

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).

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), red blood count (RBC), and mean corpuscular volume (MCV).
- Consider contact with 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 (IEF), or high performance liquid chromatography (HPLC), shows FEA pattern. DNA studies may be used to confirm genotype.

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

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

Utah Department of Health

http://health.utah.gov/newbornscreening/Disorders/HB/Hb_E_Disease_EE/FactSheet_Provider_HbEE_En.pdf