Heterozygous Advantage & Sickle Cell Anemia

Red blood cells are able to transport oxygen because they are filled with a protein called hemoglobin, which picks up oxygen in the lungs and drops it off where it is needed in tissues and organs. A mutated version in one of the hemoglobin genes leads to Sickle Cell Anemia by changing the hemoglobin protein in such a way that it tends to clump up into long chains inside red blood cells. Instead of maintaining the usual flexible disc-like shape that enables them to squeeze through even the tiniest blood vessels, the red blood cells of people with the disease twist into stiff crescents that are not efficient at transporting oxygen. "Sickled" red blood cells can clog small blood vessels, preventing oxygen from making it to certain parts of the body. The condition is life-threatening.

In the United States, one in every 500 African-American births and one out of every 1,000 to 1,400 Hispanic births is affected by Sickle Cell Anemia. Another two million Americans carry the sickle cell trait. As devastating as the disease can be, it turns out there is a reason Sickle Cell Anemia is so common and has NOT been "weeded out" of the human population. Usually a DNA change that causes a serious disease quickly gets pushed out of a population's gene pool. But researchers have found that the version of the gene that causes Sickle Cell Anemia has been around for thousands of years.

- That observation, and the fact that this version is mainly found in people with ancestors who lived relatively recently in Africa, the Mediterranean, India, or the Middle East, led scientists to wonder if the Sickle Cell Anemia-causing version of the gene offers some kind of benefit to people living in those regions.
- That benefit turned out to be resistance to malaria. Malaria is caused by parasites that multiply inside of human red blood cells. Because the disease can only be transferred from person to person by mosquitoes, it is confined to areas of the world where the insects thrive.
- Every year malaria infects more than 300 million people and kills more than a million, particularly young children. Carriers of the sickle cell trait are to a large extent resistant to malaria. Compared to non-carriers, they have approximately 1/10 the risk of dying from infection by the most deadly species of malaria parasite. Nevertheless, carriers are not completely protected from the disease and experts recommend that they still take precautions against malaria.
- Over the years, carriers living in malaria-ridden locales would have had a survival benefit compared to non-carriers, allowing them to live longer and have more children.
- This benefit is what evolutionary biologists call "heterozygote advantage," and it explains why the sickle cell trait has persisted in areas where malaria is common. The price for the carriers' advantage, though, is that some of their children are born with Sickle Cell Anemia.

L. Carnes