

Hemoglobin E/Beta-Thalassemia Disease

Action required

Laboratory evaluation of infant

- Hemoglobin electrophoresis at 4-6 months
- Because of presence of fetal hemoglobin, newborn screening cannot distinguish Hemoglobin E (benign) from Hgb E/beta thalassemia (clinically significant)

If Hgb E/beta-thalassemia is confirmed on electrophoresis

- Contact pediatric hematologist
- Discuss need for treatment, which can include:
 - § Blood transfusion
 - § Chelation therapy

Review with family

Family has **not** been notified of result by MDH.

Discuss with family that further testing is needed to determine if baby has Hemoglobin E (no risk for health problems) or Hemoglobin E/beta thalassemia (at risk for severe health problems). Generally, infants will be stable when family is contacted and at clinic visit.

If hemoglobin electrophoresis shows Hgb E/beta thalassemia, after discussion with pediatric hematologist, contact family to coordinate clinic visit, lab work, and referral to hematology clinic.

NICU issues

Newborn screens cannot be accurately interpreted after transfusion, or in extremely premature babies where adult hemoglobin may not be detected.

Clinical summary

Hemoglobin E/beta thalassemia is an autosomal recessive disorder that results when an individual produces little or no normal adult hemoglobin. If an infant is not screened and/or left untreated, symptoms begin to appear in infancy and can include moderate to severe microcytic anemia, poor growth, iron overload, splenomegaly, bone deformities, cardiac failure, and infection. Symptoms can range from those similar to mild thalassemia intermedia to those associated with severe thalassemia major.

Affected children may require transfusions, chelation therapy, and monitoring by both primary care and specialty providers.

Incidence: Unknown; Seen primarily in individuals of SE Asian descent - Thailand, Laos, and Cambodia

Clinical expectations

Variable phenotype possible - ranging from complete lack of symptoms to transfusion dependence.

Poor growth, anemia, iron overload, and other symptoms can occur even with treatment.

Affected children should be monitored for complications from anemia or iron overload, and need for intervention.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening