THALASSEMIAS AND HEMOGLOBINOPATHIES

Hemoglobin H Disease/ Alpha Thalassemia

What is Hemoglobin H Disease / Alpha Thalassemia?

Hemoglobin H Disease is a red blood disorder characterized by presence of fetal hemoglobin (F) and hemoglobin A, as well as hemoglobin Barts.

The newborn screening result may be Hb FA Barts > 25 %. Other diagnostic possibilities for this newborn screening result include Hb H Constant Spring disease and Alpha Thalassemia Trait.

CONFIRMATORY TESTING

DNA testing is 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

Hemoglobin Barts above 25% in the newborn indicates a possible hemoglobin H disease, a clinically significant form of alpha thalassemia. Deletion or dysfunction of 3 of the 4 alpha globin genes manifests as Hb H disease. This is characterized by variable clinical courses with some exhibiting splenomegaly and pallor especially after febrile episodes and that may require intermittent transfusions. Absence of all four alpha globin genes results in hydrops fetalis and is usually fatal, in utero or shortly after birth.

Note

For hemoglobin Bart's <25%, parents or hematologists may opt for genotyping but this will not be covered by the program and is at the own expense of the patients. This may be taken into account when counselling is done by the pediatrician/ hematologist.

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.