11

Pigmentation

When we think about human variation or race, we probably think first about color. Of all human diversity, variation in pigmentation, particularly skin color and hair color, is the most visible and most commonly used to sort and classify human beings. Skin color (and to a lesser extent hair and eye color) has consistently been the primary trait used in racial classifications over the centuries. For example, Linnaeus's preliminary division of humanity into four varieties was based primarily on geography and skin color (Klass and Hellman 1971, Kennedy 1976). The reliance on skin color as a racial trait continued through the centuries in the racial classifications of Cuvier, Blumenbach, and others.

Anthropological studies of human skin color have shown that it is a continuous trait, where people across the world range from very dark to very light. When we put all these data together, there are no gaps in the distribution, making racial classification an arbitrary exercise when considering the total range of variation across the human species. However, skin color is not randomly distributed across the planet, and there is a strong geographic relationship. While, in general, Europeans are lighter than Africans, this geographic correspondence does not replicate perfectly geographic definitions of race. Instead, the relevant correlation is with the distance from the equator; as shown below, human skin color is darkest at or near the equator and becomes increasingly lighter with greater distance from the equator, north or south. This strong correlation with latitude suggests past natural selection in response to environmental factors that also vary by latitude, although there is continued debate over the relative impact of such factors. Compared to genetic and DNA markers and craniometric traits, variation among geographic regions is greater for skin color (Relethford 2002), a pattern consistent with natural selection that varies across environments.

This chapter reviews what is known about human pigmentation, focusing primarily on skin color but also considering variation in hair and eye color.

SKIN COLOR

What factors account for the enormous range of variation in human skin color? This section examines briefly the underlying physiology of skin color and its relationship to genetic variation and environmental factors.

The Biology of Skin Color

Physiology

The brownish black **melanin** pigment affects the majority of variation in human skin color. Melanin is produced by **melanocytes**, which are cells in the epidermis (the outermost

layer of the skin). The melanin pigment is synthesized by the **melanosomes**, which are organelles inside the melanocytes. Packets of melanin are then distributed to the epidermal skin cells. While early studies suggested that all human populations have the same number of melanocytes, a recent review suggests that individuals with light skin have, on average, fewer melanocytes than those with dark skin (Beall and Steegmann 2000). Differences in degree of pigmentation (lighter versus darker) also depend on how many cells produce melanin, how they cluster together, the rate of melanin formation, and the rate of melanin transfer to the epidermal cells (Szabo 1967, Robins 1991).

Although most of the variation in human skin color is due to differences in melanin distribution, some color differences are also due to three other pigments: carotene, oxy-hemoglobin, and reduced hemoglobin. Carotene is a yellowish pigment that normally contributes very little to variation in skin color except in light-skinned individuals suffering from a disease known as carotenemia, which results in a yellowish hue to the skin (Robins 1991). Hemoglobin pigments have a more noticeable effect, particularly in light-skinned people. Hemoglobin gives oxygenated blood its reddish color. Since light-skinned people have less melanin, the reddish color shows through the skin, giving a pinkish hue.

Measurement of Skin Color

When we look at someone's skin color and attempt to classify it into discrete categories such as "white" and "black," the result is necessarily crude and subjective, and many individuals do not fit neatly into one category or the other. Adding more categories, such as "brown," does not help because it becomes increasingly arbitrary where to draw the line between one color and the next. A more objective, standardized method of assessing skin color is clearly needed.

Early attempts to quantify human skin color also used crude standards, most frequently matching of the subject's color with a series of paper or tile standards (Byard 1981). Such measurements are not only inherently subjective but also affected by the source of external lighting (much like trying to match paint samples by eye). Additionally, the use of such standards imposes discrete groupings on a trait that varies continuously.

A more precise means of quantifying skin color variation based on **reflectance spectrophotometry** was developed by Edwards and Duntley (1939). Reflectance spectrophotometers provide an accurate means of objectively measuring skin color that is not prone to *interobserver error*, the error introduced when different observers measure different groups of people (Lees et al. 1978). Light in the visible spectrum at different wavelengths, ranging from blue to red, is bounced off of human skin. Lighter skin will reflect more light back than darker skin, thus allowing the percentage of reflected light to be used as an objective measure of skin color. By varying the wavelength of visible light, different measures of skin reflectance can be produced for different wavelengths. This method was cumbersome and expensive, requiring extensive laboratory equipment that is not feasible to transport around the world to populations of anthropological interest. By the 1950s, the necessary technology had developed in the form of portable abridged reflectance spectrophotometers. These machines, often used in the textile industry and other places where color matching is needed, are portable enough to transport to remote sites. The machines are referred to as "abridged" because they do not sample the entire visible spectrum but only a small number of wavelengths. Weiner (1951) and Lasker (1954) were the first to use these machines in anthropological studies.

Table 11.1 Wavelengths of filters used in E.E.L. and Photovolt reflectometers

Reflectometer	Filter	Wavelength (nm)
E.E.L.	601	425
	602	465
	603	485
	604	515
	605	545
	606	575
	607	595
	608	655
	609	685
Photovolt	Blue	420
	Triblue	450
	Green	525
	Trigreen	550
	Triamber	600
	Red	670

From Byard 1981.

These machines shine light through a glass filter of a given color, which then allows only that color to pass through to the subject's skin. The light is reflected back from the skin (with lighter skin reflecting more) and measured by a photocell. Two machines have been used most often in anthropological studies. The most commonly used machine is known as the **E.E.L.** machine, where the initials stand for the manufacturer, Evans Electroselenium Limited (today known as "D.S.L." for the company Diffusion Systems Limited). The E.E.L. machine uses nine colored filters, known as filters 601–609, to sample the visible spectrum. The filters range from blue to green to red. At one end of the spectrum, the number 601 filter only allows light at a frequency of 425 nm to pass, producing a deep blue light. At the other end of the spectrum the number 609 filter samples the visible spectrum at 685 nm (a red light). Another machine, the **Photovolt** reflectometer, uses six filters: two each for the blue, green, and red portions of the visible spectrum. Table 11.1 lists the filters and wavelengths for both machines. While the majority of anthropological studies have used the E.E.L. machine, some, particularly a number of studies on New World populations, have used the Photovolt machine. Unfortunately, the wavelengths sampled are not the same, and the two machines also differ in other ways, meaning that the results cannot be directly compared. Several formulae have been developed to convert readings from one machine to the other (Garrard et al. 1967, Lees and Byard 1978, Lees et al. 1979). Recently, two new handheld reflectometers have been described for their anthropological utility (Shriver and Parra 2000), although conversion formulae have not yet been developed to allow direct comparison with the older E.E.L. and Photovolt machines.

