American College of Medical Genetics ACT SHEET

Newborn Screening ACT Sheet [FEA]

Hemoglobin E/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) for Hb, and mean corpuscular volume (MCV).
- Order hemoglobin profile analysis (usually performed by electrophoresis).
- Consult a specialist in hemoglobin disorders, refer if needed.
- 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 β^+ thalassemia mutation. Severely affected individuals may require life-long transfusion, splenectomy, and treatment for iron overload.

Additional Information:

Hemoglobin Disorders (Grady Comprehensive Sickle Cell Center)

Thalassemias

Genetics Home Reference

<u>Sickle Cell Disease in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths and Protocols for Management of Acute and Chronic Complications</u>

Referral (local, state, regional and national):

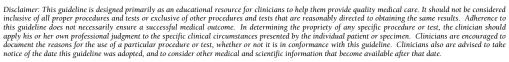
Testing

Clinical Services

Thalassemia Care Center Directory

Thalassemia Treatment Centers Directory

Find Genetic Services





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| State Resource s | ite (insert state newborn screening program website information) |
|--------------------------------|--|
| Name | |
| URL | |
| Comments | |
| | |
| | |
| Local Resource | Site (insert local and regional newborn screening website information) |
| Name | |
| URL | |
| Comments | |
| | |
| | |
| PPENDIX: Resour | ces with Full URL Addresses |
| dditional Informati | |
| http://www.scin | sorders (Grady Comprehensive Sickle Cell Center) fo.org/index.php?option=com_content&view=article&id=218:hemoglobins-what-the-results- |
| mean&catid=11 | &Itemid=21 |
| Thalassemias http://kidshealth | n.org/parent/medical/heart/thalassemias.html# |
| Genetics Home | |
| 1 | ih.gov/condition=betathalassemia |
| | rase in Children and Adolescents: Diagnosis, Guidelines for Comprehensive Care, and Care Paths or Management of Acute and Chronic Complications. |
| | s.state.tx.us/newborn/pdf/sedona02.pdf |
| | |
| | regional and national): |
| Testing | |

Clinical Services

Thalassemia Care Center Directory

http://www.cdc.gov/ncbddd/hbd/thal center list.htm

Thalassemia Treatment Centers Directory

http://www.thalassemia.org/index.php?option=com_content&view=article&id=154:thalassemia-treatmentcenters&catid=39:about-thalassemia&Itemid=27

Find Genetic Services

http://www.acmg.net/GIS/Disclaimer.aspx

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

