THALASSEMIAS AND HEMOGLOBINOPATHIES

Hemoglobin E Disease

What is Hemoglobin E Disease?

Hemoglobin E (Hb E) is a variant hemoglobin with a mutation in the ß-globin gene causing substitution of glutamic acid for lysine at position 26 of the ß globin chain. Hb E disease is defined by the coexistence of two ßE alleles, resulting in a homozygous state EE.

The newborn screening result may be Hb FE. Other diagnostic possibilities for this newborn screening result include Hemoglobin E 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?

- Contact the family to inform them of the screening result. Recommend confirmatory testing if not yet done.
- Recommend CBC at 4-6 months. If with anemia, refer to a Pediatric hematologist.

Clinical Considerations

The disorder is basically benign and does not require specialized treatment.

References:
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.