Newborn Screening ACT Sheet

FC (HbCC Disease or HbC/Beta Zero Thalassemia)

HbC/β0 Disease

Differential Diagnosis: Homozygous hemoglobin C, hemoglobin C/beta zero (β0) thalassemia, or hereditary persistence of fetal hemoglobin (Hb C/HPFH).

Condition Description: An uncommon red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin C in the absence of hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>C).

You Should Take the Following Actions

- **Contact the family to inform them of the screening result.**
- **Evaluate infant, assess for splenomegaly, and do complete blood count (CBC) and mean corpuscular volume (MCV).**
- **Contact a pediatric hematologist to determine need for further testing.**
- **Initiate timely confirmatory/diagnostic testing as recommended by consultant.**
- **Report findings to newborn screening program**

Diagnostic Evaluation: CBC and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC), shows FC pattern. DNA studies are used to confirm genotype.

Clinical Considerations: Infant is usually normal at birth. Hemoglobin CC is associated with a mild hemolytic anemia. Aplastic crises and gallstones may occur. Individuals with hemoglobin C/beta zero have a more severe anemia, splenomegaly, and rarely, bone changes. C-HPFH is clinically mild.

Additional Information:

Hemoglobin Disorders (Grady Comprehensive Sickle Cell Center)
http://scinfo.org/additional-online-books-and-articles/hemoglobins-what-the-results-mean

Hemoglobin C Disease (State Of Texas Department of State Health Services)
http://www.dshs.state.tx.us/newborn/hemo_c1.shtm

Hemoglobin C Disease
http://emedicine.medscape.com/article/200853-overview

Utah Department of Health
http://health.utah.gov/newbornscreening/Disorders/HB/Hb_C_Disease_CC/FactSheet_Family_HbCC_En.pdf

TEXAS Department of State Health Services

Newborn Screening FACT Sheet

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HbC/β0 Disease

What is Hemoglobin?
Hemoglobin is a protein in the red blood cells. It 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 hemoglobin.

What is Hemoglobin C Disease (CC)?
Hemoglobin C in the red blood cells is responsible for causing hemoglobin C disease (CC). 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 CC. Persons with 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 CC, but the gene for hemoglobin C is passed on from your child to your future grandchildren. CC disease is not contagious. Persons with CC have red blood cells that are smaller than normal and round (ball or sphere) shaped. Some hemoglobin C red blood cells become rod-shaped.

What Symptoms or Problems can CC Cause?
[Symptoms are something out of the ordinary that a parent notices.]

The round shaped hemoglobin C red blood cells 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 CC?
CC 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 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 CC can expect to lead a normal life.

What is Hemoglobin C/Beta Zero Thalassemia Disease?
Hemoglobin C/beta-thalassemia disease is a more serious disease than 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 CC and Hemoglobin C/Beta Zero Thalassemia Diseases?
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.

How Do I Get More Information about CC?
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.