

Answer to Previous Fellow's Challenge Case

The child presented as the clinical challenge in [volume 1, number 2](#), has a diagnosis of Morquio syndrome (Mucopolysaccharidosis IV or MPS IV). This autosomal recessively inherited storage disease is associated with accumulation of keratin sulfate secondary to an enzyme deficiency of either galactosamine-6-sulfatase (Morquio syndrome A) or β -galactosidase (Morquio syndrome B). Urine testing reveals increased excretion of keratin sulfate in both Morquio subtypes. Our patient was diagnosed by skin biopsy to have Morquio syndrome B which is generally less severe than Morquio syndrome A, although there is considerable phenotypic overlap.

Clinically, these children present during their second to fourth year of life with multiple musculoskeletal abnormalities. Prominent features are short-spine dwarfism, overall growth delay, and significant loss of joint range of motion with occasional mild joint swelling. They generally develop a pectus carinatum deformity, kyphoscoliosis, genu valgum and pes planus. Physical deformities progress with age. A major skeletal complication is atlanto-axial instability which results from hypoplasia of the odontoid process. Surgical fusion of the upper cervical spine is often indicated. Other clinical involvement includes corneal clouding and impaired hearing. Intelligence is normal. Patients generally survive into adulthood, but life expectancy is limited.

Radiographic findings in Morquio syndrome help to direct the clinician toward the making this diagnosis. Dysostosis multiplex is the term often used to describe the array of x-ray abnormalities seen in Morquio syndrome and other storage diseases. Vertebral changes are prominent. Platyspondyly of the thoracic and lumbar vertebrae is present, often with central anterior bony protrusion of the vertebral bodies. Cervical spine x-rays often reveal odontoid hypoplasia and atlanto-axial instability. Hip radiographs show a coxa valga

deformity, flattening of the femoral heads and hypoplasia of the lateral aspects of the acetabulum. Radiographs of the hands, with a classic abnormal metacarpal appearance, suggest the diagnosis of a storage disease. Metacarpal bones usually have diaphyseal widening and brachymetacarpia. In Morquio syndrome, the proximal ends of the metacarpal bones tend to be conically shaped.

The differential diagnosis of Morquio syndrome always includes spondyloepiphyseal dysplasia (SED) congenital. Clinical similarities include dwarfism with a short spine, pectus carinatum, kyphoscoliosis and genu valgum. Radiographic similarities include odontoid hypoplasia and platyspondyly. Although the vertebral findings in SED and Morquio syndrome look similar to the rheumatologist, the radiologist can often note differences in the vertebral appearance which are more characteristic of one diagnosis versus the other. For the clinician, the easiest way to differentiate radiographically between Morquio syndrome and SED is to note the marked metacarpal changes in the former, in contrast to the relatively normal appearance of these bones in SED.

All the fellows responding to this Fellow's Clinical Challenge selected spondyloepiphyseal dysplasia as the diagnosis.

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