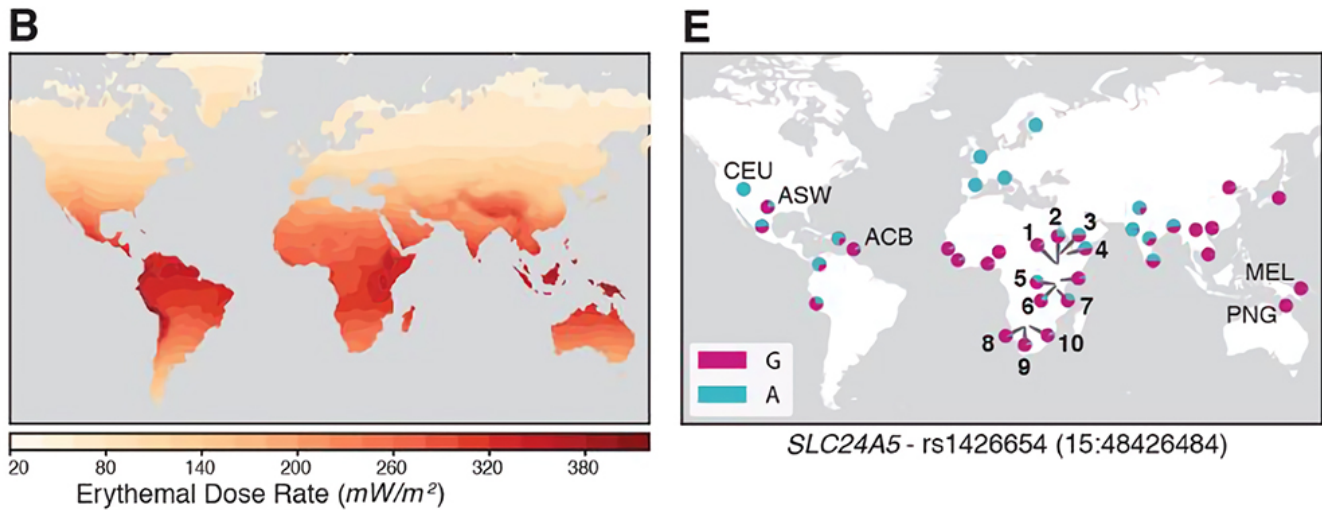




HOW TO USE THIS RESOURCE

Show the following figure and caption to your students. The accompanying Student Handout provides space below the image caption for Observations, Notes, and Questions and space next to the “Background Information” for Big Ideas, Notes, and Questions. The “Interpreting the Graph” and “Discussion Questions” sections provide additional information and suggested questions that you can use to prompt student thinking, increase engagement, or guide a class discussion about the characteristics of the graph and what it shows.



Caption: Figure B shows global measurements of erythemal dose rates. Higher erythemal dose rates indicate more exposure to ultraviolet (UV) radiation from sunlight. Figure E is based on DNA samples from human populations around the world. Each circle on the map represents a different population, and the colors in the circle indicate the proportion of that population with either the ancestral allele (“G”) or the derived allele (“A”) of the gene SLC24A5. The study focused on 10 African populations (Labels 1–10). Data from these populations were compared to published data from other populations, including Melanesian (MEL), Papua New Guinean (PNG), European ancestry (CEU), African-American Southwest US (ASW), and African Caribbean in Barbados (ACB) populations.

BACKGROUND INFORMATION

Height, weight, eye color, skin color, and many other traits are affected by genes, gene regulation, and the environment. The genetic factors contributing to variation in these traits are often unclear and thus the subject of much research. Results from this research have improved our understanding of how traits are formed, related medical conditions, and human evolution.

In this study, scientists investigated the genetic factors contributing to skin color differences, particularly within African populations. Prior to this study, research on the genetics of skin color included mostly people of European or Asian descent. The scientists collected DNA samples from 1,570 people of African descent and obtained more genetic data from other populations around the world. After comparing many DNA sequences, they identified four genomic regions that were significantly correlated with skin color differences. The most significant correlation between variation in skin color and variation in genetic sequence in Africans was at the SLC24A5 gene. This gene codes for a membrane transport protein that helps cells called melanocytes produce the pigment melanin. Melanin gives skin its color and protects against UV radiation from the sun. Although too much UV radiation can damage the skin, our bodies use exposure to small amounts of UV radiation to make vitamin D. This process is important for healthy bones, especially for people with diets low in vitamin D.

Changing just one nucleotide in the *SLC24A5* gene can change the way the protein functions. This type of genetic variation is called a single-nucleotide polymorphism, or SNP. In this study, the scientists examined one SNP of *SLC24A5* that is strongly linked to skin color differences. The ancestral form of *SLC24A5* for this SNP is a guanine (G) nucleotide. A new allele, or version, of *SLC24A5* later arose in some groups of people due to a mutation that changed the guanine to adenine (A). This mutation disrupts the function of the protein coded by *SLC24A5*, which prevents melanocytes from producing as much melanin. Although both the ancestral (G) allele and derived (A) allele exist today, these alleles have different frequencies in different populations.

INTERPRETING THE GRAPH

Comparing Figures B and E reveals that there are generally higher frequencies of the ancestral (G) allele in regions with *higher* UV exposure. Similarly, there are generally higher frequencies of the derived (A) allele in regions with *lower* UV exposure.