Skin reflectance is most often measured at the inner surface of the upper arm, slightly above the elbow (Fig. 11.1). This measurement site is easy to reach, typically socially acceptable to reveal, and a good index of skin color that has not been affected by environmental exposure, thus providing a measure of innate skin color. Some studies have also taken measurements of skin reflectance on the forehead and the back of the hand in order to also measure skin color at parts of the body that are likely to tan. The rationale
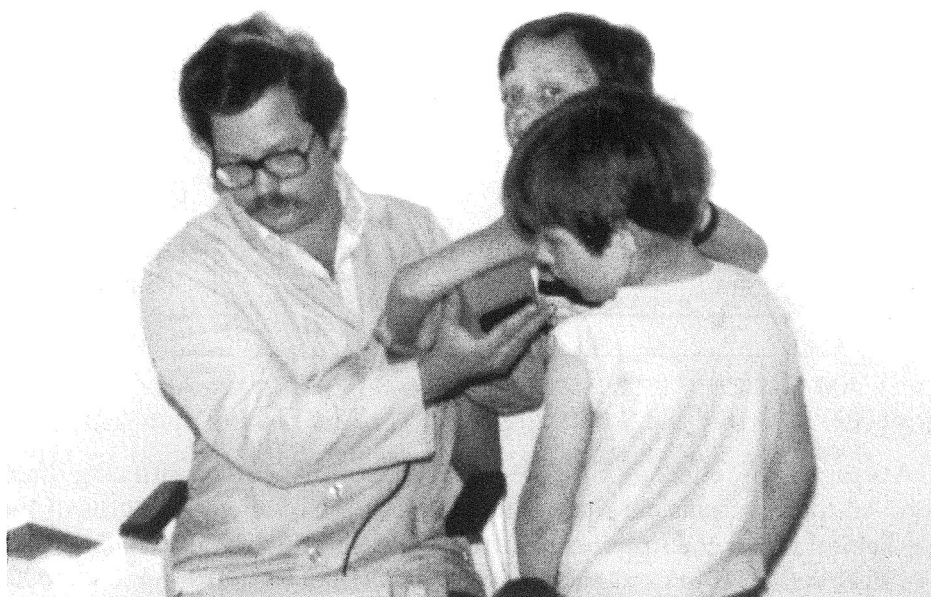
Figure 11.1. Skin color being measured by Dr. Francis C. Lees using an E.E.L. reflectance spectrophotometer. Skin color is quantified as the percentage of light reflected off of the skin at different wavelengths. (Photo by J. H. R.)

was that by comparing unexposed and exposed parts to the body, one could get an idea of a person's tanning capacity. This method has been dropped for the most part since Post et al. (1977) discovered that newborn children, who had not been previously exposed to sunlight, had differences in skin reflectance at different body sites. The foreheads of these children were 10%–12% darker than at the upper inner arm, showing an innate difference in pigmentation across the body such that any further comparisons would not necessarily indicate tanning potential.

The measurement of skin reflectance at different wavelengths allows a graphic illustration of skin color differences between samples. Figure 11.2 presents the average skin reflectance values for the nine E.E.L. wavelengths for four different populations. This graph reveals several features. First, the light-skinned population from the Netherlands shows greater skin reflectance at all wavelengths, followed by the sample from India, the Bantu, and the Fali Tinguelin population from Africa. Second, the differences between populations are most apparent in the red part of the visible spectrum; indeed, the E.E.L. filter 609 (685 nm) is often used as the best single measurement of skin color differences. Third, while reflectance generally increases with wavelength, the sample from the Netherlands shows a reduction in reflectance at 545 nm (filter 605). This reduction, known as the "hemoglobin dip," is due to the fact that hemoglobin has an absorption frequency in the green part of the visible spectrum, near filter 605. At this wavelength, the green light is absorbed by hemoglobin and therefore does not reflect back as much, making the skin reflectance lower than at other, nearby wavelengths. This dip is apparent only in light-skinned individuals, whose relative lack of melanin does not block the green light. Thus, readings at this wavelength are picking up on redness of the skin in light-skinned people due to oxygenated hemoglobin. Individuals with darker skin do not show this effect. In general, the readings at different wavelengths are highly correlated.
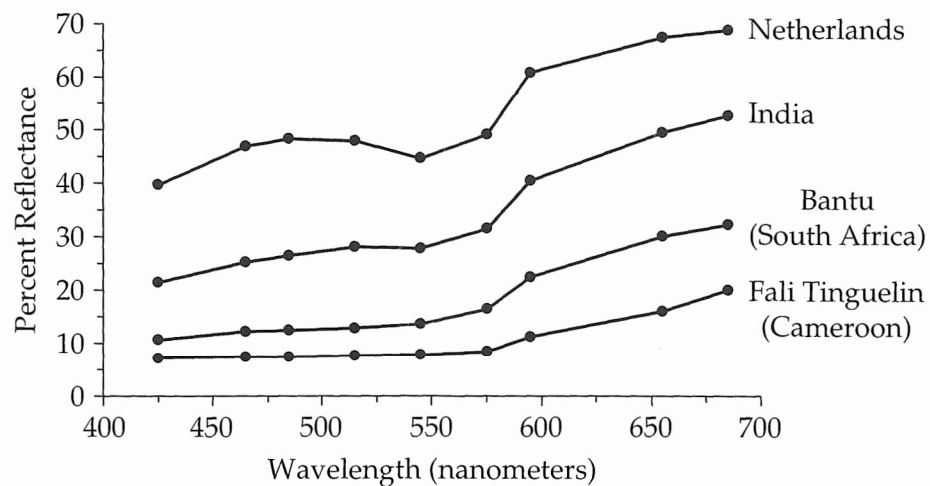
Figure 11.2. Average skin reflectance for four human populations of differing degrees of pigmentation. All four samples are males, measured at the nine wavelengths of the E.E.L. machine (see Table 11.1) at the upper inner arm. Note that the lighter the skin, the higher percentage of light reflected back at each wavelength. (Data From Rigters-Aris 1973a,b; Tiwari 1963; Wassermann and Heyl 1968.)

Korey (1980) presents a calculus-based method for deriving average skin reflectance that can suffice in many analyses of skin color variation.

Differences in skin reflectance between populations that are widely different in pigmentation reflect underlying ancestry. The children born to one light-skinned parent and one dark-skinned parent will tend to have, on average, medium-pigmented skin. This effect is shown in Figure 11.3, based on a study of English children born to light-skinned English women and dark-skinned African men (Harrison and Owen 1964). The skin
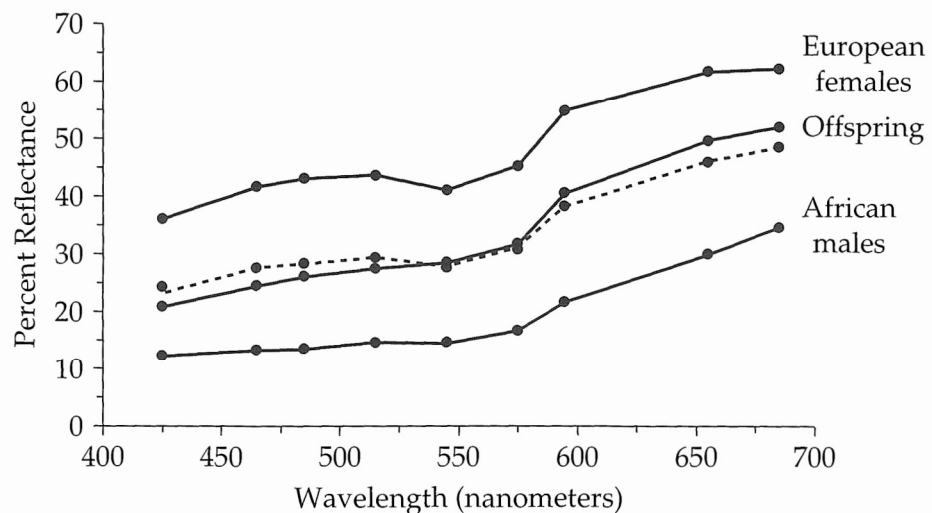

