sickle cell disease (SCD)

- RCH guideline - sickle cell disease

introduction

- sickle cell disease is a hereditary autosomal dominant haemoglobinopathy due to homozygous HbS which usually presents in childhood
- the gene is present in 8% (2 million) of black Americans who thus have sickle cell trait but only 30,000 have homozygous HbS, thus sickle cell disease occurs in 1 in 500 African Americans
- sickle cell trait is high in parts of Africa (reaching 30% is some areas) and other regions where malaria is endemic (Sicily, Greece, southern Turkey, and India) as sickle cell trait offers survival benefit in malaria prone regions.

clinical features

- painful vaso-occlusive crises
    - the most common and most distinguishing clinical manifestation of SCD
    - triggered by:
        - hypoxia
        - dehydration and acidosis
        - changes in body temperature
- acute and chronic pain in any body part due to infarctions
    - bone pain from infarction of marrow
    - hand-foot syndrome
        - dactylitis presenting as bilateral painful and swollen hands and/or feet in children
    - avascular necrosis of the femoral or humeral head
    - chronic leg ulcers
- chronic, haemolytic anaemia
    - cholelithiasis (gallstones) is common in children
- functional hyposplenism
    - high risk of invasive pneumococcal disease and infections by other encapsulated bacteria
    - dult infections are predominately with gram-negative organisms, especially Salmonella
- aplastic crisis
    - serious complication due to infection with B19V
- splenic sequestration
    - onset of life-threatening anemia with rapid enlargement of the spleen and high reticulocyte count
- growth retardation, delayed sexual maturation, being underweight
- acute chest syndrome:
    - young children present with chest pain, fever, cough, tachypnea, leukocytosis, and pulmonary infiltrates in the upper lobes
    - adults are usually afebrile, SOB with severe chest pain, with multilobar/lower lobe disease
- pulmonary hypertension
- stroke (CVA)
- eye: ptosis, retinal vascular changes, proliferative retinitis

- urologic:
  - kidneys lose concentrating capacity
    - median age at the time of renal failure in patients with SCD is 23 years
    - median survival time after the diagnosis of ESRD is about 4 years
    - median age of death is 27 years, despite dialysis treatment
  - priapism is a well-recognized complication