THALASSEMIAS AND HEMOGLOBINOPATHIES

Interacting HB D Disease With B-Thalassemia

The newborn screening result may be Hb FDA or Hb FD. HbD/ß thalassemia results from co-inheritance of a ß-thalassemia allele from one parent and the structural variant Hemoglobin D from the other.

CONFIRMATORY TESTING

Capillary Electrophoresis (CE), CBC and red blood cell indices (MCH, MCV) testing for both child and parents, and DNA testing.

What should you do?

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

Important Considerations

Infants with this result are usually normal at birth. Clinical severity is variable and depends on the specific ß-thalassemia mutation. The factor reliably responsible for the phenotype is the imbalance in the globin chain synthesis. If Hb D/ß+, a clinical presentation of thalassemia intermedia is observed. Clinical manifestations may range from microcytic, hypochromic anemia to hemolytic anemia and splenomegaly.

References:

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