Figure 11.3. Average skin reflectance for a group of offspring resulting from the mating of European females and African males residing in Liverpool. Skin reflectance was measured at the nine wavelengths of the E.E.L. machine at the upper inner arm. The dashed line indicates the expected average between adult male and female skin color. The observed curve for the offspring (solid line) is very similar to this mid-parent average. (Data From Harrison and Owen 1964.)
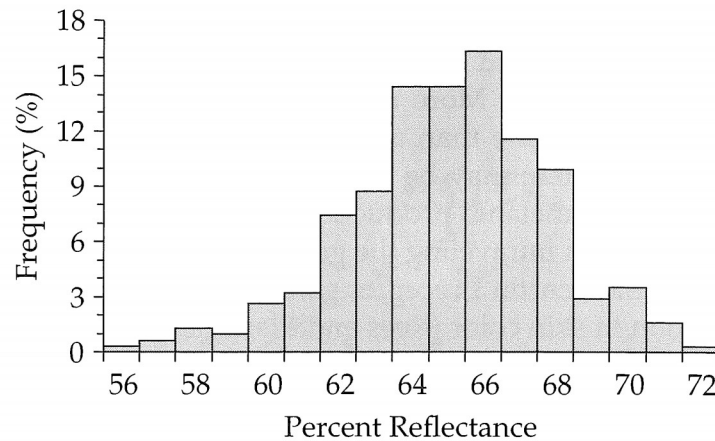
Figure 11.4. Histogram of skin reflectance as measured at E.E.L. filter 609 (685 nm) for 312 Irish male children. (Unpublished data as described in Relethford et al. 1985.)

reflectance curve of the offspring is very close to the expected curve based on a 50:50 contribution from each population. Methods have been developed using this observation to estimate degree of ancestry in admixed populations (Lees and Relethford 1978, Korey 1980, Relethford et al. 1983). Of course, these curves only indicate the average of the sample; the children do not all have the same skin color, and some are lighter and some are darker (Harrison and Owen 1964).

Within any population there is considerable variation in skin color, as shown in the example of Irish male children in Figure 11.4. Contrary to superficial visual inspection, not all people in any population have the same skin color; some are lighter, and some are darker. Some of this variation is genetic in nature, and some is environmental. The fact that some Irish boys are lighter than average and some are darker than average refutes the common misconception of human variation that "they all look alike." This is clearly not the case. Also, note that the distribution in Figure 11.4 is a normal ("bell-shaped") one, as expected in a complex trait that reflects the summation of a number of different influences. In the case of skin color, these influences are due to the action of different genes and the environment, the subject of the next section.

The Genetics of Skin Color

A number of studies have shown that skin reflectance has a moderate to high heritability (e.g., Post and Rao 1977, Clark et al. 1981, Frisancho et al. 1981), typically ranging from about 0.5 to 0.8. As always, differences in choice of population and method of estimating heritability produce a range of estimates. The higher heritabilities were derived from twin studies, which tend to inflate heritability estimates (Williams-Blangero and Blangero 1992). The most elaborate analysis to date simultaneously analyzed skin reflectance in Nepalese populations at three E.E.L. filters (601, 605, 609) in a variety of pedigrees of different sizes and found a heritability of 0.66 for filter 609 and somewhat lower heritabilities for the other filters (Williams-Blangero and Blangero 1992). These studies show that skin color variation is strongly affected by genetic variation in human populations.

Early studies of the quantitative genetics of skin color suggested that it was best explained by a polygenic model with three or four loci, each having approximately the same effect on skin color (e.g., Harrison and Owen 1964, Harrison et al. 1967). Later work

by Byard (1981) and Byard and Lees (1981) showed problems with the underlying method, and Williams-Blangero and Blangero (1992) suggested that there are likely more loci affecting skin color variation. More recent work has suggested that a major gene model might be more appropriate than a polygenic equal and additive effects model. Recent work suggests that there may be three major genes that influence human skin color through their effects on melanin production (Sturm et al. 1998, Blangero et al. 1999). The greatest success to date in unraveling the genetics of human skin pigmentation is the identification that the melanocortin 1 receptor gene (*MC1R*) located on chromosome 16 is associated with variation in skin color (Rees and Flanagan 1999, Harding et al. 2000).

Variation in Human Skin Color

Human skin color varies both within and between populations. Variation within populations is affected somewhat by sex and age, both of which are related to hormonal influences. Variation between populations is strongly correlated with latitude, suggesting differing patterns of natural selection in the past.

Sex Differences

Among adults, males are generally darker than females, possibly reflecting differences in melanin (Edwards and Duntley 1939) and sex hormones (Byard 1981). While most studies have found adult males to be darker, some have found the reverse or no effect, suggesting that in some cases there are cultural differences in exposure to sunlight, even when skin reflectance is measured at the upper inner arm site (Byard 1981). In general, the differences between adult males and females are relatively small. The situation is a bit more complicated in children and adolescents, where the darker sex varies by both wavelength and the specific age of measurement (Relethford et al. 1985). The general tendency for women to be lighter than men (usually around 3%–4%) may reflect a need in women for more calcium, particularly during pregnancy and breast-feeding. Jablonski and Chaplin (2002) suggest that women are lighter to allow slightly more ultraviolet (UV) radiation to penetrate the skin and increase production of vitamin D.

Aging

In general, females tend to show a noticeable darkening during adolescence and adulthood. While some studies of males show some aging effect, others do not; and age-related trends are usually more noticeable in females. Kalla (1974) and Byard (1981) suggest that age-related changes in skin color, where they exist, are likely to reflect the effects of changing hormone levels on pigmentation. Relethford et al. (1985) note that Irish female children were significantly darker than Irish male children for filters in the green range of the visible spectrum and suggest that this difference relates to higher subcutaneous blood flow in light-skinned females. It is difficult, however, to generalize too much since both Byard (1981) and Williams-Blangero and Blangero (1991) note that the relationship of skin color and age can vary considerably even among closely related populations.

The Geographic Distribution of Skin Color

Throughout recorded history, people have observed a definite geographic pattern to human skin color. These casual observations have since been confirmed by detailed analysis. Human skin color varies with latitude such that indigenous populations at or near the equator tend to be dark-skinned and populations that live farther away from
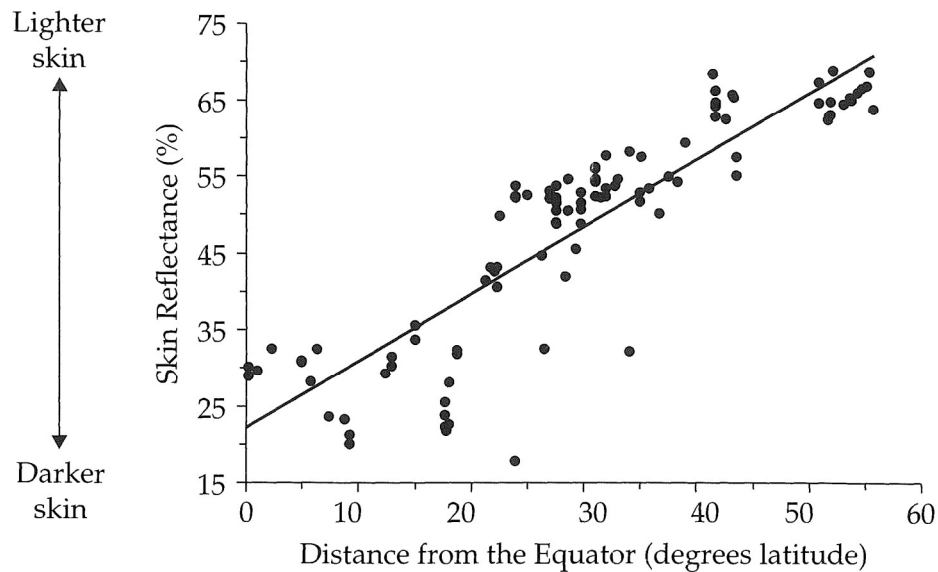
Figure 11.5. Relationship of skin reflectance (E.E.L. filter 609) and distance from the equator for 102 male samples across the Old World. (Data from Relethford 1997.)

the equator are lighter. These studies show significant positive correlations of skin reflectance with distance from the equator (Roberts and Kahlon 1976, Tasa et al. 1985, Relethford 1997, Jablonski and Chaplin 2000). Figure 11.5 provides an example of this relationship based on 102 male samples using E.E.L. filter 609, often taken to be the best single measure of melanin variation among human populations. While the relationship is not a perfect one, it clearly describes the average geographic distribution of human skin color, with latitude accounting for 77% of the total variation in skin color.

