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Association of Pediatric Hematology/Oncology Nurses

A HANDBOOK FOR FAMILIES

Sickle Cell Disease

HEMATOLOGY SERIES

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SICKLE CELL DISEASE

A Handbook for Families

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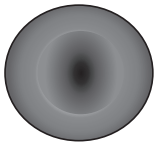
What is sickle cell disease?

Sickle cell disease is an inherited blood disorder that affects the ability of red blood cells to carry oxygen throughout the body. Normally, red blood cells are round and bendable. Sickle cell disease causes red blood cells to change into a curved, or sickle, shape. These sickle-shaped red blood cells stick together, blocking blood flow to the hands, feet, joints, bones, and major organs. When blood flow to these areas is decreased, the tissues are not getting enough oxygen, causing pain and other problems. To really understand how sickle cell disease affects the body, it helps to understand a little more about blood.



Red blood cell

Sickled red blood cell

What is blood?

Blood is a mixture of cells, proteins, and a watery substance called plasma. The cells in blood are made in our bone marrow, the spongelike center of some of the bones in our skull, spine, pelvis, and ribs. Like water being pumped through a hose, our blood is pumped by our heart through small tubes called blood vessels. There are two types of blood vessels: arteries and veins. The arteries carry blood away from the heart and bring oxygen to the body's tissues. The veins then return the oxygen-poor blood from the tissues back to the heart.

Blood has many different functions. It carries nutrients and gases throughout the body to our tissues and organs, and removes waste products from these tissues and organs so that the waste can be removed from the body. Blood also helps fight infection and heal wounds.

Blood has four major components:

- *Plasma*—a yellowish liquid that is mostly water. Plasma acts as a river, helping the white blood cells, red blood cells, and platelets flow freely through the blood vessels. It also carries nutrients, hormones, proteins, and waste products around the body.
- *White blood cells*—the fighter cells, also known as leukocytes. White blood cells, which are part of the immune system, protect the body from infection and disease.
- *Platelets*—the component that helps stop bleeding. Platelets form a scab after skin or tissue has been injured.

- *Red blood cells*—making up the majority of the cells in our blood, these small cells, also known as erythrocytes, are slightly flattened, so they look like a doughnut with the hole filled in. The function of red blood cells is to carry oxygen, carbon dioxide, and nutrients throughout the body. Red blood cells contain a protein called hemoglobin, which carries oxygen through the body. The red blood cells pick up oxygen in the very small vessels of the lungs and carry it to the body's tissues and organs. The tissue cells use the oxygen and create carbon dioxide, a waste product. After red blood cells drop off oxygen at the tissues, they pick up carbon dioxide and carry it back to the lungs to be exhaled from the body.

How does sickle cell disease affect the blood?

Red blood cells normally contain hemoglobin A, or adult hemoglobin. In sickle cell disease, the child inherits an abnormal gene for the production of hemoglobin, which is called a *gene mutation*. This gene mutation causes the body to make hemoglobin S, or sickle hemoglobin, instead of hemoglobin A. Sickle hemoglobin is not very efficient at carrying oxygen. Under certain conditions in the body, such as fever and dehydration, the red blood cells change into a rigid sickle shape, like a crescent moon. When this happens, the sickle-shaped cells get caught on each other and on the blood vessel walls, instead of flowing easily through the vessels like soft, round, normal blood cells. When these sickle-shaped cells become caught on each other, they block the flow of blood through that vessel so the tissues cannot get fresh, oxygenated blood. This is what causes pain, the most well-known symptom of sickle cell disease. However, this lack of oxygen is also responsible for many other problems, which will be explained later.

Red blood cells that contain hemoglobin A live for almost 120 days. However, because they can sickle and become rigid, cells containing hemoglobin S live only 10–20 days before breaking apart. This breaking apart of red blood cells is called *hemolysis,* and the result is a low hemoglobin level in people living with sickle cell disease.

Who gets sickle cell disease?

Sickle cell disease is a genetic disorder, meaning it can be inherited from a parent who carries the gene mutation for sickle cell disease. Many people associate sickle cell disease with Africans. Although it is very common in Africa, sickle cell disease actually began in four areas of the world with a high incidence of malaria: Africa, India, the Mediterranean, and the Middle East. It is believed that the sickle-cell mutation developed as a way to protect people living in these areas from malaria. Throughout the years, intermarriage between ethnicities and population migration to other countries have made sickle cell a global disease. Today, there are people with sickle cell disease living in almost every country.

How is sickle cell disease inherited?

Sickle cell disease is inherited in the same way that many of our genetic traits, such as eye color, are inherited from our parents. Sickle cell disease is a recessive condition that occurs when a child inherits the sickle cell gene from both parents, one from the mother and one from the father. If a child only inherits one sickle cell gene from either parent, then he or she will have sickle-cell trait and be referred to as a "carrier." People who live with sickle-cell trait do not experience the symptoms or problems that those who live with sickle cell disease experience.