



My son has SICKLE CELL TRAIT ... WHAT NOW?

Guide for orientation on
SICKLE CELL TRAIT





Introduction

The objective of this manual is to provide information about Sickle Cell Trait to patients and their relatives, as we know that medical information and its technical jargon can be difficult or incomprehensible to most people. Thus, we hope it can answer your questions.

Any comments are welcome, either about the clarity or omission of some information considered important they can be sent as suggestions to HEMORIO or by e-mail ouvidoria@hemorio.rj.gov.br

FOR MORE INFORMATION VISIT:
WWW.HEMORIO.RJ.GOV.BR



What is Sickle Cell Trait?

Sickle cell trait is not a disease. People with sickle cell trait inherit a gene for usual hemoglobin (Hb A) from one parent, and a sickle hemoglobin gene (Hb S) from the other one. It is a condition in which the person, although not present anemia at a routine examination (CBC) carries a kind of modified hemoglobin gene, called the "S".

Sickle cell trait is a genetic disorder inherited from one parent who is not strong enough to manifest as disease.

What is hemoglobin?

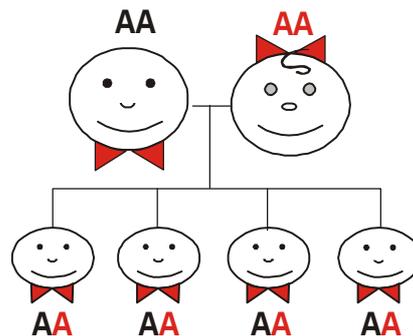
Hemoglobin is a protein that is inside red blood cells and their function is to carry oxygen to body tissues.

A normal hemoglobin gene is called the A and people are considered AA, because they receive a share of the father and one mother.

What is a genetic disorder?

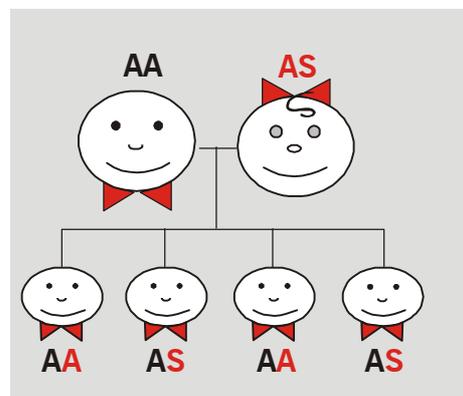
Genetic disorder or hereditary disorder is a condition that is caused by abnormalities in genes that pass down through a family.

The following diagram represents the inherited genes for the formation of hemoglobin A, that is the usual hemoglobin.

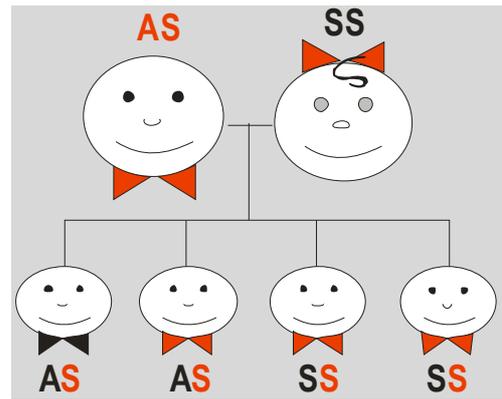


In the case of Sickle Cell Trait, there are two possibilities represented for the following diagrams:

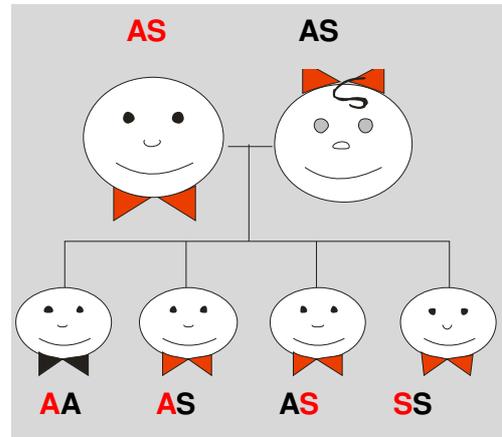
- Sickle Cell Trait people inherit one gene for the usual hemoglobin (A) from one parent and one gene for hemoglobin S from the other parent.
- There is a 50% chance with **each** pregnancy that the child will have sickle cell trait, as well as a 50% chance the child will be unaffected (Hb AA).