What reason could there be for this strong geographic distribution? Correlations of this magnitude suggest natural selection and some sort of environmental factor that also varies by latitude. Most often, the correlation between skin color and latitude is interpreted as a reflection of the geographic distribution of UV radiation, which is strongest at the equator and diminishes with increasing distance away from the equator. In this sense, latitude is considered a proxy for the average intensity of UV radiation. Jablonski and Chaplin (2000) note that skin reflectance is also strongly correlated with direct estimates of UV as obtained from spectrophotometric analysis of ozone mapping from space satellites.

The relationship of skin color, UV radiation, and latitude turns out to be more complicated. UV radiation tends to be higher in the Southern Hemisphere (below the equator) than at the same latitude in the Northern Hemisphere because of a variety of factors, including hemispheric differences in the ozone layer and the eccentricity of the earth's orbit, where the Southern Hemisphere is closer to the sun during its summer months (McKenzie and Elwood 1990, Relethford 1997, Relethford and McKenzie 1998). Given this hemispheric difference in UV radiation and the suggestion that worldwide variation in skin color is linked to UV radiation, we should expect to see a hemispheric difference in skin color at equivalent latitudes. Figure 11.6 presents the same graph as in Figure 11.5 but with each data point labeled as belonging to a Northern Hemisphere population or a Southern Hemisphere population. The Southern Hemisphere populations clearly tend to have lower average skin reflectance, and therefore darker skin color, than Northern Hemisphere populations at the same distance from the equator, which is
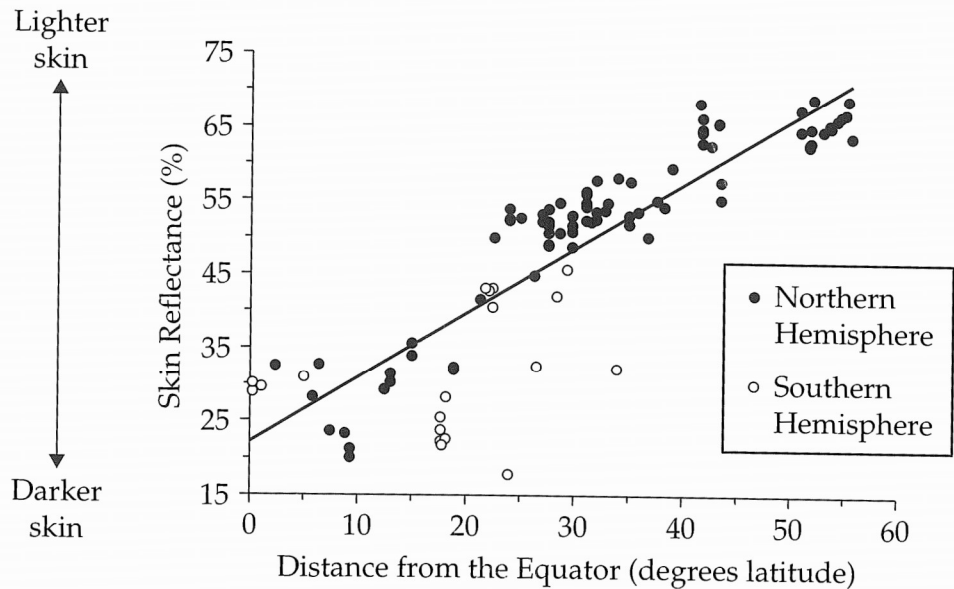
Lighter
skin

Darker
skin

Figure 11.6. Relationship of skin reflectance (E.E.L. filter 609) and distance from the equator for 102 male samples across the Old World showing differences between populations located in the Southern Hemisphere (*open circles*) and the Northern Hemisphere (*filled circles*). (Data from Relethford 1997.)

expected if UV radiation levels are higher in the Southern Hemisphere. Relethford (1997) extended this observation by developing a mathematical model that allows the relationship between skin reflectance and distance from the equator to be different in each hemisphere. As shown in Figure 11.7, skin reflectance is lowest (darkest) near the equator and

Lighter
skin

Darker
skin
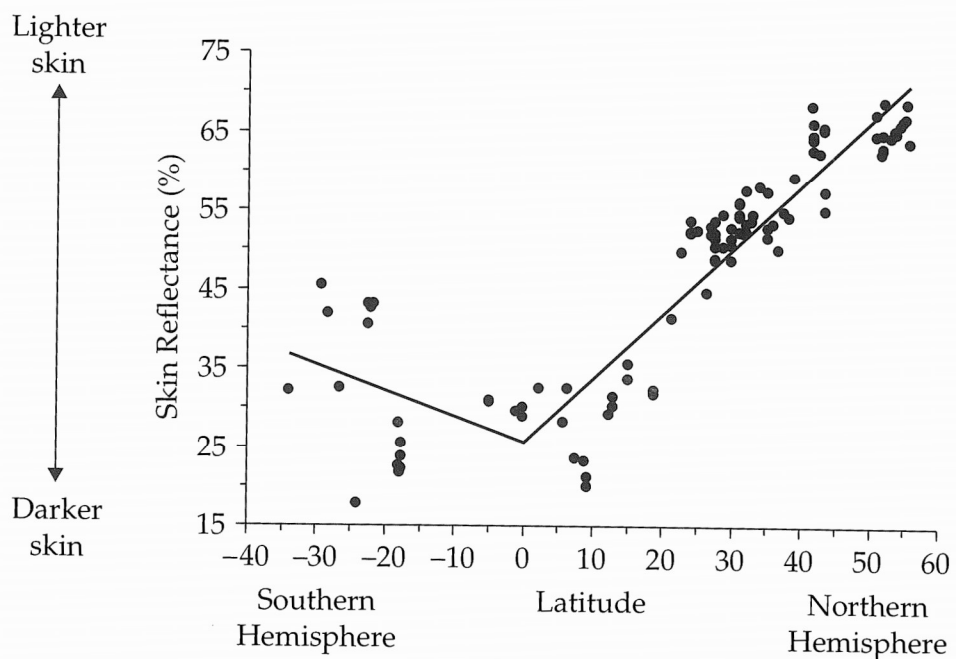
Figure 11.7. Relationship of skin reflectance (E.E.L. filter 609) and distance from the equator for 102 male samples across the Old World using a nonlinear model that allows for different slopes in the Southern and Northern Hemispheres. The change in skin reflectance with distance from the equator is less in the Southern Hemisphere than in the Northern Hemisphere. (Data from Relethford 1997.)

increases (lighter) farther away from the equator in both hemispheres but not at the same rate. Skin reflectance increases 8.2% for every 10 degrees of latitude in the Northern Hemisphere, but the rate of increase is less in the Southern Hemisphere, roughly 3.3% for every 10 degrees of latitude, as expected given higher UV radiation levels in the Southern Hemisphere.

