THALASSEMIAS AND HEMOGLOBINOPATHIES

Other Rare Interacting Thalassemias and Hemoglobinopathies

Other rare interacting thalassemias and hemoglobinopathies may be detected via the newborn screening. This will warrant confirmatory testing.

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?

- Evaluate infant and assess for splenomegaly
- Refer patient to a pediatric hematologist.

Clinical Considerations

The clinical manifestations of the interacting thalassemias and hemoglobinopathies may vary depending on the combination. This may vary from mild to severe presentation.

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.