

CARRIER SCREENING FOR SICKLE CELL DISEASE AND THE THALASSEMIAS

Sickle cell disease and Thalassemia disease are inherited genetic conditions. These diseases are examples of autosomal recessive conditions which means that a person must have two traits for that condition in order to be affected (one inherited from their mother and one inherited from their father). Each of us carries a few recessive traits or genes that do not work properly. Because there are thousands of genes, the likelihood that we would meet and have children with another person who carries the same recessive gene or trait that we do is small. However, that chance increases if we come from the same ethnic group as our partner, because people from the same ethnic group share common ancestors, and therefore, common genes.

In large ethnic groups, it is difficult to pinpoint which recessive conditions a person is at risk for, but a few specific risks are known. For example, individuals with an African ancestry are at an increased risk to carry sickle cell trait and individuals who are from Mediterranean countries (Greece, Italy, Turkey) are at increased risk to carry β -thalassemia trait. Both sickle cell disease and thalassemias affect a part of the blood, the hemoglobin, which carries oxygen and iron to other tissues in the body.

WHAT IF I HAVE A FAMILY HISTORY OF ONE OF THESE CONDITIONS?

If you have a family history of sickle cell disease or a thalassemia, such as a brother, sister, or cousin, then you may have a higher chance of being a carrier. Your specific risk is determined based on how you are related to the person in your family with the condition. If the trait, or change in the gene, is known for your family member, then testing can look for those specific changes and rule out whether or not you have inherited them. Genetic counseling is recommended for people who have a family history of an autosomal recessive genetic condition.

WHAT AM I AT RISK FOR IF I HAVE AFRICAN ANCESTRY?

If you are African-American, Afro Caribbean, or have another type of African ancestry, then you may have an increased chance to have a child with a form of sickle cell disease. About 1 in every 10 people (10%) with African ancestry carries a form of sickle cell trait and 1 in every 400 babies born to parents of African ancestry will have sickle cell disease. People who are not of African ancestry can also have sickle cell trait; however, it is less common (for example, about 1 in every 100 Caucasians).

There are two main types of sickle cell trait: "S" and "C". The classic form of sickle cell disease occurs when a child inherits two "S" or sickle cell traits (SS). People with sickle cell disease are at increased risk for pain crises where they have pain in their joints and organs and may feel weak. Eventually, damage may build up in the liver and spleen. People with sickle cell disease are more prone to infections and the disease may lead to a shorter lifespan. Sickle cell-C disease (SC) has one sickle cell trait and one C trait. Children with sickle cell- C disease (SC) often have fewer pain crises than people with SS disease, but the severity is different from person to person. In addition, if a child inherits an "S" from one parent and a β -thalassemia trait from another parent, they will have sickle β -thalassemia (S- β -thal) with problems similar to sickle cell disease. About 1 in every 50 to 75 people with African ancestry carries β -thalassemia trait.

Testing for sickle cell trait can be done several ways. Sometimes a sickle dex, also called a sickle quick or sickle prep, will be performed. However, this type of testing only looks for "S" trait so people with "C" trait or β -thalassemia trait will not be detected. A hemoglobin electrophoresis with A₂ quantitation is the best test for detecting all types of sickle cell trait. Sickle cell screening is routinely drawn as a part of the prenatal labs on pregnant women of African ancestry. Talk to your doctor to find out what type of testing they use and/or consider having screening prior to becoming pregnant. It is also important to know that all babies born in South Carolina are screened for sickle cell disease on their newborn screening test.

WHAT IF I AM OF MEDITERRANEAN ANCESTRY?

People from countries that surround the Mediterranean Sea such as Italy, Greece, Turkey, Morocco and the Middle East have a higher chance to carry β -thalassemia trait. Approximately 1 in every 20 to 1 in every 30 people with ancestry from this region will carry β -thalassemia trait. If both members of a couple are of Mediterranean ancestry, then they have a 1 in 3,000 chance to have a child with Thalassemia major, or Cooley's anemia, which occurs when a child inherits two β -thalassemia traits.

Cooley's anemia causes the body to not make enough red blood cells and leads to enlargement of the liver, spleen, and heart. Children with Cooley's anemia receive blood transfusions to help with oxygen and red blood cell supply. Iron contained in the transfusions can build up in the body leading to other problems. The lifespan of someone with Cooley's anemia is typically shortened.

Screening for β -thalassemia trait can be done by looking at two of the sections of a complete blood count, or CBC. If someone has low MCV and MCH scored on their CBC, then they might be a carrier of β -thalassemia trait. Often the doctor will then draw a hemoglobin electrophoresis with A₂ quantitation to see if the A₂ is elevated. A low MCV, low MCH, and high A₂ indicate a person probably carries β -thalassemia trait. DNA studies can be pursued if both members of a couple are suspected to carry β -thalassemia trait and the couple is interested in prenatal diagnosis.

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WHAT IF I AM OF ASIAN OR SOUTH PACIFIC ANCESTRY?

If you are Asian-American and/or have ancestry from China or other Southern Pacific countries, you have a higher chance to carry α -thalassemia (alpha thalassemia) trait. The chance to carry α -thalassemia trait is about 1 in 20, or 5%. Each person actually has four copies of the α -thalassemia gene. As long as a person has at least two working copies of the α -thalassemia gene, they are not expected to have problems with their blood cells. People of Asian ancestry who are carriers of the α -thalassemia trait are usually missing two copies of the gene on the same chromosomes. If their partner is also a carrier of α -thalassemia trait and is missing two copies of the gene on the same chromosome, then the couple can have child who has no working copies of the α -thalassemia gene. Babies cannot normally survive without any working α -thalassemia genes. Children who inherit just one working copy of the gene survive but have some health problems like a larger than normal spleen.

Screening for α -thalassemia trait can be done by looking at one portion of the complete blood count, or CBC, that is usually done as a part of routine blood work. If the MCV is low, then the doctor may draw a hemoglobin electrophoresis with A_2 quantitation to see if the A_2 is low and may do iron studies to make sure you are not anemic. If the MCV is low, the A_2 is low, and the iron studies are normal, then you probably have α -thalassemia trait. DNA studies can be pursued if both members of a couple are suspected to carry α -thalassemia trait and the couple is interested in prenatal diagnosis of future pregnancies.

WHAT HAPPENS IF I AM A CARRIER FOR SICKLE CELL OR THALASSEMIA TRAIT?

If BOTH members of the couple are carriers of the SAME genetic trait, then the couple has a 25% risk (1 on 4) with each pregnancy to have a child with that condition. The couple may consider DNA studies for use in prenatal diagnosis by CVS or amniocentesis, may test a child at birth, or may consider alternative parenting options such as adoption, egg or sperm donation, or preimplantation genetic diagnosis. Specific testing options vary based on the condition involved.

Genetic counseling is recommended when both members of a couple are found to be carriers. During a genetic counseling session many questions can be answered, such as explaining the inheritance of these conditions, teaching more about the diseases, and helping a couple understand their risks and reproductive options. If one member of the couple is a carrier, but the other member of the couple is found not to be a carrier, then no further testing is recommended. Some risk for the condition may still exist depending on the detection rate of the screening test used for the couple.

SHOULD I HAVE CARRIER SCREENING?

The decision to pursue screening is a personal one. Some women/couples want to know if their chance to have a child with a genetic condition is increased prior to or during pregnancy. Other women/couples do not feel like the risk of these conditions is high enough for them to consider screening.

Talk with your doctor and/or arrange to speak with a genetic counselor at the USC Department of Obstetrics and Gynecology Genetic Counseling office.

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