IDPH
ILLINOIS DEPARTMENT OF PUBLIC HEALTH

NEWBORN SCREENING
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Hemoglobin C Disease

Information for Physicians and Other Health Care Professionals

Definition
Hemoglobin C is an inherited variant of hemoglobin A. Individuals with hemoglobin C disease do not produce hemoglobin A; they only produce hemoglobin C.

Clinical Symptoms
A mild hemolytic anemia develops in the first few months of life as fetal hemoglobin decreases and hemoglobin C increases. Individuals with hemoglobin C disease may develop an enlarged spleen or jaundice. Pigmented gallstones may develop in adulthood.

Newborn Screening and Definitive Diagnosis
In Illinois, high performance liquid chromatography (HPLC) is used to test for the presence of hemoglobin variants, such as hemoglobin C. **If results indicate presence of hemoglobin C, referral should be made to a pediatric hematologist.** Confirm diagnosis with infant blood tests and test the parents to rule out hemoglobin C thalassemia disease.

Treatment
Treatments are available to relieve symptoms caused by the disease.

Incidence
Hemoglobin C disease is seen most frequently among people of African descent and those with families from Italy, parts of the Middle East, and parts of Central and South America; however, anyone can have the disease.

Inheritance Patterns
Hemoglobin C is an autosomal recessive disease that occurs when an individual inherits two hemoglobin C genes, one from each parent. Parents of a child with hemoglobin C are usually unaffected, healthy carriers of the disease. They have one copy of the hemoglobin C gene and are said to have hemoglobin C trait.

If both parents have hemoglobin C trait, there is a 25 percent chance that they will have a child with hemoglobin C disease; a 50 percent chance for a child with hemoglobin C trait; and a 25 percent chance for an unaffected child. **Genetic counseling is recommended for families planning future pregnancies.**

Physiology
Hemoglobin C disease is caused by a glutamic acid substitution (in place of lysine) at the sixth position of an individual's beta globin chains. The resulting hemoglobin variant is unstable and causes red blood cells to be broken down more quickly than normal (anemia).

Key Points for Parents
Hemoglobin C disease is a mild hemolytic anemia. Avoid overly alarming the child's parents, especially if the diagnosis has not yet been confirmed.

If results indicate presence of hemoglobin C, make certain the parents understand the importance of following the pediatrician's and/or pediatric hematologist's recommendations for additional testing and referrals.

Follow-up After Confirmation of Diagnosis
These guidelines should be followed after a diagnosis of hemoglobin C disease has been confirmed:

1) Follow-up with the child's hematologist as recommended.
2) Parents should understand that hemoglobin C is a mild disease; symptoms may not appear in every affected individual and treatment is not always necessary.
3) Recommend genetic counseling services to help the parents understand the complexity surrounding the carrier state and inheritance of this disease.
4) Additional information about newborn screening can be found at:

   - Baby's First Test: http://www.babysfirsttest.org/
     Health Resource and Service Administration (HRSA), Grant no. U36MC16509, Quality Assessment of the Newborn Screening System.

   - National Center for Biotechnology Information: http://www.ncbi.nlm.nih.gov/gtr/
     National Center for Biotechnology Information, U.S. National Library of Medicine 8600 Rockville Pike, Bethesda MD, 20894 USA.