THE EVOLUTION OF HUMAN SKIN COLOR

What events took place in the evolution of our species that led to such dramatic differences in skin color across the world? Comparing humans at different latitudes and with other primates suggests that there have been several major shifts in skin pigmentation during the past 6 million years or so of human evolution. Most of the skin of living primates is unpigmented except for exposed areas not covered with hair, suggesting a primitive feature in primates from which our ancestors changed. The best model for our own ape-like ancestors is probably the condition found in living chimpanzees, which have lightly pigmented skin covered with dark hair. As hominids adapted to the tropical savanna, the density of body hair most likely decreased and the density of sweat glands increased, both leading to a relatively naked skin best adapted for heat stress. As such, the hair no longer provided protection against the potentially harmful effects of UV radiation, and skin color most likely darkened as a selective response (Jablonski and Chaplin 2000).

Until almost 2 million years ago, all hominids lived in Africa and presumably had dark skin. As early humans expanded out of Africa into parts of Asia and Europe, they moved into northern latitudes with less UV radiation. The present-day distribution of human skin color suggests that, over time, populations living farther away from the equator became increasingly lighter. This distribution raises two basic questions: (1) Why specifically is dark skin more adaptive at or near the equator? (2) Why did skin color become lighter farther away from the equator? Can the same factor be responsible for both, or did different selective factors operate at different latitudes?

Selection for Dark Skin

Several models have been proposed to explain the evolution of dark skin in human populations at or near the equator, all linked to the greater amount of UV radiation found in that part of the world. The common element of these models is that dark skin acts as a natural "built-in" sunscreen (Jablonski and Chaplin 2002, 75).

Sunburn

UV radiation can cause severe damage to unprotected skin. The most immediate effect is sunburn. While we have a tendency to think of sunburn as a temporary, although painful, nuisance, severe sunburn can have major consequences. Severe sunburn can damage the sweat glands, which in turn can have a negative impact on a person's ability to handle heat stress (Byard 1981, Jablonski and Chaplin 2000). Given the fact that hominids first evolved in a tropical environment, the problem of heat stress may have been particularly acute if, as we expect, the transition to savanna life included a reduction in hair density and the increased production of sweat glands. Under these conditions, light-skinned individuals prone to sunburn and damaged sweat glands would be

at a disadvantage and would be selected against. According to this scenario, dark skin appeared in the first hominids as an adaptive response to protect against UV radiation damage to sweat glands. Another consequence of severe sunburn is an increase in infection to damaged skin cells (Byard 1981, Rees and Flanagan 1999, Jablonski and Chaplin 2000), which could also influence relative survival and render light-skinned individuals at a disadvantage. It seems reasonable to suggest that darker skin evolved near the equator at least partially as a means of protecting against UV radiation damage.

Skin Cancer

Damage to the skin due to UV radiation over extended periods of time can lead to skin cancer, brought about by UV radiation damage to a gene that normally inhibits cancerous growths. The risk of developing skin cancer varies among human populations by as much as 100-fold, a difference that appears to be strongly related to skin color (Rees and Flanagan 1999). There are two lines of evidence that link skin color, UV radiation, and skin cancer rates. First, there is a strong correlation of skin cancer incidence and mortality with latitude, with both decreasing with greater distance from the equator. Second, skin cancer rates are lower in darker-skinned populations than lighter-skinned populations at the same latitude. In Texas, for example, the incidence of nonmelanoma skin cancer is 5 per 100,000 among nonwhite residents compared to 113 per 100,000 among white residents (Beall and Steegmann 2000).

Given these data, one suggestion for the evolution of dark skin in humans living at or near the equator is that light-skinned individuals would be selected against due to increased susceptibility to skin cancer. It has been argued, however, that skin cancer is unlikely to have been a significant factor in the evolution of skin color because cancer tends to occur later in life, often after the reproductive years (Jablonski and Chaplin 2000). Since natural selection operates through differential survival and reproduction, any disease influencing survival after the reproductive years would not be subject to natural selection. Others argue that while the incidence of skin cancer among younger individuals may be lower, it is still sufficient for the action of natural selection. Robins (1991) compared skin cancer rates among albinos and nonalbinos in Africa. Since albinos lack any pigment, they are more subject to skin cancer. All of the albinos showed skin cancer or premalignant lesions by the age of 20, suggesting early onset, at least in this sample. Skin cancer might have been a factor in the evolution of dark skin among our ancestors, although it was probably not the only factor. Rees and Flanagan (1999) suggest that although skin cancer was a likely factor in the evolution of dark skin in the tropics, risks related to severe sunburn were probably more important.

Vitamin D Toxicity

Vitamin D is an essential nutrient for humans. With the exception of fatty fish, such as salmon, most foods are generally low in vitamin D. In the United States, we are used to associating vitamin D with milk, but in this case the vitamin had been added to the milk during processing; cow's milk is generally low in vitamin D. Most vitamin D in humans is obtained from a biochemical synthesis in the skin brought about by the action of UV-B (wavelengths between 280 and 320 nm), which converts 7-dehydrocholesterol into a substance known as **previtamin D**, which then converts to vitamin D (Robins 1991).

Too little vitamin D can cause a variety of problems, as discussed below. In terms of the adaptive value of dark skin under high UV radiation levels, Loomis (1967) suggest

that dark skin evolved to prevent toxic effects of producing too much vitamin D. High dosages of vitamin D are sometimes administered to people suffering from vitamin D deficiency, and in some cases an overdose can result in toxic levels of vitamin D, which can lead to excess calcium absorption, kidney failure, and death (Robins 1991). Loomis (1967) argues that dark skin evolved to protect against the possibility of vitamin D toxicity since dark skin prevents up to 95% of the UV radiation from reaching the deep layers of the skin, where vitamin D is synthesized.

While logical, this hypothesis must be rejected given measurements that show that humans do not synthesize toxic levels of vitamin D. Holick et al. (1981) examined the effect of artificial UV light on skin samples from light- and dark-skinned humans in order to determine the rate of previtamin D synthesis. They found that synthesis of previtamin D increases only up to a certain level, after which it remains at a plateau. After this point, 7-dehydrocholesterol converts into two inert substances rather than previtamin D. Therefore, vitamin D toxicity due to UV radiation is not possible, and the hypothesis is rejected.

Folate Photolysis

Several nutrients are subject to **photolysis**, chemical decomposition brought about by visible light. One such nutrient, **folate** (a compound belonging to the vitamin B complex group), has been implicated in the evolution of dark skin. Branda and Eaton (1978) found that folate concentrations in human blood plasma decreased significantly after brief exposure to UV light. In order to show that the same effect occurs in living humans, they further compared serum folate levels in 10 light-skinned patients who were undergoing therapeutic exposure to UV radiation with 64 healthy light-skinned people. The patients undergoing UV exposure had significantly lower serum folate levels than the controls, suggesting photolysis.

Branda and Eaton (1978) suggest that light-skinned individuals are likely to show folate deficiency in environments with high levels of UV radiation. Jablonski and Chaplin (2000, 2002) note that folate deficiency is clearly linked to differential survival and fertility. There is a link between folate deficiency and the incidence of neural tube defects, congenital malformations that affect the development of the nervous system. Two of these malformations, anencephalus and spina bifida, are more common in light-skinned populations and account for a notable percentage of infant deaths. It is also worth noting that Africans and African Americans, with darker skin, show lower levels of folate deficiency and neural tube defects. Folic acid prevents 70% of neural tube defects, and it seems reasonable to assume that low levels of folate can predispose individuals to neural tube defects during development of the embryo. Jablonski and Chaplin (2000, 2002) also note that there is a link between folate deficiency and differential fertility. Several studies have shown that induced folate deficiency leads to problems in sperm production and increased incidence of male infertility.

