THALASSEMIAS AND HEMOGLOBINOPATHIES

Interacting A-Thalassemia With Hemoglobin E

Hemoglobin (Hb)E is the most common abnormal hemoglobin in Southeast Asia countries. Alpha thalassemia is also prevalent in the region. Thus, the coinheritance of Hb E with **α**-thalassemia is frequently observed.

CONFIRMATORY TESTING

Capillary Electrophoresis (CE), CBC and red blood cell indices (MCH, MCV) testing for both child and parents, and DNA testing are used to confirm the diagnosis.

What should you do?

- Evaluate infant and assess for splenomegaly
- Refer patient to a pediatric hematologist.

Clinical Considerations

The clinical manifestation of Hb E/**α**-thalassemia will depend on the number of deletions in the **α** gene. Hb E/ -**α**/**αα** (1 deletion), Hb E/ -**α**/-**α** (2 deletions) or Hb E/ --/**αα** (2 deletions) will give rise to a mild type of hypochromic anemia with mild splenomegaly. Hb E/ --/-**α** (3 deletions) may give a severe form of HbH disease requiring blood transfusions.

References:

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