

ACT SHEET FOR POSITIVE NEWBORN SCREENING RESULT (FE) HEMOGLOBIN EE OR E BETA ZERO THALASSEMIA (Hb EE or Hb E β^0)

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.
- **Repeat NBS to confirm FE result.**
- 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.

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^0 is variable with the most severely affected requiring life-long transfusion, splenectomy and treatment for iron overload.**

Confirmation of Diagnosis: Confirmation may be done by **submitting a repeat dried blood spot filter paper specimen.** Parental or DNA studies may be done as indicated.

Additional information:

- <https://www.orpha.net/data/patho/GB/uk-HbE.pdf>
- hemoglobinopathy Podcasts: mededportal.com search: hemoglobinopathy screening
- www.scinfo.org/sickle-cell-resources/blog → scroll down to: What do the Hemoglobin results mean? Click on question.
- <http://newbornscreeningcodes.nlm.nih.gov/nb/sc/condition/Hb-F-E>
- Hemoglobinopathy Video with CME available at: www.childrensomaha.org/medicaleducation

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