



Newborn Screening ACT Sheet

FEA (HbE/ Beta Plus Thalassemia)

HbE/ β + Disease

Differential Diagnosis

Hb E beta plus thalassemia.

Condition Description

A red blood cell disorder characterized by presence of fetal hemoglobin (F) and hemoglobin E and hemoglobin A. The hemoglobins are listed in order of the amount of hemoglobin present (F>E>A).

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), red blood count (RBC), and mean corpuscular volume (MCV);
- Repeat newborn screen if second screen has not yet been done;
- Consider contact with a pediatric hematologist to determine need for further testing;
- Initiate timely confirmatory/diagnostic testing as recommended by consultant; and
- Report findings to newborn screening program

Diagnostic Evaluation

CBC and MCV. Hemoglobin separation by electrophoresis, isoelectric focusing (IEF), or high performance liquid chromatography (HPLC), shows FEA pattern. DNA studies may be used to confirm genotype.

Clinical Expectations

Infants are usually normal at birth. Clinical severity is variable depending on the specific beta plus thalassemia mutation.

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](#)