Joint Bone Spine 86 (2019) 121–122

Available online at
ScienceDirect
www.sciencedirect.com

Elsevier Masson France
EM|consulte
www.em-consulte.com/en

Correspondence

Joint manifestations can provide diagnostic clues in Morquio syndrome. Comment on: "Mucopolysaccharidoses seen in adults in rheumatology" by Mitrovic et al., Joint Bone Spine 2017;84:663–70

ARTICLE INFO

Keywords:
Mucopolysaccharidoses
Morquio syndrome
Skeletal involvement
Joint involvement

We read the valuable manuscript of Mitrovic et al. entitled "Mucopolysaccharidoses seen in adults in rheumatology" [1]. We aimed to contribute their study by drawing attention to mucopolysaccharidoses (MPS) with this case presentation.

An 8-year-old boy was referred to our clinic with pain and limitation in hip movements. His medical history and systemic evaluation was unremarkable. There was no evidence of growth or mental failure. Joints were also normal except slightly reduced hip movements.

In the radiological evaluation, middle-beaked vertebraes in thoraco-lumbar region, rounded iliac wings, capital femoral epiphyseal dysplasia and lower iliac wing tapering in pelvis were observed (Fig. 1).

There was a marked deficiency of galactose-6-sulphatase in his blood sample which is consistent with a diagnosis of MPS IVA. He was referred to the pediatric metabolism clinic for treatment possibilities.

MPS are a group of inherited rare syndromes caused by the absence or malfunctioning of lysosomal enzymes [2]. Especially attenuated phenotypes may be sometimes difficult to recognize for clinicians not familiar with the syndromes. Eight distinct types and several subtypes of MPS have been defined. MPS IV, also known as Morquio syndrome, is consists of two forms (MPS IVA and B) with similar clinical features. Both types can present severe or mild forms, depending upon the amount of residual enzyme activity.

Although several extraskeletal involvements such as corneal opacities, hepatosplenomegaly, valvular heart disease, hearing loss and enamel hypoplasia may be seen, MPS IV is mainly characterized by skeletal involvement. Due to early skeletal signs, the clinicians interested in leukomotor system may sometimes be the first clinician to consider the diagnosis [1]. In some cases, skeletal manifestations may be the only presenting sign providing diagnostic clues [3]. Dysostosis multiplex, short stature, atlantoaxial instability, thoracolumbar kyphosis, pectus carinatum, hip dysplasia, genu valgum are common. Spinal cord compression that can be life-threatening may occur later in the course [4].

Radiographic changes can be observed even prior to clinical manifestations. Most patients exhibit dysostosis multiplex which generally involves malformations of the skull, thorax, spine, pelvis, long bones and hands. Vertebral bodies are typically middle-beaked. The pelvis is often characterized by rounded iliac wings and inferior tapering of the ilea. The long bone deformities include hypoplastic epiphyses (especially capital femoris), proximal humeral notching, narrow femoral necks and thickened short diaphyses. Metacarpals can be short and thick with thin cortices. Also abnormal shaped ribs and enlarged skull can be seen. These typical radiologic manifestations can indicate the presence of MPS and warrant further clinical examination [5].

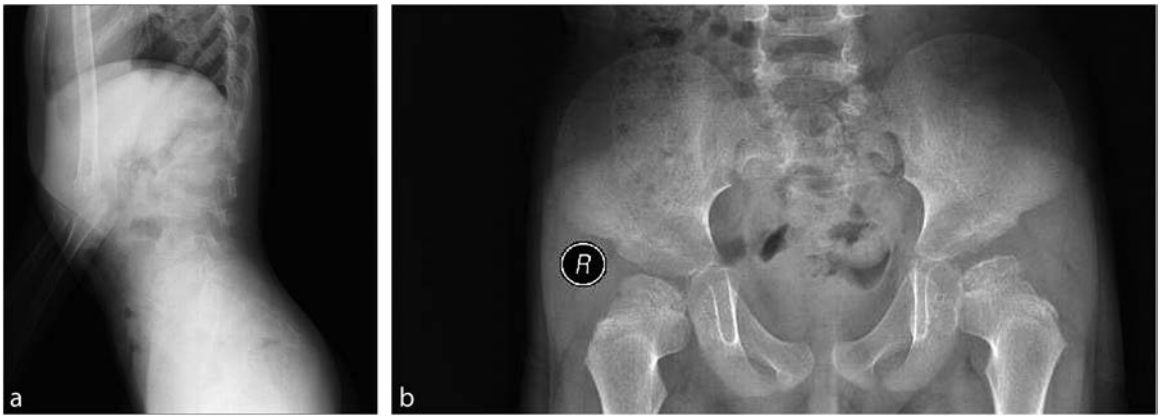

Fig. 1. Middle-beaked vertebraes in thoraco-lumbar region (a), rounded iliac wings, capital femoral epiphyseal dysplasia and lower iliac wing tapering in pelvis (b).

https://doi.org/10.1016/j.jbspin.2018.06.012

The diagnosis of MPS requires demonstration of an abnormal enzyme activity. Enzyme replacement therapy is a therapeutic option and most patients need orthopedic surgical procedures besides rehabilitative approaches.

In conclusion, the cases with MPS IVA, especially in attenuated phenotypes, may be sometimes difficult to recognize for clinicians not familiar with the syndrome. Skeletal signs are generally the earliest features of the disease. An increased awareness of the syndrome among the clinicians interested in leukomotor system will contribute to diagnosis.

Disclosure of interest

The author declares that he has no competing interest.

References

[1] Mitrovic S, Gouze H, Gossec L, et al. Mucopolysaccharidoses seen in adults in rheumatology. Joint Bone Spine 2017;84:663–70.
[2] Melikoglu MA, Kocabas H, Sezer I, et al. Legg-Perthes disease-like joint involvement and diagnosis delay in Scheie syndrome: a case report. Clin Rheumatol 2007;26:1937–9.
[3] Baz AB, Akalin S, Arik H, et al. Proximal realignment surgery for unilateral chronic patella dislocation in Morquio syndrome: a case report. Acta Orthop Traumatol Turc 2011;45:466–9.
[4] White KK. Orthopaedic aspects of mucopolysaccharidoses. Rheumatology (Oxford) 2011;50:26–33.
[5] Lachman R, Martin KW, Castro S, et al. Radiologic and neuroradiologic findings in the mucopolysaccharidoses. J Pediatr Rehabil Med 2010;3:109–18.

Meltem Alkan Melikoglu
Ataturk University, Faculty of Medicine, Physical Medicine and Rehabilitation, Rheumatology Department, Erzurum, Turkey
E-mail address: mamelikoglu@gmail.com