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My46 Trait Profile

Sickle cell disease

Sickle cell disease is a rare condition that affects the red blood cells and is characterized by chronic anemia (i.e., low blood count), frequent infections, and episodes of pain. It is caused by mutations in the HBB gene that encodes a protein called beta-globin.

Characteristics of Sickle cell disease

Sickle cell disease (SCD) is one of a group of conditions that affect hemoglobin, a molecule that is responsible for delivering oxygen to various parts of our body. SCD is characterized by chronic anemia (i.e., low blood count), frequent infections, and episodes of pain in different body areas such as the bones, kidneys, joints and lungs. Individuals with SCD may also develop an enlarged spleen due to damaged red blood cells accumulating in the spleen, and are also at risk for stroke. Babies born in the United States are screened for SCD by newborn screening.

Diagnosis/Testing

Individuals with SCD have changes or mutations in the HBB gene that encodes the beta-globin protein. This protein, along with other globin proteins, makes up a molecule called hemoglobin. Hemoglobin is found in red blood cells and is responsible for carrying oxygen throughout the body. The red blood cells of most individuals contain mainly hemoglobin A. Individuals with SCD have mostly hemoglobin S, which is also called “sickle hemoglobin” because of the “sickle” shape of the red blood cells. Because of their sickle shape, they cannot hold onto oxygen as well as normal red blood cells. The abnormal shape also makes it is difficult for them to pass through blood vessels, thus they can get stuck resulting in episodes of severe pain. Additionally, sickle red blood cells break down earlier than normal red blood cells and this can lead to anemia.

Management/Surveillance

Management of individuals with SCD often involves regular blood transfusions, physical exams, chest x-rays, abdominal ultrasounds, head ultrasounds (an ultrasound test uses sound waves to image tissues and organs such as the brain), and blood tests to monitor the progression of the condition. Some individuals with SCD take medication (e.g., hydroxyurea) that helps to reduce the number of sickled red blood cells in their bodies, and thus reduce the frequency of symptoms. It is important for individuals with SCD to stay hydrated, avoid extreme temperatures, and stay current with their immunizations in order to protect against harmful infections. It is recommended that young children with SCD take daily doses of antibiotic medication (e.g., penicillin prophylaxis) to also help prevent infections.

Mode of inheritance

SCD is inherited in an autosomal recessive pattern. This means that an individual has to inherit two HBB mutations (i.e., one from each parent) to be affected with SCD. If both parents are carriers of a HBB mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with SCD. Babies born in the United States are screened for SCD by newborn screening.

Risk to family members

Parents of a child with SCD are carriers of SCD. If a sibling of a child with SCD is unaffected, he/she has a 2 in 3

(or 66%) chance of being a carrier of SCD.

Special considerations

Most carriers of SCD, also referred to as sickle cell trait, do not have any symptoms of SCD. However, in rare cases, some carrier individuals may experience pain when exposed to low oxygen levels in the air (e.g., when mountain climbing), increased pressure in their surroundings (e.g., when scuba diving) or in cases of dehydration.

Resources

Centers for Disease Control and Prevention: Sickle Cell Disease
http://www.cdc.gov/ncbddd/sicklecell/index.html
Genetics Home Reference: Sickle cell disease
http://ghr.nlm.nih.gov/condition/sickle-cell-disease
Sickle Cell Disease Association of America, Inc.
http://www.sicklecelldisease.org
American Sickle Cell Anemia Association
http://www.ascaa.org
About Sickle Cell Disease
http://www.sicklecellinfo.net
Medical Home Portal: Sickle cell disease
http://www.medicalhomeportal.org/newborn/sickle-cell-disease

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