THALASSEMIAS AND HEMOGLOBINOPATHIES

Hemoglobin D Disease

What is Hemoglobin D Disease?

Hemoglobin D (Hb D) is a variant hemoglobin with a mutation in the ß globin gene causing substitution of glutamine for glutamic acid in the ß globin chain. Hb D disease is defined by homozygous state DD.

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

CONFIRMATORY TESTING

Capillary Electrophoresis (CE), CBC and red blood indices (MCH, MCV) testing for both child and parents, and DNA testing.

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

Most individuals with Hb D disease do not have symptoms. However, some may have mild haemolytic anemia which may be associated with a slightly enlarged spleen. Treatment is usually not necessary. Individuals with Hb D are expected to live a normal life.

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.