Sickle Cell Disease: Overview

Definition

Sickle cell disease (SCD) is a group of inherited red blood cell disorders caused by abnormal hemoglobin resulting in acute and chronic complications.

Pathophysiology

- Point mutation in β-globin gene changing amino acid Glu → Val at position 6, resulting in HbS
- HbS polymerizes → red blood cell sickling → vaso-occlusion, changes in blood viscosity, hemolysis, endothelial dysfunction, and inflammation

Presentation

Acute Complications

- Vaso-occlusive Crisis (Acute Pain Episode)
- Acute Chest Syndrome
- Sepsis
- Splenic Sequestration
- Stroke
- Dactylitis
- Priapism

Chronic Complications

- Poor growth
- Renal disease
- Retinopathy
- Osteonecrosis
- Functional asplenia
- Cholelithiases
- Learning & cognitive difficulties
- Silent cerebral infarcts
- Chronic lung disease
- Pulmonary hypertension
- Congestive heart failure

Diagnosis

Newborn screening in many provinces
Hemoglobinopathy investigation

Management

Targeted towards managing & preventing complications

Involve hematology prior to any blood product transfusion or procedure

Education
Teach families splenic palpation and to urgently seek care with rapid splenic enlargement/fever

Prophylactic Penicillin
Prophylaxis against encapsulated organisms as functionally asplenic. Continue to minimum age 5 years

Immunizations
Routine immunizations + PPSV 23

Transcranial Doppler
Screening for stroke prevention begins at age 2

Hydroxyurea
Increases total fetal Hb, decreases WBC. Decreases admissions for Acute Chest Syndrome and pain episodes.

Red Blood Cell Transfusions
Simple or exchange transfusions. Indicated only in certain clinical situations as high risk alloimmunization. Reduces the HbS%.

Hematopoietic stem cell transplant
Currently, the only cure for SCD

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