THALASSEMIAS AND HEMOGLOBINOPATHIES

Sickle Cell Disease

What is Sickle Cell Disease?

Sickle cell disease patients have predominant Hb S. This condition is most common in Africa, Middle East and the United States. Affected infants are usually normal at birth but develop anemia later when the Hb S concentration increases and the Hb F decreases. These patients are particularly susceptible to encapsulated bacterial infections such as Streptococcus pneumonia, Hemophilus influenzae, Staphylococcus aureus, and Salmonella.

The newborn screening result may be Hb FS. Other diagnostic possibilities for this newborn screening result include Sickle cell-ß thalassemia and sickle cell-hereditary persistence of fetal hemoglobin.

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.
- Immediately refer the patient to a pediatric hematologist.
- If there are no pediatric hematologists, then start oral penicillin if below 6 months.

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.

Clinical Considerations

Infants with this finding are usually normal at birth. However, severe anemia may develop in the first few months of life. Complications include growth retardation, intercurrent infections, progressive hepatosplenomegaly, skeletal abnormalities, and periodic episodes of pain. These episodes can occur when sickled red blood cells, which are stiff and inflexible, get stuck in small blood vessels. These episodes deprive tissues and organs of oxygen-rich blood and can lead to organ damage, especially in the lungs, kidneys, spleen, and brain. Comprehensive care including family education, immunizations, regular blood transfusions, pain control and prompt treatment of acute illness reduces morbidity and mortality.