ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT
(Fe)
HEMOGLOBIN EE OR E BETA ZERO THALASSEMIA
(Hb EE or Hb E β°)

Disease Category: Hemoglobinopathies

Meaning of the Screening Result: Hemoglobin FE pattern on newborn screen is highly suggestive of homozygous hemoglobin E. Infant may also have hemoglobin E beta zero thalassemia.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact a specialist in hemoglobinopathies for consultation or referral on diagnostic evaluation and management.
- Contact the family to inform them of the screening result.
- Do confirmatory testing (hemoglobin electrophoresis).
- Evaluate the infant with a complete blood count with reticulocyte count at first visit and at six months. If the child develops significant anemia, consult a specialist in hemoglobinopathies.
- Report results to Nebraska Newborn Screening Program.

Pediatric specialists in hemoglobinopathies are available through the centers at Children’s Hospital (402) 955-3950 and UNMC/Nebraska Medical Center (402) 559-7257.

Condition Description: Individuals with hemoglobin FE are homozygous for hemoglobin E or compound heterozygous for hemoglobin E and beta zero thalassemia.

Clinical Expectations: Individuals with Hb EE are not anemic but have microcytosis and target cells on blood smear. Clinical expression in Hb E β° is variable with the most severely affected requiring life-long transfusion, splenectomy and treatment for iron overload.

Confirmation of Diagnosis: Diagnosis is confirmed by hemoglobin electrophoresis and parental or DNA studies as indicated.

Additional information:
- Acute and Chronic Complications - www.tdh.state.tx.us/newborn/sc_guide.htm
- Grady Comprehensive Sickle Cell Center Web Site - www.scinfo.org
- Sickle Cell Disease Association - www.sicklecelldisease.org
- Hemoglobinopathy podcasts http://feeds.feedburner.com/Pediatrics bytes