THALASSEMIAS AND HEMOGLOBINOPATHIES

Interacting A-Thalassemia With B-Thalassemia

Co-inheritance of the two types of thalassemia is relatively common in Southeast Asia. In a study on a Chinese population, one in every six ß-thalassemia carriers co-inherits **α**-thalassemia.

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 **α**-/ß-thalassemia interaction shows considerable clinical heterogeneity. The clinical improvement depends on the severity of the ß-thalassemia alleles and the number of functional **α**-globin genes. Coinheritance of two **α**-globin gene deletions or a non-deletional **α**2-globin gene mutation in ß0 thalassemia homozygotes is more likely to produce a thalassemia intermedia phenotype. On the other hand, co-inheritance of a single **α**-globin gene deletion in the same group of patients is usually associated with a thalassemia major phenotype. The SEA deletion (--/**αα**) improves the clinical presentation of ß0/ß+ but not necessarily of ß0/ß0-thalassemia. Co-inheritance of a single **α**-globin gene deletion in homozygous or compound heterozygous ß+-thalassemia produces an improved phenotype. On the other hand, co-inheritance of Hb H disease with ß0/ß+-thalassemia present as ß thalassemia minor.

Just as presence of **α**-thalassemia can improve the clinical severity of ß-thalassemia major, the co-inheritance of ß-thalassemia can improve presentation of non-deletional Hb H disease.

References:

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