THALASSEMIAS AND HEMOGLOBINOPATHIES

Hemoglobin C Disease

What is Hemoglobin C Disease?

Hemoglobin C (Hb C) is a variant hemoglobin with a mutation in the ß globin gene causing substitution of glutamic acid for lysine at position 6 of the globin chain. Hb C disease is defined by the co-existence of 2 ßC alleles (homozygous state CC). The heterozygous form is benign.

The newborn screening result may be Hb FC. Other diagnostic possibilities for this newborn screening result include Hemoglobin C thalassemia.

CONFIRMATORY TESTING

Capillary Electrophoresis (CE), CBC and red blood 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.
- Refer patient to a Pediatric hematologist.

Clinical Considerations

An affected neonate is likely to appear healthy, but has a risk for mild anemia and minor complications. Complications may include splenomegaly, jaundice and increased risk for gallstones.

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.