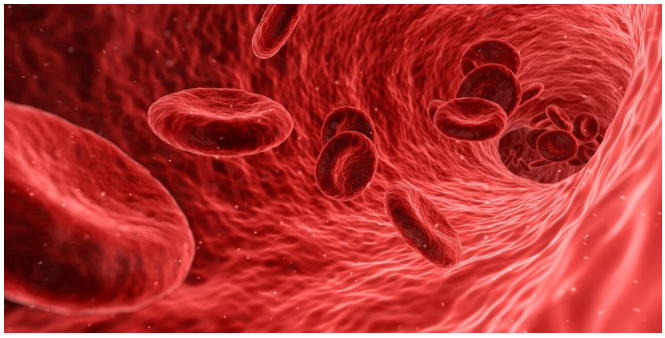
Understanding sickle cell disease

10 September 2021, by Laurel Kelly



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Sickle cell disease, which is a group of inherited red blood cell disorders, affects approximately 100,000 people in the U.S., according to the Centers for Disease Control and Prevention.

September is National Sickle Cell Awareness Month, which makes this a good time to learn about two of the most common types of sickle cell disease: sickle cell anemia and thalassemia.

Sickle cell anemia

Sickle cell anemia is caused by a mutation in the gene that tells your body to make hemoglobin, the iron-rich compound that makes blood red and enables red blood cells to carry oxygen from your lungs throughout your body. In sickle cell anemia, the abnormal hemoglobin causes red blood cells to become rigid, sticky and misshapen. Both mother and father must pass the defective form of the gene for a child to be affected.

If only one parent passes the sickle cell gene to the child, that child will have the sickle cell trait. With one normal hemoglobin gene and one defective form of the gene, people with the sickle cell trait make both normal hemoglobin and sickle cell hemoglobin. Their blood might contain some sickle cells, but they generally don't have symptoms. They're carriers of the disease, however, which means they can pass the gene to their children.

Signs and symptoms of sickle cell anemia usually appear around 5 months of age, and they vary from person to person and change over time. Signs and symptoms can include anemia, episodes of pain, swelling of the hands and feet, frequent infections, delayed grown or puberty, and vision problems.

Management of sickle cell anemia usually is aimed at avoiding pain episodes, relieving symptoms and preventing complications. Treatment can include medications and blood transfusions. For some children and teenagers, a stem cell transplant might cure the disease.

Thalassemia

Thalassemia also is caused by a mutation in the gene that tells your body to make hemoglobin, and the mutations are passed from parents to children.

Hemoglobin molecules are made of chains called alpha and beta chains that can be affected by mutations. In thalassemia, the production of either the alpha or beta chains are reduced, resulting in either alpha-thalassemia or beta-thalassemia. In alpha-thalassemia, the severity of the disease depends on the number of gene mutations inherited. The more mutated genes, the more severe the disease. In beta-thalassemia, the severity of the disease depends on which part of the hemoglobin molecule is affected.

Because there are several types of thalassemia, the signs and symptoms depend on the type and severity of the condition. Signs and symptoms can include fatigue, weakness, pale or yellowish skin, facial bone deformities, slow growth, abdominal swelling, and dark urine.

Mild forms of thalassemia don't need treatment. For moderate to severe thalassemia, treatment can include frequent blood transfusions, chelation therapy and stem cell transplant.

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