THALASSEMIAS AND HEMOGLOBINOPATHIES

Interacting HB C Disease With B-Thalassemia

The newborn screening result may be Hb FCA or Hb FC. Hb C/ß-thalassemia results from co-inheritance of a ß-thalassemia allele from one parent and the structural variant Hemoglobin C 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.

Important Considerations

Infants are usually normal at birth. The combination of Hb C/ß thalassemia on whether there is complete or partial production of Beta chain. If Hb C/ß+, the clinical presentation will correspond to thalassemia intermedia. The clinical manifestations may range from mild to moderate hemolytic anemia, splenomegaly and bone changes. Serious hemolytic anemia may be observed in the complete absence of Beta chain (ß°).

References:

- American College of Medical Genecs. Newborn Screening ACT Sheet. Retrieved from: https://www.acmg.net/PDFLibrary/Hemoglobin-C-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.