**Newborn Screening ACT Sheet**

# Beta Thalassemia Major
## (Hemoglobin F [Fetal] Only)

**Differential Diagnosis:** Homozygous beta zero thalassemia (thalassemia major), hereditary persistence of fetal hemoglobin (HPFH), and prematurity.

**Condition Description:** A red blood cell disorder characterized by a lack of normal beta globin production and absence of Hb A (F [fetal Hb] only).

**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) for Hb, red blood count (RBC), 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 the newborn screening program**

**Diagnostic Evaluation:** CBC, RBC, and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing, or high performance liquid chromatography (HPLC), shows F-only pattern. DNA studies should be used to confirm genotype.

**Clinical Considerations:** Newborn infants with this finding are usually normal, but severe anemia may develop in the first few months of life. Complications eventually include growth retardation, intercurrent infections, progressive hepatosplenomegaly, skeletal abnormalities, and severe iron overload. Comprehensive care including family education, immunizations, regular transfusions, and prompt treatment of acute illness reduces morbidity and mortality.

**Additional Information:**

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

**Thalassemias**
http://kidshealth.org/parent/medical/heart/thalassemias.html

**Genetics Home Reference**
http://ghr.nlm.nih.gov/condition/beta-thalassemia

**American College of Medical Genetics and Genomics**
https://www.acmg.net/StaticContent/ACT/HBF(F).pdf

**NIH Genetics Home Reference**
http://ghr.nlm.nih.gov/condition/beta-thalassemia

**National Center for Biotechnology Information**
http://www.ncbi.nlm.nih.gov/books/NBK1426/

TEXAS Department of State Health Services

**Newborn Screening FACT Sheet**

# Beta Thalassemia Major
## (Hemoglobin F [Fetal] Only)

### What is Beta Thalassemia Major?

Beta Thalassemia Major is a life-long blood disorder that decreases the production of hemoglobin. Hemoglobin is a protein in the red blood cell that carries oxygen throughout the body. Without enough hemoglobin, red blood cells do not develop normally which leads to anemia as well as a lack of oxygen supply to the body.

### What Causes Beta Thalassemia Major?

Beta Thalassemia Major occurs when a baby inherits two mutated or altered genes, each of which carries the thalassemia trait. If you have one altered gene, then you are a carrier and usually have no symptoms or mild anemia. If you have two altered genes however, you may have beta thalassemia disease and have moderate to severe anemia.

### What Problems Occur with Beta Thalassemia Major?

Signs and symptoms usually appear between 6 and 24 months of age. These problems include:

- anemia
- feeding problems and poor weight gain (failure to thrive)
- paleness
- jaundice (yellowing of the skin and whites of the eyes)
- diarrhea
- irritability
- recurrent fevers
- splenomegaly
- bone abnormalities

### What is the Treatment for Beta Thalassemia Major?

**Hematology Referral** - Your child's pediatrician will refer your baby to also see a pediatric hematologist, who is a physician that specializes in blood disorders.

**Blood Transfusions** - For children with Beta Thalassemia Major, anemia can be life-threatening so frequent blood transfusions are usually required due to the lack of normal red blood cells.

**Chelation Therapy** - With frequent blood transfusions, iron (found in red blood cells) can build up in the body. Since iron is a heavy metal, too much can harm the body. Chelation therapy helps remove the extra iron from the body.