TEXAS Health and Human Services | Texas Department of State Health Services

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).

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);**
- **Repeat newborn screen if second screen has not yet been done;**
- **Contact a pediatric hematologist to determine need for further testing;**
- **Initiate timely confirmatory/diagnostic testing as recommended by consultant; and**
- **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 Expectations

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

American College of Medical Genetics and Genomics – Hemoglobin E ACT Sheet
Utah Department of Health & Human Services – Hemoglobin C, D, and E Disorders
Kids Health – Thalassemias
U.S. National Library of Medicine, Medline Plus – Beta thalassemia

Revised: 5/23/2024