THALASSEMIAS AND HEMOGLOBINOPATHIES

Interacting HB E Disease With B-Thalassemia

The newborn screening result may be Hb FEA or Hb FE. HbE/ß-thalassemia results from co-inheritance of a ß-thalassemia allele from one parent and the structural variant Hemoglobin E 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 are used to confirm the diagnosis.

What should you do?

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

References:

- American College of Medical Genetics. Newborn Screening ACT Sheet. Retrieved from: https://www.acmg.net/PDFLibrary/
- Hemoglobin-E-ACT-Sheet.pdf. Accessed 8 April 2020.
- Fucharoen S and DJ Weatherall. The Hemoglobin E Thalassemias. Cold Spring HarbPerspect Med. 2012; 2:a011734.
- Kohne, E., Compendium of Hemoglobinopathies, pp 49-53
- Gepte, MB., Naranjo, ML., Bahjin, RR., De Castro, Jr. R., Fajardo, P., Maceda EB., Paclibar MLF. (2022, October) Disorder[Thalassemias Experts Committee Session]. Newborn Screening Reference Center, National Institutes of Health, University of the Philippines Manila.

Important Considerations

Infants with this result are usually normal at birth. Clinical severity is influenced by the instability of the Hb E and the degree and severity of the ß thalassemia mutation. Hence, Hb E/ß° will correspond to the clinical picture of thalassemia major and will require life-long transfusion. While Hb E/ß+ will be similar to the presentation of thalassemia intermedia.