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What is the test for sickle cell anemia

Sickle cell anemia is a genetic disorder that causes abnormal hemoglobin production, leading to irregularly shaped red blood cells. Hemoglobin S test results can detect the presence of this abnormal protein in a blood sample and identify gene mutations associated with sickle cell disease. Normal hemoglobin A makes up most of the hemoglobin found in adult red blood cells, while fetal hemoglobin F is present before birth and gradually replaced by Hb A after birth. Inheriting mutated genes can result in abnormal hemoglobins like Hb S and Hb C. Sickle cell trait carriers typically don't experience symptoms but can pass the mutation to their children. The abnormal shape of red blood cells causes oxygen transport issues, reduced lifespan, and increased risk of complications. Testing for sickle cell disease (SCD) or sickle cell trait is used for various purposes, including newborn screening, carrier screening, general screening, and diagnosis. Newborn screening is mandatory in all states, while carrier screening is recommended for pregnant women or those considering pregnancy. General screening identifies asymptomatic parents who have an affected child or family members of individuals with SCD or sickle cell trait. Diagnosis detects SCD in those with a positive screening test or symptoms of unexplained anemia or abnormal blood count results. Several types of tests are available, including hemoglobin S solubility and sodium metabisulfite tests for screening individuals 6 months old or older, and hemoglobinopathy evaluation methods like electrophoresis, fractionation by HPLC, isoelectric focusing, mass spectrometry, and capillary zone. Genetic testing (DNA analysis) determines whether someone has one or two gene copies of the Hb S mutation or has two different mutations in hemoglobin genes. This test can be used for carrier testing, diagnosis, and monitoring treatment response. Understanding test results, implications, and risks of passing genetic disorders to potential children is crucial. Additional tests that aid in evaluating individuals with suspected or confirmed sickle cell trait or disease include: Complete blood count (CBC), which assesses red blood cell counts, hemoglobin levels, and RBC size and shape; Blood smear analysis to detect abnormal RBCs like sickle-shaped ones; Iron studies to evaluate iron storage and usage, as people with sickle cell anemia may experience iron overload due to repeated blood transfusions. Sickle cell tests are typically conducted shortly after birth for newborn screening, especially among African Americans where the disease occurs in 1 out of every 365 births. Carrier screening is also offered during pregnancy or pre-pregnancy consideration. When a woman is found to be a carrier, testing is recommended for her partner. The tests may also be ordered when abnormal CBC and blood smear results are seen alongside signs and symptoms suggesting sickle cell anemia, such as recurring pain due to sickle cell crises, anemia, increased infections, or acute chest syndrome. People with the sickle cell trait mainly produce normal hemoglobin A, while those with the disease will mostly have Hb S without any Hb A. Individuals who inherit two different types of hemoglobin genes will typically have varying amounts of both. For instance, they may produce both Hb S and Hb C but no Hb A. Genetic testing can identify sickle cell status by detecting specific gene mutations. If two copies of the Hb S gene mutation are found, it indicates sickle cell disease. Having one gene for Hb S and a normal gene means someone has sickle cell trait. Individuals with one Hb S copy and another mutation, like Hb C or beta thalassemia, may experience symptoms similar to those with sickle cell disease. Sickle cell testing can produce various results, which are listed in the following table: | Results Seen | Condition | Genes | | --- | --- | --- | | Slightly decreased Hb A; Moderate amount Hb S (about 40%) | Sickle cell trait | One gene copy for Hb S (heterozygous) | | Majority Hb S; Increased Hb F (up to 10%); No Hb A | Sickle cell disease | Two gene copies for Hb S (homozygous) | It's essential to note that sickle cell anemia symptoms and complications can vary greatly from person to person, even within the same family. Recent blood transfusions may cause false-negative test results in some cases. People with sickle cell trait are generally healthy but may experience symptoms under specific conditions, such as intense exercise, dehydration, or high altitudes. Sickle cell carriers produce both Hb A and some Hb S, which can lead to sickling when exposed to significant stress that reduces oxygen levels in the body. Sickle cell disease is more prevalent among those of African ancestry and those who can trace their roots to the Mediterranean area, South and Central America, the Middle East, India, and the Caribbean. Historically, sickle cell offered protection against malaria in these areas, but its prevalence has increased worldwide due to migration. According to a 2013 study, the rates of sickle cell anemia are projected to affect over 400,000 newborns by 2050, with India and sub-Saharan Africa showing the sharpest increases. Newborn screening identifies most cases of sickle cell trait and disease, but some adults may not have been tested. The National Collegiate Athletic Association (NCAA) recommends testing college athletes who haven't documented their sickle cell status due to the risk of "exertional sickling" during intense training. Your healthcare practitioner may request DNA sequencing for further evaluation. The purpose of this test is to identify abnormal types of hemoglobin that might be present in a person's blood. This is not a standard procedure, but rather an additional step taken if there's suspicion of a rare type of hemoglobin (variant), such as hemoglobin F. DNA sequencing is used here instead of testing for specific gene mutations; it involves determining the order of DNA building blocks in a person's genetic code. This method can help detect hemoglobin disorders caused by less common mutations. A sickle cell test, which includes blood work and high-performance liquid chromatography (HPLC), is used to check for sickle cell trait or disease, an inherited condition that affects red blood cells' shape due to the presence of abnormal hemoglobin S instead of normal hemoglobin A. If the results from HPLC are inconclusive, a genetic test may be performed to confirm the findings. People can inherit genes in various combinations, leading to different types of hemoglobin: two sets of genes producing normal hemoglobin, one set making hemoglobin A and another making hemoglobin S (carriers), or two sets of genes making hemoglobin S (sickle cell disease). The test is especially important for identifying sickle cell trait in couples planning a family. It's also recommended by the United States Preventive Services Task Force that all newborns be tested for this condition, and it may be done for individuals at high risk or with previous blood transfusions within the last four months. A small amount of blood is drawn from an arm vein or, for babies, a heel prick is used to collect several drops of blood. The procedure is generally quick and causes minimal discomfort, although there's a slight risk of bruising at the injection site. Given text here hemoglobin is found in the blood, and it's crucial for babies to be tested for sickle cell disease at six months old. This can help determine if they have genetic information that makes them a carrier of the condition. The test also helps identify people who may be at risk due to their family history or immigration status. Hemoglobin is a protein in red blood cells that carries oxygen from the lungs to other parts of the body. Individuals can discover their carrier status for sickle cell disease through a simple blood test, which can be taken at any point in life. This screening is particularly valuable for those with a family history of the condition or if their partner has been identified as a carrier. Both men and women can undergo this test to determine their risk level. If you suspect you might carry the sickle cell trait, consult your general practitioner about scheduling an appointment for the blood test.