THALASSEMIAS AND HEMOGLOBINOPATHIES

Beta Thalassemia Major

What is Beta Thalassemia Major?

Beta Thalassemia Major is a red blood disorder characterized by a reduction in or lack of normal beta globin production and absence of Hb A (F [fetalHb] only).

The newborn screening result may be Hb F only. Other diagnostic possibilities for this newborn screening result include premature infants.

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.
- Evaluate infant, assess for splenomegaly
- Refer patient to a Pediatric hematologist.

Clinical Considerations

Infants with this disorder may be normal until 4-6 months of life. With beta-thalassemia, severe anemia may develop in the first few months of life. Moderate pallor may appear at this age often requiring regular blood transfusions. Complications later in childhood may include growth retardation, sexual immaturity, intercurrent infections, progressive hepatosplenomegaly, skeletal abnormalities, and severe iron overload. Comprehensive care including family education, immunizations, regular transfusions partnered with compliance to iron chelation therapy reduces morbidity and mortality.

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.