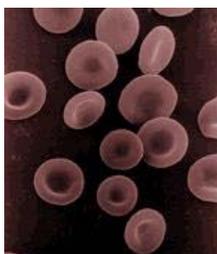


Parents' Guide to Hemoglobin C Disease

You have just learned that your infant has hemoglobin C disease. Naturally you are concerned and have many questions. This information sheet will help answer some of your questions, however an informed discussion with your baby's doctor (primary care provider) is recommended.

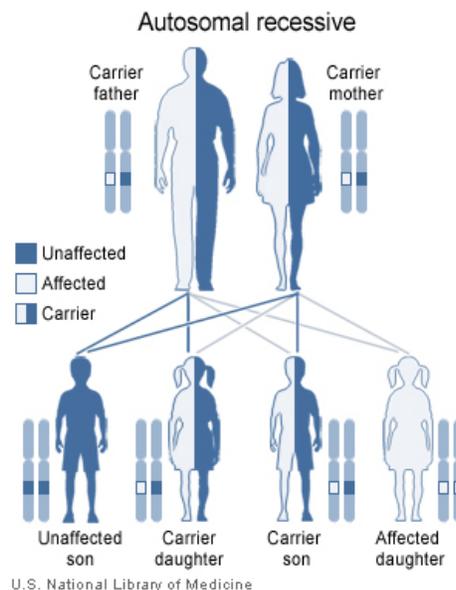
What is Hemoglobin (HB) ?



Hemoglobin is a protein in the red blood cells. HB carries oxygen from the lungs to all parts of the body and gives blood its red color. There are many hemoglobin types (this is not the same as a blood type). Hemoglobin is inherited through genes, one from each parent. Most people have hemoglobin A, also called adult or normal hemoglobin. The presence of hemoglobin A in red blood cells makes them smooth and shaped with a large, rounded depression on each side of the cell, known as a biconcave disk shape (see picture). This shape allows normal red blood cells to be very flexible, moving easily through the blood vessels to deliver oxygen to the body.

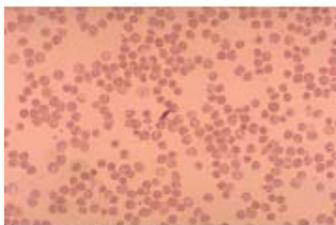
What is Hemoglobin C Disease (CC)?

Hemoglobin C in the red blood cells is responsible for causing hemoglobin C disease. Children *inherit* this disease from their parents as a recessive genetic disorder. This means a hemoglobin C gene is passed from *both* mom and dad to the baby causing hemoglobin C disease (hemoglobin CC). Persons with hemoglobin CC have only hemoglobin C and no adult or normal hemoglobin. When both parents have one hemoglobin C gene, there is a 1 in 4 or 25% chance with each pregnancy that an infant will inherit two hemoglobin C genes. There are no serious health problems associated with hemoglobin CC, but the gene for hemoglobin C is passed on from your child to your future grandchildren. Hemoglobin C disease is not contagious.



Persons with hemoglobin C disease have red blood cells that are smaller than normal and round (ball or sphere) shaped instead of the biconcave disk shaped normal red blood cell (pictured above). Some hemoglobin C red blood cells become rod-shaped because straight-edged, six-sided crystals form inside of the red blood cell.

What Problems can Hemoglobin C Disease (CC) Cause?



The round shaped hemoglobin C red blood cells (see picture on left) are not very flexible in moving through blood vessels and have a smaller outside surface area to carry oxygen. They are very fragile and are more likely to burst than normal red blood cells; so their lifespan is shorter. (Normal blood cells live about 120 days). This leads to mild anemia and decreases the ability of red blood cells to hold onto oxygen. Bilirubin is produced from the remains of the ruptured red blood cells,

which can cause jaundice and gallstones. Most people do not have symptoms. Some persons may experience bone, joint and muscle pain.

What is the Frequency of Hemoglobin C Disease (CC)?

Hemoglobin C disease affects both sexes equally. It occurs in less than one percent (<1%) of the population both in the United States and throughout the world. The disease occurs most often in northern Africa and Italy and in persons with African, Sicilian or Hispanic ancestry.

What can be done to Treat Hemoglobin C Disease (CC)?

Treatment is usually not necessary. Folic acid supplements may be prescribed by your child's doctor to help his or her body to produce normal red blood cells and improve the symptoms of anemia. Treatment is needed if gallstones develop. People with hemoglobin C disease can expect to lead a normal life.

What is Hemoglobin C/Beta-Thalassemia Disease?

Hemoglobin C/beta-thalassemia disease is a more serious disease than hemoglobin C disease (CC). Children with hemoglobin C/beta-thalassemia *inherit* one gene for hemoglobin C from one parent and one beta-thalassemia gene from the other parent. The beta-thalassemia gene causes the body to make less than the normal amount of hemoglobin. Hemoglobin C/beta-thalassemia disease causes moderate destruction of the red blood cells. Persons who are affected may have a more severe anemia and the spleen may be enlarged. Your baby's doctor will do a complete blood count (CBC) and smear of the red blood cells to look for beta-thalassemia when your baby is approximately 6 to 9 months of age.

What are the Most Important Things to Remember about Hemoglobin C Disease (CC) and Hemoglobin C/Beta-Thalassemia Disease?

- Work closely with your child's doctor and hematologist (a doctor who is a blood specialist). Make sure your child has regular checkups with them.
- Call your child's doctor when you have questions and have your child seen if you have any medical concerns.

What about Sickle Cell Trait (FAS)?

- If you have HB C disease, it is important your partner be tested for hemoglobinopathies, especially Sickle cell trait (FAS), before you have children of your own. There is a 50/50 chance of having a child with Hemoglobin C disease when your partner has sickle cell trait (FAS).

How Do I Get More Information about Hemoglobin C Disease?

- Talk with your baby's doctor. You may also want to have a genetic consultation for you and your family to see how these diseases might affect future children or grandchildren.

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