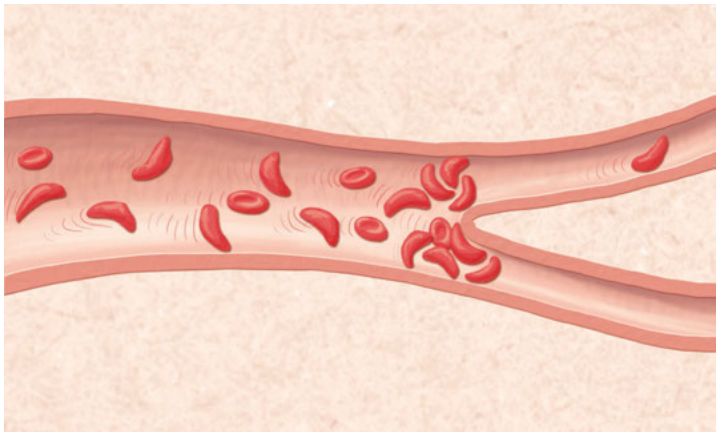
Teacher Tune-up

Quick Content Refresher for Busy Professionals

How do genes that cause disease stay in populations?

Gregor Mendel's findings allowed scientists to predict how genes are passed from one generation to the next, but as we integrate our understanding of genetics with the theory of evolution, we are left with a looming question: how do genes that cause disease stay in populations? Based on Darwin's theories, genes that harm an organism's chances of survival and reproduction would disappear from populations as they wouldn't be passed along; and genes that provide better chances would spread through the population. Diseases like sickle cell disease, cystic fibrosis, and Huntington's are all single-gene diseases that still persist in populations. In recent years, scientists have come up with some interesting ideas as to why.

Sickle cell disease is caused by a mutation to the hemoglobin gene, which affects the protein that carries oxygen and helps give red blood cells their distinctive round shape. The mutated gene causes the red blood cells to have a concave, abnormal shape, somewhat like a sickle, hence the name. These abnormal cells can get rigid and sticky, clogging blood vessels and reducing oxygen flow to the body.



Sickle cell disease shows a pattern of inheritance known as co-dominance: a recessive sickle cell allele paired with a dominant normal allele will lead to the production of both sickle-shaped cells and normal-shaped cells.

While having two copies of the sickle cell gene leads to a serious disease, being a carrier of the gene, or just having just one copy, is not as problematic. In fact, having one copy turns out to be a benefit for people living in places with high rates of malaria, as having some sickle-shaped cells seems to prevent malaria from taking hold in the body.

This phenomenon helps explain why the gene persists in populations that live where malaria exists – if you are a carrier, you are more likely to survive malaria, and thus canpass on the gene to your offspring. Rates of sickle cell trait, or having just one sickle cell allele, are highest in populations who have ancestors from places like Sub-Saharan Africa, South Asia, the Middle East, and Southern Europe, all locations where malaria is prevalent. In environments without malaria, rates of sickle cell disease are very low, as there is no selective advantage to being a carrier, and there is a clear disadvantage to having two copies of the gene.

https://vimeo.com/251539516

A similar pattern may exist for cystic fibrosis, which is found in higher rates in populations with ancestors from Europe. While scientists are still working on their research, early findings suggest that being a carrier for cystic fibrosis may have provided

some resistance to cholera, a disease that was common in Europe many years ago. Since cholera is no longer a high risk in Europe, scientists predict that eventually genes for cystic fibrosis will decrease in populations, since there is no longer any advantage to being a carrier.

Other genetic diseases such as Huntington's, which leads to the death of brain cells, persist in the population for different reasons. The gene for Huntington's disease is a dominant one, which means that if one of your parents has the allele, you have a 50% chance of having the allele, and thus a 50% of getting the disease. Why does this devastating disease persist in the population? One reason may be that having this gene doesn't tend to affect a person's chances of reproduction, because the disease affects adults later in life. By the time an adult is afflicted with Huntington's disease, they have likely already passed on the gene to their children.

Look Out! Common Student Misconceptions

One common misconception among students is that a disease such as sickle cell is a "black" disease, and that genetic diseases are associated with race. Race is not a genetic trait, but rather a cultural construct. While skin color is determined by genes, knowing someone's skin color doesn't tell much about any of their other genes.

For example, someone who is black may have ancestors from a region in Africa where malaria is not prominent, and thus he or she will not be more likely to carry the sickle cell gene. People whose genealogy traces back to regions of the world such as the Middle East, Southern Europe, and South Asia are not black, but have an elevated risk of carrying the gene for sickle cell because they come from regions where malaria was endemic. A person's genetic make-up may be influenced by their ancestor's environment, but skin color is insufficient to tell us much about his or her genes. Mendel's Law of Independent Assortment reminds us that genes are inherited independently of one another, so a gene for skin color is unlikely to be a strong index of other genetic traits.

Geographical ancestry, however, may indeed have an effect on genetic traits, as seen in the examples of sickle cell disease and cystic fibrosis. Helping students distinguish between geographical ancestry, skin color, and modern notions of race will help them appreciate that race is not a meaningful concept in genetics.