraviolet light, protecting dark-skinned people against skin cancer and folate deficiency, while still allowing enough ultraviolet light through for adequate synthesis of Vitamin D.

There are no known genes that code specifically for skin color (except that the allele for albinism, present in all human populations, prevents synthesis of all melanin pigment and results in pale white skin). Apart from environmental effects (such as suntanning), there are probably dozens of genes that produce enzymes that influence the synthesis of melanin and other skin pigments—so many that it is difficult to study any one of them apart from the others. Nevertheless, natural selection has favored different levels of pigmentation in different geographic regions.

Nutritional sources of vitamin D in the far north. For the reasons given in the preceding sections, populations living in the high latitudes are generally light-skinned and populations that are adapted to living in tropical latitudes are generally dark-skinned. There is one very interesting exception: the Inuit populations of Arctic regions, sometimes known as Eskimos. (These people have always called themselves Inuit; the name ‘Eskimo’ was a pejorative name used by their enemies.) The Inuit are not very light-skinned, nor do they expose themselves much to sunlight. Most Inuit people live in places so cold that the exposure of bare skin poses a greater danger than any benefit of ultraviolet rays could overcome, and most Inuit are fully protected by clothing that offers hardly any exposure to the Sun. So how do they get enough vitamin D? The Inuit have discovered their own way of staying healthy. One of the world’s richest sources of vitamin D is fish livers, especially those of cold-water fishes. (Cod liver oil is a very rich source of both A and D vitamins.) Moreover, the vitamin D in fish oils is fully synthesized and needs no sunlight to activate it. So, instead of having pale skins and traditions of exposing their skins to sunlight, the Inuit have traditions of catching cold-water fish (Figure 7.13) and eating them whole,
liver and all. These traditions have allowed them to stay healthy in a climate that is too cold and too sunless for most other populations.

In all of the above examples, a population that has lived in a particular geographic area for long periods has become adapted to the temperature, humidity, sunlight, and other conditions of their environment. The evidence presented in this chapter and in Chapter 5 suggests that natural selection is largely responsible for these adaptations.

**THOUGHT QUESTIONS**

1. If people differ in their resistance to extreme cold or heat, does this mean that the difference is genetic? What would you need to know to answer this question? How could an experiment be arranged to test this?

2. Blood type O is statistically associated with duodenal ulcers, one of many such correlations between a blood type and a disease. Does a correlation demonstrate a cause? Does a correlation imply a mechanism of some kind? Does a correlation suggest new hypotheses? How can scientists learn more about whether there is a causal connection between the blood type and the disease?

**Concluding Remarks**

Throughout the history of biology, scientists have developed various ways of describing groups of people. Some of these groupings have been known as races. Some concepts of race have attempted to find biological explanations for the racial groupings already established by various societies. Morphological concepts of race divided humans on the basis of their physical appearance. Biologists and anthropologists of the past gathered descriptive data about the physical characteristics of different populations and assumed that each group was distinct and unchanging. More recently, biologists have abandoned these concepts, in part because of the racism that has flowed from them, but also because these ideas no longer fit the data that we now have. The population genetics theory of human variation views human populations as varying continuously, with no group being uniquely different from any other. Biological differences among human populations are products of evolution. Like any other species, humans can evolve only when genetic variation is present in a population. When a population encounters some agent of natural selection, such as disease or climate, people with certain genotypes survive in greater numbers and leave more offspring than those with other genotypes. Over long periods, this process results in the adaptation of a population to its environment, with allele frequencies differing from one population to the next. This evolution continues today, although the increased mobility of people and technological alterations of the environment are
slowly making populations less distinct than in past centuries. Populations vary in the frequencies of traits; they do not carry any unique traits. There is no biological phenotype, genotype, or DNA sequence that can assign an individual to a race or to a population. Although our biological concepts about race and other human variation have changed over time, racism will continue to exist if one group of people is held to be more valuable than another.