- Sickle Cell Trait people inherit one gene for the usual hemoglobin (A) from one parent and one gene for hemoglobin S from the other parent.
- There is a 50% chance with **each** pregnancy that the child will have sickle cell trait, as well as a 50% chance the child will have Sickle Cell Disease (Hb SS).



- Sickle Cell Trait people inherit one gene for the usual hemoglobin (A) from one parent and one gene for hemoglobin S from the other parent.
- There is a 50% chance with **each** pregnancy that the child will have sickle cell trait, as well as a 25% chance child will be unaffected (Hb AA) and 25% chance the child will have Sickle Cell Disease (Hb SS).



How many types of abnormal hemoglobins are there?

There are **other types of abnormal** inherited **hemoglobin** such as Hemoglobin C, D, and E. Sickle cell trait is also called "hemoglobinopathy AS", while others receive the designation for the changed type of hemoglobin. The most frequent hemoglobinopathy traits in our midst, are:

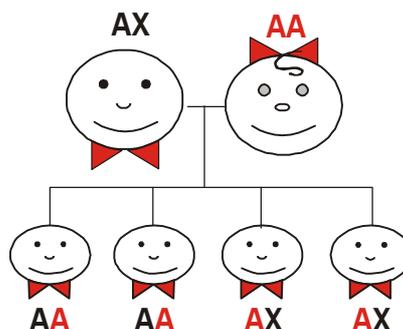
Hemoglobin S trait or hemoglobinopathy AS

Hemoglobin C trait or hemoglobinopathy AC

Hemoglobin D trait or hemoglobinopathy AD

Hemoglobin E trait or hemoglobinopathy AE

The diagram represents the genes for inherited types of hemoglobinopathy trait, being represented by "X", the gene for changed hemoglobin.



Who can have sickle trait?

The gene comes from Africa, and Brazil is a country composed by people from different backgrounds, especially African people. Therefore, sickle cell trait is very frequent in our population.

What is the best time to make the diagnosis of hemoglobinopathy trait?

It should be done at birth, through newborn screening tests. A small amount of blood is collected from the heel of the newborn and sent to the laboratory for the detection of some diseases, including hemoglobinopathies.

➤ In the case of hemoglobinopathy trait, early diagnosis has the purpose to detect hemoglobin in the family.

➤ No caso do traço, o diagnóstico precoce visa detectar a existência da hemoglobina alterada, na família.



Who has sickle cell trait is healthy?

Yes. People with hemoglobinopathy trait have a normal life, because it is not a disease. However, parents with trait can have babies with sickle cell disease.

What are the symptoms of sickle cell trait?

Usually hemoglobinopathy trait is asymptomatic. However, it is advisable that whenever he goes to any consultation attendance for some reason, this condition should be informed to the doctor.

If sickle cell trait does not have any health problem, why do I have to know about it?

The most important in identifying people with sickle cell trait is informing them of their risk of having a baby with sickle cell disease.

Should my family be tested?

If you have a baby with sickle cell trait you or someone in your family can be a carrier. Information on the presence of hemoglobinopathy trait in the family is helpful during the pre-nuptial tests of any member of your family. Soon, the doctor should check if you are at risk of having a child with hemoglobinopathy.

What is sickle cell disease and what are their consequences?

Sickle cell disease occurs when a person inherits the hemoglobin S gene from each parent (Hb SS). Sickle cells cannot pass through blood vessels (veins) properly, causing the main signs and symptoms of the disease, which are: anemia, jaundice (yellow eyes), fatigue, pain crises and severe infections.

Patient with sickle cell disease need specialized medical support, which can be done at the Blood Center and Hematology Service for your area.

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