According to Jablonski and Chaplin (2000, 2002), folate photolysis was the major factor in the evolution of dark skin among early hominids in equatorial Africa. Those with lighter skin would be more likely to suffer from the photodestruction of folate, thus leading to increased mortality due to increased incidence of neural tube defects and decreased fertility due to the increased incidence of male infertility. Light-skinned individuals would thus be selected against because of differences in both survival and reproduction, while those with darker skin would be selected for because of the protective effect of dark skin on folate photolysis.

The folate photolysis hypothesis is supported by epidemiological and physiological data, and it seems reasonable to assume that folate deficiency was the major force shaping the evolution of dark skin in equatorial hominids. This does not mean, however, that other factors, such as sunburn and skin cancer, did not also influence overall fitness. Dark skin protects against UV radiation damage, which in turn can include both nutrient photolysis and damage to the skin and sweat glands.

Selection for Light Skin

The available evidence suggests a protective role of dark skin against the damaging effects of UV radiation in populations at or near the equator. The current distribution of human skin color shows that human populations that moved away from the equator, particularly those that moved far to the north, evolved lighter skin. The potential damage from UV radiation decreases as one moves away from the equator, but this means only that lighter skin *could* evolve; it does not explain why it did. To fully explain the current distribution of human skin color, we need a way to explain why light skin is found at distances farther from the equator. We can rule out random genetic drift since it is extremely unlikely that random variation in skin color in the absence of selection would lead to the observed high correlation of skin color and latitude.

Vitamin D Deficiency

The most widely accepted model for the evolution of light skin focuses on the role of insufficient UV radiation. UV radiation declines with increasing distance from the equator. This change means that the potential damage due to UV radiation also decreases, as shown by studies of latitudinal differences in skin cancer rates, among other findings reviewed above. For light skin to be favored in such environments, however, it is necessary to consider problems in survival and/or reproduction that would result from receiving too little UV radiation. Specifically, the idea here is that darker skin would be selected against (and hence lighter skin selected for) because reduced UV radiation leads to a deficiency in the production of vitamin D.

Loomis (1967) noted that nontropical regions receive low amounts of UV radiation during the year and suggested that individuals with dark skin would be at a selective disadvantage because they would suffer from a deficiency in vitamin D synthesis. The main effect of vitamin D deficiency is the childhood disease rickets, the defective growth of bone tissue. Severe rickets can obviously lead to reduced fitness, particularly given the active hunting and gathering life of our ancestors. Further, rickets can directly impact on differences in reproduction when pelvic bones are deformed during growth, thus leading to later complications and possible death during childbirth. The area of the pelvic inlet in women with rickets is only 56% that of healthy women, suggesting a greater incidence of difficulties during childbirth. Studies in the 1950s, performed prior to the increased use of vitamin supplements, showed that African American women had a much higher incidence of a deformed pelvis (15%) compared to European American women (2%) (Frisancho 1993). This suggests, but does not prove, a relationship with skin color as we must consider other genetic and/or environmental differences between the two ethnic groups. Nonetheless, this ethnic difference is consistent with the vitamin D deficiency hypothesis.

Dark skin reduces the amount of vitamin D synthesis, and it takes longer for previtamin D synthesis in dark skin. Holick et al. (1981) measured the rate of previtamin D

synthesis in skin samples exposed to the same amount of UV radiation as found at the equator and found that it took 30–45 minutes for lightly pigmented skin to reach a maximal level and 3–3.5 hours for dark skin to reach a maximal level. These findings support the vitamin D deficiency hypothesis, which argues that dark-skinned individuals would be at a selective disadvantage outside of the tropics.

Not everyone agrees. Robins (1991) argues against the vitamin D deficiency hypothesis. He notes that rickets is a disease that came about in more recent times, with the spread of civilization and cities, and tends to be found in polluted cities but to be rare in rural areas. Robins (1991) also notes that vitamin D can be stored in body tissues, allowing people to synthesize enough vitamin D during part of the year (summer) to last through times of lower UV radiation (winter). Also, while dark-skinned individuals are less effective at synthesizing vitamin D, they are still capable of producing enough even at higher latitudes to allow survival. Robins's main argument here is that differences in vitamin D synthesis between light- and dark-skinned people are relative, and the reduced rate of synthesis in the latter would not pose a selective disadvantage. He cites several studies suggesting that, outside of polluted urban areas, dark-skinned people would be able to produce enough vitamin D. As further evidence, Robins also points out that there is little evidence of rickets in skeletons from preindustrial Europe, raising the question of whether rickets was present in sufficient degree to constitute a selective factor. Others have contested Robins's arguments, noting that the evidence for high levels of vitamin D storage is lacking (Frisancho 1993). Beall and Steegmann (2000) note that skeletal analysis of rickets focuses on the most severe manifestations of low vitamin D levels, and lower values might also have biomedical significance. Finally, Jablonski and Chaplin (2000) analyzed UV radiation levels across the planet to estimate the expected levels of previtamin D at different latitudes. Their analyses support the vitamin D deficiency hypothesis by showing that medium and dark skin would be at a disadvantage at higher latitudes, being less likely to be able to synthesize adequate levels of vitamin D.

Cold Injury

While many anthropologists have accepted the vitamin D deficiency hypothesis as the most likely explanation for the evolution of light skin, others have suggested that variation in temperature might also have an impact (Post et al. 1975). The cold injury hypothesis proposes that heavily pigmented skin is more susceptible to damage, such as frostbite, and would therefore be selected against in cold environments. Since average annual temperature tends to vary with latitude (hottest near the equator), this model further suggests that the observed correlation of skin color and latitude actually reflects, to some extent, an underlying correlation with temperature (recall from Chapter 10 that latitude is often used as a proxy for temperature in studies of human adaptation).

There is some statistical evidence for the influence of temperature on skin color variation. Roberts and Kahlon (1976) found that annual temperature was significantly related to skin reflectance even after variation in latitude was taken into account. More direct support for the cold injury hypothesis comes from medical records of soldiers, particularly during the Korean War, where winter conditions were severe. For example, Ethiopian troops, primarily dark-skinned, had the highest rates of frostbite of all nationalities, with rates almost three times that of the U.S. forces. This comparison is crude because the U.S. forces included both white and black soldiers. However,

comparisons within the U.S. forces showed that frostbite among blacks was between four and six times that among whites, even after controlling for other factors (Post et al. 1975).

Physiological studies also suggest a relationship between pigmentation and susceptibility to cold injury. Post et al. (1975) cite a number of examples, including studies of piebald guinea pigs (having black and white–spotted skin). They froze black and white areas of the skin of anesthetized guinea pigs and found that cold damage was more severe in the darker skin. Cold injury was always more severe in the pigmented skin of the guinea pigs.

While some scientists have accepted the plausibility of the cold injury hypothesis (e.g., Robins 1991), others have not. Beall and Steegmann (2000) argue that black–white differences in cold injury might instead be due to known differences in the vascular response to cold and have nothing directly to do with variation in pigmentation. In other words, the cold injury hypothesis may be based on a spurious correlation. They also suggest that the guinea pig experiments are not directly relevant to human cold injury, primarily because the temperatures used on the guinea pigs were more extreme than typically found in likely human environments. Instead, they make the case, as do Jablonski and Chaplin (2000), that light skin evolved in response to vitamin D deficiency. Of course, these two models are not mutually exclusive. It is possible that both vitamin D deficiency and cold injury contributed to the evolution of light skin farther away from the equator. Although the evidence supporting the vitamin D hypothesis is strong, this does not preclude the possibility that differential cold injury was also a contributing factor. As is often the case in evolution, one's overall fitness reflects the net effect of phenotypic response in a given environment.

