



POSNA

The Core Curriculum

Sickle cell disease and related hemoglobinopathies

Objectives

1. Describe different combinations of normal and abnormal hemoglobins and their phenotypic expression
2. Describe the pathophysiology of vasoocclusion secondary to hemoglobinopathies
3. Define and describe hand-foot syndrome
4. Describe features of osteomyelitis secondary to sickle cell disease and its differentiation from bone infarct
5. Describe features of avascular necrosis of bone associated with sickle cell disease
6. Discuss the complication rate of orthopaedic surgery for problems secondary to sickle cell disease
7. Describe the major non-orthopaedic problems associated with sickle cell disease

Discussion

Sickle cell disorders are prevalent and a frequent cause of orthopaedic admissions. Hemoglobin S results from an abnormality of the beta-globulin gene on chromosome 11. The gene for hemoglobin S is common in the African and African-American race; estimated as being present in 8% of the African-American population. Hemoglobin C results from a different amino acid substitution on the same gene. Patients homozygous for hemoglobin S have sickle cell disease; patients with hemoglobin S and C have SC disease. The severity of disease in S-thalassemia is variable, depending on the ability to synthesize hemoglobin A. Patients who synthesize no hemoglobin A have a course similar to SS disease, this category is referred to as S-beta(0) thalassemia. Those who synthesize some hemoglobin A S-beta(+) thalassemia, have a course similar to SC disease, which is not as severe.

Low oxygen tension polymerizes the hemoglobin S molecule, distorting the erythrocyte and making it more fragile. At the same time, sickle cells are more viscous, predisposing to an ability to occlude small vessels. This is the basis for virtually all the problems related to sickle cell disease. Obviously, there is considerable variation in severity of this phenomenon in the family of hemoglobinopathies related to sickle cell disease, as indeed there is variation in those with homozygous sickle cell disease.

Non-orthopaedic conditions include cardiomegaly, anemia, hepato and splenomegaly, leg ulcers, priapism, and gallstones. Pubertal growth and skeletal maturation are delayed.

The major orthopaedic complications of the sickle hemoglobinopathies are bone infarct, osteomyelitis, and avascular necrosis of the femoral and humeral heads. Bone infarct of the hand and/or foot is common in the first two years of life, presenting as swelling and pain and fever below 38 degrees. Radiography demonstrates initial soft tissue swelling, followed by periosteal reaction and bone lysis. Salmonella osteomyelitis has been reported concomitantly with hand-foot syndrome, also known as dactylitis.

The most frequent cause for admission in children with sickle cell disease is extremity pain, with a differential diagnosis of infarct versus osteomyelitis. A great deal of effort can be expended trying to differentiate between the two. Recent reports favor gadolinium enhanced MRI and ultrasound as diagnostic aids, as differentiation can be difficult. Subperiosteal fluid of greater than 10mm was almost always associated with osteomyelitis. Children with frequent bone infarcts are not surprisingly more predisposed to osteomyelitis. Nevertheless, a study of admissions to a large children's hospital revealed that only 1.6% of admissions for musculoskeletal pain were identified as being secondary to osteomyelitis. Staphylococcus and salmonella have predominated as the major offending organism in larger series, both are common.

Avascular necrosis is also common, with an overall prevalence of 9.8% in the hip in a study of 2890 patients. Those with Ss disease and alpha-thalassemia were at greatest risk. Children with onset during the middle years tend to have a better outcome than those affected shortly before skeletal maturity, presumably because of greater time to remodel. Results of total hip and shoulder arthroplasties have not been good, with a high complication rate. A study of complications following orthopaedic procedures on patients with sickle cell disease revealed a serious complication rate of 67%, the most two common complications were excessive blood loss and acute chest syndrome.

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