

Information on a Newborn Screen Result for Sickle Cell Trait (AS)

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MARITIME
Newborn Screening
Dépistage néonatal
DES MARITIMES

What is newborn screening?

These are routine tests done soon after birth. A few drops of blood from a baby's heel are put onto a piece of absorbent paper (blotter). The blood is tested for rare, treatable conditions.

Newborn screening can also find differences in hemoglobin.

What does it mean if a baby has a positive newborn screen result for sickle cell trait (AS)?

This means the baby has sickle cell trait (AS). This is also known as "genotype AS" or being a "hemoglobin S carrier". No further testing is needed to confirm the baby's carrier status. Babies with sickle cell trait usually have no symptoms or health concerns, and do not need special medical treatment. Sickle cell trait will not change into sickle cell disease later in life. Families may feel worried about the baby's newborn screen result. It is important to remember that the baby does not have sickle cell disease and is not expected to have health problems because of sickle cell trait.

How is a positive newborn screen result reported for sickle cell trait (AS)?

Maritime Newborn Screening sends the result and information packages to the primary care provider/clinic. The primary care provider will explain the result to the family.

What is sickle cell trait (AS)?

Sickle cell trait is an **inherited** (genetic) difference affecting the hemoglobin in a person's red blood cells. Hemoglobin is a part of a red blood cell. Hemoglobin carries oxygen throughout the body. The hemoglobin genes tell the body how to make hemoglobin. Each person has two copies of the hemoglobin genes because they get one gene from each parent. Hemoglobin genes typically make normal hemoglobin, called "hemoglobin A". A person with sickle cell trait (AS) has one gene that makes hemoglobin A (normal hemoglobin), and one gene that makes hemoglobin S (sickle hemoglobin). People with sickle cell trait (AS) still make normal hemoglobin, which is why they do not have symptoms like someone with sickle cell disease.

How many babies have sickle cell trait (AS)?

Sickle cell trait is found most often in people of African (in West Africa, 1 in 6 people have sickle cell trait), Mediterranean, Middle Eastern, Caribbean, Central American, and Asian ancestry. However, it is possible for a person of any ancestry to have sickle cell trait. People with sickle cell trait may not know they have it because there are typically no health problems.

How does a baby get sickle cell trait (AS)?

Sickle cell trait is **inherited**, meaning it is passed on from parent to child. It is not contagious, and it is not caused by germs. Someone with sickle cell trait (AS) inherited a normal hemoglobin gene (making hemoglobin A) from one parent, and a changed hemoglobin gene (making hemoglobin S) from the other parent. This is called autosomal recessive inheritance (see Figure 1).

Why screen for sickle cell trait (AS)?

Knowing trait status can be important for family planning. When both partners know their trait status, they can know the chance to have a child with **hemoglobin disease**. After knowing a baby's trait status, they can be told later in life that they have sickle cell trait so their future partner can be offered testing. It is also important to know because if a baby has sickle cell trait it means at least one parent also has sickle cell trait. It is possible that both parents could have a hemoglobin trait. If both parents have trait, it could be possible to have a pregnancy/baby with hemoglobin disease.

Summary:

- Sickle cell trait does not usually cause health problems.
- If both parents have sickle cell trait, they could have a baby with sickle cell disease.
- **Parents can be tested for sickle cell trait.** If parents have already been tested outside of Canada, it is recommended to **re-test to confirm the result.**

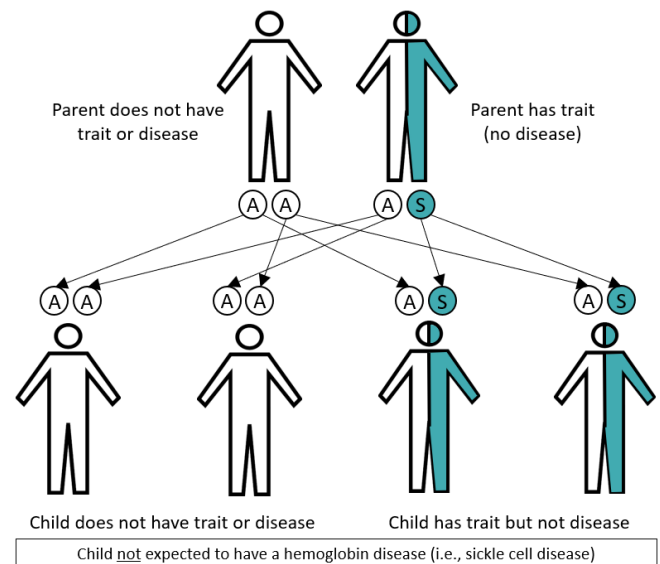


Figure 1. Autosomal recessive inheritance when one parent has sickle cell trait (AS).