For example, populations of West African descent have higher frequencies of the ancestral (G) allele. Individuals with this allele typically have darker skin because their cells produce more melanin. This melanin protects the individuals from the high levels of UV near the equator, while still allowing enough UV for sufficient vitamin D production. Note that some populations, such as those of East Asian descent, also have high frequencies of the ancestral (G) allele even though their skin colors aren't typically as dark. Other genetic variations — some of which the study also investigated (see the other panels of Figure 1 in the [original paper](#) for examples), but many of which have yet to be identified — may contribute to the lighter skin colors in these populations.

On the other hand, populations of European or Central Asian descent have higher frequencies of the derived (A) allele. Individuals with this allele typically have lighter skin because their cells produce less melanin. This is likely because these individuals live in regions with more limited UV exposure, which limits vitamin D production. Mutations that reduce melanin production, such as the mutation in the derived allele, can thus increase vitamin D production to healthy levels.

Using genetic analyses, the scientists estimated that the derived (A) allele first appeared in Eurasia 29,000 years ago. Within the last 10,000 years, this allele was likely introduced into African populations by humans migrating from West Eurasia to Africa. However, certain populations in eastern and southern Africa have higher frequencies of the derived allele than expected, possibly due to strong natural selection for the allele. It remains unclear why the derived allele may be advantageous in these populations.

Teacher Tip: Prompt your students to explain the parts of the graph as applicable:

Figure B:

- **Graph type:** Heat map
- **Legend:** Erythemal dose rate in milliwatts per square meter (mW/m^2). Erythemal dose, a measure of the amount of UV radiation required to temporarily turn fair skin red, is used as a proxy for average UV exposure in a region. Darker colors indicate a higher erythemal dose rate and, thus, higher UV exposure.

Figure E:

- **Graph type:** Map with pie charts. Each pie chart corresponds to a different population.
- **Legend:** In each pie chart, purple represents the proportion of the population with the ancestral (G) allele of *SLC24A5*, and blue represents the proportion with the derived (A) allele.
- **Pie chart labels:** Labels indicate specific populations that the scientists had data from. (Note that not all of the populations are labeled.) Labels 1 to 10 correspond to the 10 African populations that were the focus of the study (1: Ethiopia Nilo-Saharan, 2: Ethiopia Omotic, 3: Ethiopia and Tanzania Cushitic, 4: Ethiopia Semitic, 5: Tanzania Nilo-Saharan, 6: Tanzania Hadza, 7: Tanzania Sandawe, 8: Botswana Bantu, 9: Botswana San/Bantu admixed, and 10: Botswana San). The remaining labels represent other populations

with published genomic data, including MEL (Melanesian), PNG (Papua New Guinean), CEU (European ancestry), ASW (African-American Southwest US), and ACB (African Caribbean in Barbados).

- **Bottom label:** The name of the gene (*SLC24A5*), the SNP identification number (rs1426654), and the SNP's chromosomal location (15:48426484).

DISCUSSION QUESTIONS

- What patterns do you notice in Figure B, the erythemal dose rate map? How about in Figure E, the map with the pie charts?
- Compare the patterns in the two maps. What similarities and differences do you observe?
- In Figure E, what do the colors in each pie chart represent? Describe the typical genetic and phenotypic differences between the individuals represented by each color.
- Explain the difference between an “ancestral allele” and a “derived allele.”
- Which allele of *SLC24A5*, ancestral (G) or derived (A), would you expect to find in an individual of Scandinavian descent? Why?
- Which regions of the world do individuals with darker skin color typically descend from? Lighter skin color? Use evidence from the figure to support your answer.
- High frequencies of the ancestral (G) allele can be found in populations of East Asian descent. However, these populations often have lighter skin colors than populations of African descent do. How can you explain these differences?
- What health recommendations would you give to a person with darker skin color living in a low-UV area? What about a person with lighter skin color living in high-UV area? Why?
- One definition of evolution is “a change in allele frequency.” Based on this definition, do you think that humans are evolving? Provide evidence from the figures.
- Do you think skin color is an adaptation? If so, what might be its selective pressures and selective advantages? Provide evidence from the figures.
- How might modern cultural practices affect the context for natural selection on skin color? Have certain selective pressures on skin color changed over time?
- How might human migration affect the patterns shown in Figure E?
- Some mutations in *SLC24A5* have been linked to medical conditions like melanoma (a type of skin cancer) and albinism. Why do you think this is?
- The deletion of a large portion of the *SLC24A5* gene causes an albino phenotype in white Doberman pinschers. What does this observation suggest about the conservation of gene function over the course of evolution?

KEY TERMS

allele, human migration, melanin, single-nucleotide polymorphism (SNP), skin pigmentation, ultraviolet (UV) radiation

SOURCE

Figure 1b and 1e from:

Nicholas G. Crawford, Derek E. Kelly, Matthew E. B. Hansen, Marcia H. Beltrame, Shaohua Fan, Shanna L. Bowman, Ethan Jewett, et al. “Loci associated with skin pigmentation identified in African populations.” *Science* 358, 6365 (2017). <https://doi.org/10.1126/science.aan8433>.

An annotated version of the article is also available from [Science in the Classroom](#).

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