Coarse Facial Features, Short Stature, Spinal Deformity

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Case History: Skeletal survey for coarse facial features, short stature, spinal deformity in male child of normal intelligence.

Case Study: Coarse Facial Features, Short Stature, Spinal Deformity

Case History: A male child of normal intelligence belonging to a nonconsanguineous marriage was sent to the radiology department for skeletal survey in view of coarse facial features, short stature, and spinal deformity.

Male child of normal intelligence belonging to a nonconsanguineous marriage.
Figure 1. AP radiograph of the hand showing bullet-shaped metacarpals and slanting distal growth plates of the radius and ulna.
Figure 2. Lateral radiograph of the skull showing frontal bossing and J-shaped sella.
Figure 3. Lateral radiograph of the spine showing platyspondyly with central beaking of the vertebral bodies.
Diagnosis: Morquio syndrome

**Imaging Findings**

Radiograph of the hand (Figure 1) revealed shortened metacarpals with proximal tapering of the bases of 2-5 metacarpals (bullet-shaped). The carpal bones were small in size and reduced in number for the patient's age. The growth plates of the distal ulna and the radius were inclined towards each other. Lateral radiograph of the skull (Figure 2) revealed frontal bossing and J-shaped sella. Lateral radiograph of the spine (Figure 3) revealed platyspondyly with central vertebral beaking.
Discussion

Morquio syndrome is a genetic disorder caused by a lack of the enzyme responsible for the breakdown of the mucopolysaccharide keratin sulphate.¹ Patients with Morquio syndrome do not develop mental retardation, as the metabolism of heparan and dermatan sulphate is normal.

Clinical Features²

- Marked dwarfism in early childhood
- Kyphoscoliosis
- Muscular hypotonia
- Pectus carinatum
- Coarse facies
- Corneal clouding
- Enlarged wrists and deformed hands are noted.
- Atlantoaxial dislocations occur owing to odontoid hypoplasia

Patients with Morquio syndrome do not develop mental retardation.
Case Study: Coarse Facial Features

**Radiological Features**\(^1,2\)

- Central beaking of vertebral bodies (Hurlers: anteroinferior beaking)
- Universal platyspondyly
- Hypoplastic or absent odontoid
- Dysplastic acetabula and capital femoral epiphyses
- Short metacarpals with proximal tapering of bases of the metacarpals
- Inclination of the distal growth plate of the radius and ulna
- J-shaped sella

**Differential Diagnosis**

- Hurler’s syndrome and other mucopolysaccharidosis
- Spondyloepiphyseal dysplasia

Differential diagnoses: Hurler's syndrome and other mucopolysaccharidosis
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Published on Diagnostic Imaging (http://www.diagnosticimaging.com)

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