Case 10851

Morquio type B syndrome
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Section: Musculoskeletal system
Area of Interest: Musculoskeletal bone Musculoskeletal joint Musculoskeletal spine Computer applications
Procedure: Diagnostic procedure
Procedure: Education
Procedure: eLearning
Imaging Technique: MR
Special Focus: Congenital Dysplasias Genetic defects
Case Type: Clinical Cases
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Patient: 14 years, female

Clinical History:

A 14-year-old girl presented with a waddling gait, genu valga and mild scoliosis since the age of 5 years. Intelligence was normal. At the age of 12, growth delay was noticed. Plain films of the spine, pelvis and full-leg radiograph as well as MRI of the hip joint were performed.

Imaging Findings:

Plain radiographs showed diffuse changes throughout the whole skeleton. The most remarkable findings were hypoplasia of the odontoid process, scoliosis, and flattened, wedge-shaped vertebral bodies of the entire spine (Fig. 1, 2). Full leg radiograph revealed genu valga (Fig. 3). Radiographs of the hands showed „squaring” of the metacarpal heads, delayed ossification of the carpals (particularly of the scaphoid bone) and hypoplasia of the distal radial and ulnar epiphyses (Fig. 4). Pelvic abnormalities consisted of marked hip dysplasia with flattening of the femoral heads and shallow acetabuli (Fig. 5). MRI of the hip joints showed severe collapse of the femoral heads, bilateral subluxation and joint effusion (Fig. 6). Biochemical testing suggested the diagnosis of Morquio syndrome type B, which has been confirmed by the result of genetic tests (p.Trp273Leu mutation).

Discussion:

Morquio syndrome or mucopolysaccharidosis (MPS) type IV is a rare genetic disorder characterised by defective degradation of keratan sulphate due to an enzymatic deficiency [1]. Two non-allelic variants, A and B, exist. Out of these two, type B is less severe and much less common [2]. It was first described by O’Brien et al. [3] in 1976. In contrast to patients with Morquio type A, in type B central nervous system is spared [4]. Skeletal manifestations, due to keratan sulphate deposition in bone and cartilage, are similar in both types and resemble those of other MPS (“dysostosis multiplex congenita”).

Clinical findings include short stature, corneal clouding and chest deformity. In contrast to other MPS, intelligence is normal.

Physical examination of the musculoskeletal system in Morquio B patients reveals ligamentous laxity of the distal joints and stiffness of the proximal ones, resulting in a waddling gait [5]. Typical radiographic features include odontoid hypoplasia, platyspondyly, tongue-like appearance of the vertebra (with “central beaking”), underdevelopment of epiphyses, hip dysplasia, shallow acetabuli, overconstricted iliac wings, coxa and genu valga. Destructive hip disease is usually more severe than in other MPS. Hand radiographs may reveal hypoplasia of the
distal radius and ulna, squaring of the metacarpal heads and small or absent carpals. MRI may be used to evaluate the degree of hip destruction. It may show steeply oblique acetabular roof with, flattening of the femoral heads and premature degenerative changes [6]. Urine test may not be sufficient for confirmation of MPS IVB and enzymatic analysis is mandatory to assess the β-galactosidase deficiency. Although the diagnosis of Morquio is based on the combination of clinical examination, biochemical and genetic testing, medical imaging may add valuable information on disease progression disease and monitoring of complications. Most importantly it allows detection of dens hypoplasia, which can lead to atlanto-axial subluxation and eventually death [7].

Although new therapeutic options are emerging, nowadays management of patients suffering from Morquio syndrome is still mainly symptomatic and limited to ensuring control of acute and chronic pain symptoms. Rehabilitation and correction of genu valga deformity are crucial to preserve functionality. Enzyme replacement therapy (ERT) and haematopoietic stem cell transplantation (HSCT) are currently under investigation for MPS IV patients. Both ERT and HSCT, although not cures, may be able to alter the natural history of the disease [8]. Other promising therapeutic approaches, such as gene therapy or small molecule therapy, are currently being scrutinised.

**Differential Diagnosis List:** Mucopolysaccharidosis type IVB (Morquio syndrome type B), Perthes disease, Other mucopolysaccharidoses, Other spondyloepiphyseal dysplasias, Dystrophic dysplasias

**Final Diagnosis:** Mucopolysaccharidosis type IVB (Morquio syndrome type B)

**References:**


**Figure 1**

**Description:** There is flattening of the vertebral bodies and hypoplasia of the dens (letter D). Note broad atlantodental interval (blue arrow). **Origin:** Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium
**Description:** Plain radiographs of the thoracic (a) and lumbosacral (b) spine. Note platyspondyly and central vertebral "beaking" with tongue-like appearance (arrows). **Origin:** Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium
Description: Genu valga. Origin: Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium
Description: There is a distal ulnar and radial hypoplasia and delayed ossification of the carpals. Note also „squaring” of the metacarpal heads (arrows). Origin: Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium
**Figure 5**

Description: Note flattening and fragmentation of the femoral heads (arrows) and shallow acetabuli.

Origin: Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium
Figure 6

Description: Coronal fatsuppressed T2-WI (a) and coronal T1-WI (b) of the pelvis. There is marked destruction and subluxation of both femoral heads (arrows). Note also bilateral joint effusion. Origin: Parizel PM, Department of Radiology, Antwerp University Hospital & University of Antwerp, Antwerp, Belgium