Sexual Selection

Another model also considers the evolution of light skin to be a consequence of past natural selection, but instead of relating light skin to environmental variation, this model rests upon the concept of sexual selection. First developed by Charles Darwin, sexual selection occurs when there is competition for mates or where there are preferences for mating with members of the opposite sex that have certain physical characteristics (Aoki 2002). A classic example of the latter is the preference of females in a number of species of birds and fish to choose brightly colored males as mates. Given this preference, there would be an advantage to having bright coloration, and any genes underlying this coloration would be selected for over time.

The sexual selection hypothesis as applied to human skin color rests on the assumption that human males prefer females with lighter skin color. Aoki (2002) suggests that if this assumption is correct, then the present-day geographic distribution of human skin color could be explained by a balance between natural selection and sexual selection. His model suggests that natural selection tends to favor darker skin but that (as discussed above) the intensity of this selection diminishes with increasing distance from the equator. Consequently, any preference for lighter-skinned mates would have its greatest impact when the counteracting natural selection for dark skin is lowest; that is, farther away from the equator. Aoki's model represents a balance between selection for dark skin and (sexual) selection for light skin.

What is the link between a preference for light skin and selection? Jones (1996) suggests that given a tendency for female skin color to darken with age, lighter skin might have acted as a signal for greater fecundity. However, this connection has not been

demonstrated, and as noted earlier, the relationship between skin color and age varies even among closely related groups (Williams-Blangero and Blangero 1991).

Although the sexual selection model seems reasonable, it rests upon the critical assumption that there was widespread preference for light skin in past human populations. Van den Berghe and Frost (1986) performed a cross-cultural analysis of ethnographic accounts and found preferences for light skin color in mates in 47 out of 51 societies, with males expressing this preference more often. There remains the question of whether these preferences, recorded for recent human societies, apply to our past as well. In other words, is a preference for light skin part of our species' evolutionary history, or does it represent the widespread adoption of culturally based preferences, particularly standards spread via colonialism? Although some authors argue that a preference for light skin existed historically throughout much of the world prior to European colonialism (Jones 1996, Aoki 2002), we of course have no evidence regarding mating preferences from earlier historic or prehistoric times. Perhaps the major problem with the sexual selection hypothesis is showing a link between mating preference and mating behavior. The hypothesis would be strengthened with a demonstration that mating preferences have a significant impact on the actual choice of a mate. Although the sexual selection hypothesis has its proponents (e.g., Aoki 2002), other anthropologists feel that although sexual selection is perhaps a contributing factor, the evolution of light skin has been affected more by the link between UV radiation and vitamin D production (e.g., Jablonksi and Chaplin 2002).

No Selection?

Another view on the evolution of light skin is that there is no selective advantage for light skin, either through natural selection or sexual selection. Brace (2000) and Brace and Montagu (1977) have argued that when humans moved farther away from the equator, selection for dark skin decreased. Over time, mutations for light skin arose; and in the absence of selection against them, these neutral mutations became more common. The underlying assumption in this model is that any mutations would interfere with the production of melanin and therefore lead to lighter skin.

Although this neutral mutation model has not received much support, the idea that light skin may not reflect a selective advantage has resurfaced with Harding et al.'s (2000) analysis of the *MC1R* gene. Using statistical methods to detect past natural selection from genetic diversity measures, they found evidence for strong selection in Africa but a lack of evidence for selection in European populations. Instead, they argue that patterns of *MC1R* diversity in Europe are consistent with neutral expectations, suggesting a lack of significant selection for light skin. However, other analyses of *MC1R* diversity do suggest a selective interpretation (Rana et al. 1999). In addition, it is not clear to what extent variation (and selection) for the *MC1R* gene is related to variation and selection in skin color. Variation in *MC1R* explains only part of the variation in skin color, and it is possible that other genes affecting skin color are influenced by adaptation (Harding et al. 2000).

OTHER ASPECTS OF HUMAN PIGMENTATION

In addition to skin color, anthropologists have studied variation in hair and eye color, although to a lesser extent.

Hair Color

Although hair color is an immediately noticeable phenotypic trait, less is known about the underlying causes of hair color variation in the human species.

Biology of Hair Color

Hair color, like skin color, is related to variation in melanin, which in this case is extruded into the hair from the hair follicles. Although the term *melanin* has been used previously, there are actually two different types of melanin: (*1*) **eumelanin**, the black–brown pigment found in all melanocyte-producing tissues, and (*2*) **pheomelanin**, a pigment that is most often reddish brown and found in mammalian hair (and chicken feathers) (Robins 1991). While skin color is influenced only by eumelanin, hair color is due to the relative amounts of eumelanin and pheomelanin. In people with black hair, eumelanin dominates and masks the pheomelanin. Other combinations of eumelanin and pheomelanin result in a range of hair color from dark brown to blonde. Red hair occurs when pheomelanin is more frequent, but the exact shade depends on the contribution of eumelanin (Brues 1977).

Like skin color, hair color can be measured in several different ways. Some studies have used standardized color scales (e.g., Braüer and Chopra 1980), while others have used reflectance spectrophotometry, measuring hair color at the crown of the head, side of the head at ear level, and (given sufficiently long hair) the hair ends (e.g., Little and Wolff 1981). When the latter is used, reflectance tends to increase with wavelength, with red hair showing a noticeable increase starting in the green range of the visual spectrum (see Fig. 11.8).

The genetics of hair color is not clearly understood, although it appears that it is a polygenic trait, where darker shades are in general dominant over lighter shades. One study observed hair color in 100 German families and estimated a heritability of 0.61, showing moderate genetic influence but also a fair amount of environmental effect (Braüer and Chopra 1980). Hair and skin color may be related to some extent, particularly in individuals with red hair, who are generally paler and more prone to freckles (Little and Wolff 1981).
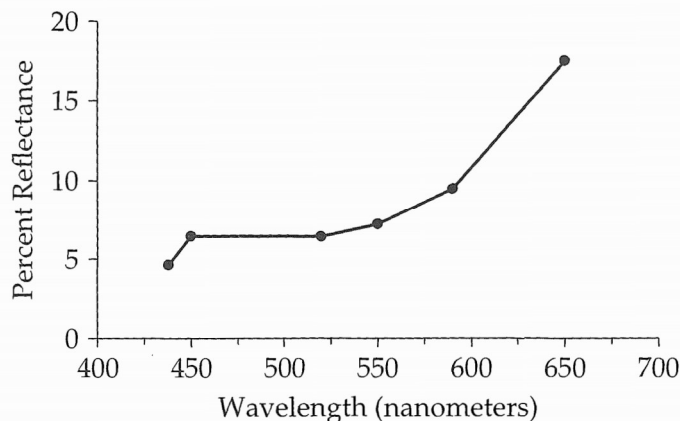

Figure 11.8. Percent reflectance of hair measured at the crown for 33 redheaded women using the Photovolt machine. The sharp increase in reflectance in the green range of the visible spectrum (around 550 nm) is characteristic of people with red hair. (Data from Little and Wolff 1981.)

Recently, the genetics of red hair color has become better understood in terms of variants of the *MC1R* gene and their effect on a switch in production from eumelanin to pheomelanin within melanocytes (Rees and Flanagan 1999, Harding et al. 2000). Three different mutations (Arg151Cys, Arg160Trp, and Asp294His) are associated with red hair. In one study, every person who had at least two of these mutations had red hair. Other studies confirmed this, showing that most redheads had two of the three mutations or were homozygous for one of them (Rees and Flanagan 1999).