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What is hemoglobin disease?

Someone with hemoglobin disease can only make abnormal hemoglobin. They have gene changes (variants) in both of their hemoglobin genes. The symptoms can be very different depending on what hemoglobin variants are present. An example of hemoglobin disease is **sickle cell disease** (also called “**genotype SS**”).

Could a family have another baby with sickle cell trait (AS) or hemoglobin disease?

Yes. If a child has sickle cell trait it means that likely one biological parent has sickle cell trait, and it is possible for both parents to have a hemoglobin trait. **If a baby is known to have sickle cell trait parents have the option of being tested.** Testing is a blood test. Some of the hemoglobin traits that parents are tested for include sickle cell trait, hemoglobin C trait (AC), and beta-thalassemia trait.

If parents have been tested for hemoglobin traits outside of Canada, we recommend **repeating** this testing to confirm the results. Testing in some countries may miss sickle cell trait and other hemoglobin traits

If **both** parents have a hemoglobin trait there is a 25% (1 in 4) chance in every pregnancy to have a child with hemoglobin disease. Some sickle cell diseases are mild, and some can be severe. Sickle cell disease can cause significant health problems and need ongoing medical care. See Figure 2 for an example of when both parents have sickle cell trait (AS).

Where can I get more information?

If **both** parents have a hemoglobin trait, a referral to **Maritime Medical Genetics Service** can be offered to talk about the following:

- inheritance
 - information for future pregnancies
 - testing options for other family members
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- For more information on newborn screening, please visit our website at www.maritimenewbornscreening.ca or call the newborn screening genetic counsellor at 902-470-2783.
 - Websites for families:
 - St. Jude’s Hospital: <https://together.stjude.org/en-us/medical-care/inherited-risk-genetic-testing/sickle-cell-trait.html>
 - KidsHealth: <https://kidshealth.org/en/parents/sickle-cell-trait.html>
 - Websites/resources for healthcare providers:
 - UpToDate: <https://www.uptodate.com/contents/sickle-cell-trait>
 - GeneReviews: <https://www.ncbi.nlm.nih.gov/books/NBK537130/>
 - SOGC Carrier Screening Guidelines: <https://www.jogc.com/article/S1701-2163%2816%2932975-9/pdf>

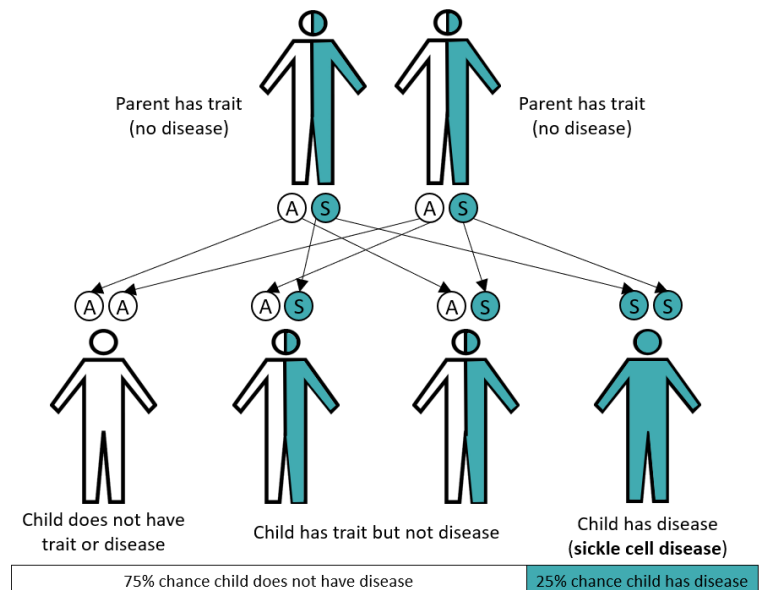


Figure 2. Autosomal recessive inheritance when **both** parents have sickle cell trait (AS).

NOTE TO PARENTS/GUARDIANS: This material is provided for informational purposes and provides basic information only. This material is not intended to be and does not take the place of medical advice, diagnosis, or treatment. Please talk to your health care provider if you have any questions or concerns.



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