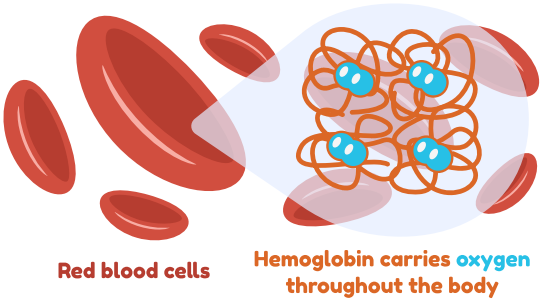
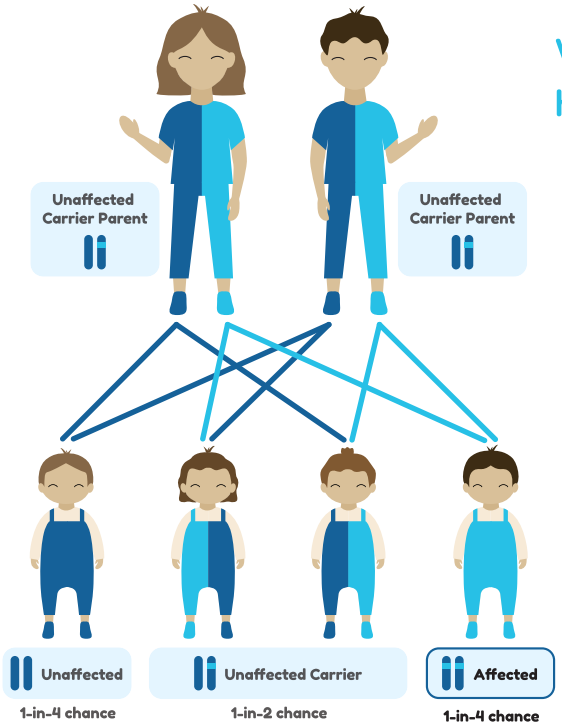
A Patient's Guide to Hemoglobinopathies

What are hemoglobinopathies?

- Hemoglobinopathies are genetic conditions that affect a protein called **hemoglobin**, which carries oxygen in red blood cells to all parts of the body. Examples of hemoglobinopathies are sickle cell disease, alpha-thalassemia, and beta-thalassemia.



- We all inherit genes that help make hemoglobin from our parents. Hemoglobinopathies are caused when a person inherits differences in these genes that prevent hemoglobin from being made correctly from *both* of their parents.
- When hemoglobin isn't made correctly, the number and shape of red blood cells can be affected. This can make it harder for oxygen to travel through the body and lead to symptoms like extreme tiredness, pain, and organ damage. While mild hemoglobinopathies may not need any medical treatment, more severe cases require treatment early in life.



What does it mean to be a carrier of a hemoglobinopathy?

- A **carrier** is a person who inherits one normal copy of a hemoglobin gene and one different copy of a hemoglobin gene. Carriers of a hemoglobinopathy do not show signs of these conditions, and often do not know they are carriers until they are tested.
- If two carriers of a hemoglobinopathy have a child, each of their children has a 25% or 1-in-4 chance of inheriting the different gene from both parents and being affected by the condition. The types and severity of symptoms depend on factors such as the specific genetic differences that are involved.

What are my next steps if I am a carrier?

- If you are a carrier of a hemoglobinopathy, testing your reproductive partner will inform the chance that your child could be affected by a hemoglobinopathy. Partner carrier testing is done with a blood test.
- The chance that your partner is a carrier of a hemoglobinopathy depends on factors such as ancestry. Hemoglobinopathies are most common in people of African, Mediterranean, Middle Eastern, Southeast Asian, and West Indian descent.

If your partner is also a carrier:

The chance of having a child with a hemoglobinopathy is 1-in-4.

The chance of having a child who is also a carrier is 1-in-2.

If your partner is not a carrier:

The chance of having a child with a hemoglobinopathy is very low.

What are my options if my partner and I are both carriers?

If you and your partner are both carriers, a genetic counselor or doctor who is an expert in genetics can talk with you more about what this means for you and your future children.

- If you are currently pregnant, prenatal tests such as chorionic villus sampling (CVS) or amniocentesis can be carried out to see whether your baby is affected by a hemoglobinopathy. It is up to you whether you would want to test your baby prenatally or once they are born.
- If you are not currently pregnant, you have several options you may consider for a future pregnancy:
  - You may decide to become pregnant naturally and test the baby prenatally to see whether they are affected. You may also decide not to test the baby until birth.
  - You may decide to become pregnant through in vitro fertilization (IVF). You may be able to use your own eggs and sperm and select an unaffected embryo, or use eggs or sperm from a donor who is not a carrier.
  - You may decide to become pregnant through intrauterine insemination (IUI) with sperm from a donor who is not a carrier.
  - You may decide to adopt a child.

Where can I find more information?

- Talk to your doctor about any questions you have and meeting with a genetic counselor, or call (844) 743-6384 to make an appointment with a genetic counselor directly.
- For information on hemoglobinopathy carrier screening, visit www.acog.org/womens-health/faqs/carrier-screening-for-hemoglobinopathies.
- For information on hemoglobinopathy newborn screening, visit www.babysfirsttest.org/newborn-screening/conditions/hemoglobinopathies.
- Visit www.cdc.gov/ncbddd/sicklecell for information on sickle cell disease, or visit www.cdc.gov/ncbddd/thalassemia for information on thalassemias.