Variation in Hair Color

One of the more obvious characteristics of hair color is how it changes with age. In general, hair color darkens with age (Robins 1991), particularly around puberty, which may reflect some hormonal effect (Brues 1977). Interestingly, lighter shades are sometimes found in young children even when the typical adult color is very dark; the best example is the observation that a number of Australian Aborigine children have blondish hair, although most adults are dark. Hair color can also lighten with increased age, producing a gray color that results from the mixture of pigmented and nonpigmented (white) hairs.

Outside of Europe, most human populations tend to have darker shades of hair color. The frequency of blond hair tends to be highest in northwestern Europe. However, there is variation even within smaller geographic areas, such as the noticeable cline showing an increasing frequency of blond hair from southern to northern Italy (Harrison et al. 1988). Red hair is also confined primarily to Europe, having the highest frequency in northern Europe, particularly Scotland and Wales (Little and Wolff 1981), and appears to be associated with a series of specific mutations of the *MC1R* gene (Rees and Flanagan 1999).

The cause of these geographic distributions is not clear. Some have suggested an adaptive value of dark hair color in hot climates because of the ability of darker hair to absorb heat. Since hair tends to act as an insulator, dark hair can absorb and then radiate heat that otherwise might be harmful in a hot climate (Brues 1977). This hypothesis has not been confirmed, but even if correct, it still leaves unexplained the higher frequency of lighter hair colors in Europe. Perhaps we are seeing an example of relaxed selection, such that dark hair is not as necessary in colder climates, thus allowing other shades to increase due to random genetic drift.

Eye Color

Eye color is perhaps even more complicated in terms of biology and measurement.

Biology of Eye Color

Eye color can be assessed using visual inspection, comparison with color scales, or comparison with glass eyes of given colors. In any case, there is a great deal of subjectivity in resolving between different shades, such as light versus medium brown. Measurement is complicated further by the numerous factors that influence eye color, including distribution of melanin and optical scattering. Eye color is to a large extent determined by the amount of melanin present in the anterior layer and stroma of the iris. Brown and dark brown eyes result from heavy concentrations of melanin. Blue eyes are not due to blue pigment but instead result from a lack of pigment combined with optical scattering; in the absence of melanin (or very little), the shorter blue wavelengths of light are scattered, giving rise to a bluish color much in the same way that the sky appears blue. Other shades, such as hazel and green, reflect a balance between the amount of melanin and

optical scattering (Robins 1991). Mixed colors (each eye has a different color) sometimes occur, and it appears to be more common among females, suggesting a sex-linked effect (Brues 1977).

There is some correlation of eye color with skin and hair color, suggesting that there is some pleiotropic effect related to pigmentation. Among redheaded women, for example, those with lighter eye color tend to have lighter skin color and lighter hair color (Little and Wolff 1981).

Eye color is typically considered to be inherited. However, a study of 100 German families (Braüer and Chopra 1980) estimated a heritability of 0.8, indicating that 20% of the variation in eye color did not reflect inheritance in this sample. It is possible that some of this variation is due to problems in measurement and coding of eye color. The genetics of eye color is sometimes taught in school as a simple Mendelian trait affected by a single locus, where the "brown-eyed gene" is dominant over the "blue-eyed gene." Such statements are only rough approximations, and although the genetics of eye color are still poorly understood, it is likely that eye color is polygenic to some extent. It is true that darker eye color does tend to be dominant over lighter eye color but only in a general sense if only "brown" and "blue" categories are used and other variations are lumped together (Brues 1977, Robins 1991).

Variation in Eye Color

Newborns typically have blue or violet–blue eyes, which will often darken quickly with age. Infants with violet–blue eyes tend to develop brown eyes (Brues 1977). Some studies have shown some darkening later in life (Robins 1991).

Although light-colored eyes are found to a limited degree across the world, they tend to be very common only in some European populations (Brues 1977). The reason for this geographic distribution is not clear. Perhaps the distribution of blue and other light-colored eyes reflects, to some extent, variation in overall pigmentation since it is light-skinned populations in which light-colored eyes are most common. Some suggestions have been made that eye color has undergone a different pattern of selection. For example, one hypothesis was that light-colored eyes had enhanced ability to perceive shorter wavelengths, which would presumably be adaptive in early hunting and gathering populations living in glacial Europe with foggy weather. However, this hypothesis was tested, and no such differences in perception were found (Robins 1991). Other suggested evolutionary adaptations of light-colored eyes (see Brues 1977) have also not been supported or rely on unreasonable assumptions. To date, there is no accepted model of the evolution of eye color.

CHAPTER SUMMARY

One of the most visible signs of human biological variation is the difference in skin, hair, and eye color. Skin color is influenced primarily by variation in the melanin pigment and is best measured using reflectance spectrophotometry, which measures the percentage of light reflected off of the skin at different wavelengths. The genetics of skin color is still not completely known, but many studies show a high heritability. Recent studies have shown that variation in the gene MC1R is associated with human skin color variation. Females are generally lighter than males by a small amount, and there is some evidence for slight darkening with age, particularly among adolescent females.

Human skin color shows a strong environmental correlation. Skin color is darkest in populations at or near the equator and is progressively lighter with increasing distance from the equator. The geographic distribution of skin color matches that of UV radiation, which is strongest at the equator and diminishes farther away from the equator. Skin color is somewhat darker in the Southern Hemisphere than in the Northern Hemisphere at equivalent latitude, paralleling the known incidence of UV radiation.

A number of models have been proposed to explain the evolution of human skin color differences. Explaining the evolution of human skin color requires an explanation for dark skin at or near the equator and for light skin in populations farther away from the equator. Several explanations have been offered for the origin and maintenance of dark skin in equatorial populations, all focusing on dark skin protecting against injurious effects of UV radiation, including folate photolysis, sunburn, and skin cancer. The evolution of lighter skin farther away from the equator has usually been explained by lowered levels of UV radiation causing a reduction in vitamin D synthesis, which in turn can lead to a number of medical problems, including rickets. According to this model, light skin evolved as an adaptation to reduced levels of UV radiation. Some have suggested a relationship between latitude, temperature, and skin color, noting that light skin is less susceptible to frostbite than dark skin. Others have suggested that light skin evolved due to sexual selection and a preference for mates with lighter skin color. It is likely that the evolution of human skin color at all latitudes reflects a balance between several selective forces.

Hair color is influenced by both the blackish brown eumelanin and the reddish brown pheomelanin, leading to a range in hair color from dark to light and including various red shades. Most human populations have dark hair, and the greatest amount of variation tends to occur in European populations, although the evolutionary reasons are not known. Eye color is caused by variation in melanin in the iris combined with optical effects such that eyes with little pigment appear blue. As with hair color, European populations show the greatest degree of variation in eye color.

SUPPLEMENTAL RESOURCES

Jablonski N G and Chaplin G (2000) The evolution of human skin coloration. *Journal of Human Evolution* 39:57–106.

Jablonski N G and Chaplin G (2002) Skin deep. *Scientific American* 287:74–81. A short introduction to current views on the evolution of human skin color.

Relethford J H (1997) Hemispheric difference in human skin color. *American Journal of Physical Anthropology* 104:449–457.

Jablonski and Chaplin (2000) and Relethford (1997) review the evidence for the geographic distribution of human skin color. Jablonski and Chaplin (2000) includes a complete review of different models for the evolution of human skin color.

Robins A H (1991) *Biological Perspectives on Human Pigmentation*. Cambridge: Cambridge University Press. A thorough review of human pigmentation focusing on skin color.