Chapter Summary

- Human populations vary geographically. Phenotypic and genotypic variation within populations usually exceeds variation between them.
- Phenotypic variation within a population can be either continuous or discontinuous. Continuously variable traits, in which all intermediate values are possible, can be described in terms of average values for each population. Discontinuously variable traits, such as those that are either present or absent, can be described in terms of phenotypic frequencies or allele frequencies.
- Differences among populations have historically been described in terms of culturally defined or morphological races.
- Population genetics allows us to describe groups of populations that differ from one another by certain characteristic allele frequencies.
- Most allele frequencies vary gradually and continuously among populations, without abrupt boundaries. Continuous variation from one population to another is best described in terms of geographic gradients, also called clines. Clines can be plotted on maps for average values of continuously variable traits such as height, or for population frequencies of discontinuously variable traits such as particular blood groups or alleles or DNA sequences.
- When more than one allele of a gene persists in a population this is called a polymorphism.
- The Hardy–Weinberg equilibrium describes the conditions under which allele frequencies remain constant in a population.
- Populations that were at one time small may have allele frequencies that have been shaped in part by genetic drift.
- Aside from genetic drift, most geographic variation among human populations has resulted from natural selection producing adaptation of the population to the environment.
- Disease is an important force of natural selection. Malaria, a widespread parasitic infection, has selected in different regions for high frequencies of alleles associated with sickle-cell anemia, thalassemia, and G6PD deficiency, all of which protect heterozygous individuals against malaria. Malaria and other diseases result in balanced polymorphism whenever the heterozygous genotype enjoys maximum fitness.
- Temperature selects for geographic variation in the alleles influencing body size and body shape.
• Ultraviolet light at different latitudes selects for geographic variation in the population frequencies of alleles influencing skin color. Alleles producing pale skin are selectively favored at high latitudes as an adaptation to absorb more ultraviolet light and prevent vitamin D deficiency. Alleles producing dark skin are favored near the Equator as a protection against skin cancer from too much ultraviolet exposure.

CONNECTIONS TO OTHER CHAPTERS

Chapter 1
Every study of human variation is conducted in a cultural context.

Chapter 1
Studies of human variation have ethical implications, including those arising from inappropriate use of the results.

Chapter 3
Many human variations have a genetic basis; such alleles arose ultimately from mutations.

Chapter 5
Human population variations reflect evolutionary processes, including mutation, natural selection, and genetic drift, all of which continue to work in modern populations.

Chapter 9
Nearly all human populations are growing, and some are growing much faster than others. Population growth and migrations change various allele frequencies.

Chapter 10
Different populations sometimes have different ways of meeting their nutritional requirements.

Chapter 12
Some types of cancer are more frequent in some human populations and less frequent in others.

Chapter 18
Human variation is an example of biodiversity at the population level.

Chapter 19
Because of damage to Earth’s ozone layer, people are being exposed to increased ultraviolet radiation, which may select over time for a shift in allele frequencies leading to a darkening of skin pigmentation in human populations.

PRACTICE QUESTIONS

1. How many different genotypes can code for the blood group B phenotype? What are they? Are they heterozygous or homozygous?

2. How many different genotypes can code for the blood group AB phenotype? What are they? Are they heterozygous or homozygous?

3. How many different genotypes can code for the blood group O phenotype? What are they? Are they heterozygous or homozygous?

4. How many different genotypes can code for the Rh+ phenotype? Are they heterozygous or homozygous?

5. How many different genotypes can code for the Rh- phenotype? Are they heterozygous or homozygous?
6. If the allele frequency of $Hb^S$ in a population is 0.1, how many people in that population will be heterozygous $Hb^AHb^S$? (Review the Hardy–Weinberg equation.)

7. How many different host species does the *Plasmodium* parasite need to complete its life cycle?

8. Why do people who are heterozygous for sickle-cell anemia have less severe anemia than people who are homozygous $Hb^SHb^S$?

9. How does the bottleneck effect alter the allele frequencies of a population?

10. What is a balanced polymorphism? Give an example.