Spondyloepiphyseal Dysplasia Secondary to Morquio Syndrome (Mucopolysaccharoidosis IV): A Case Report

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Morquio syndrome is an autosomal recessive mucopolysaccharoidosis (MPSs) including Type IVA, a deficiency of N-acetylgalactosamine-6-sulfatase and Type IVB a deficiency of β-galactosidase. Plain films of the entire spine, pelvis, chest, knees, hip, and knees demonstrated the characteristic skeletal changes of this disease. The main abnormalities were platyspondyl, genu valgum deformity. Radiographs are crucial to provide substantial information about evolution of the skeletal and joints changes, and the rehabilitation strategies to be followed. From detailed history, physical examination, investigations, and treatment, different clinical, radiographic and biochemical studies it is clear that Morquio’s disease is an inherited error of MPS metabolism characterized by deficiency of galactose 6-sulfate sulfatase activity.

Keywords: Autosomal recessive, Deficiency, Syndrome

INTRODUCTION

Morquio syndrome is a rare inherited disorder of mucopolysaccharide (MPS) catabolism. It is an autosomal recessive MPSs, which includes the Type IV-A (a deficiency of N-acetylgalactosamine-6-sulfatase) and the Type IV-B (a deficiency of beta-galactosidase) resulting in a defective degradation of keratan sulfate.¹ In 1929, Morquio a pediatrician in Uruguay and Brailsford a radiologist in England independently and simultaneously described Morquio-Brailsford syndrome.²

The Morquio syndrome is characterized by severe skeletal changes, which include hypoplasia of odontoid process, short neck, and barrel chest with pectus carinatum, thoracic kyphoscoliosis and dwarfism. Other characteristics are joint laxity, dental abnormalities, acoustic deafness, together with cardiac abnormalities, and respiratory problems.³

Some of the X-ray features of Morquio’s disease include wide flaring of the ilium, shallow acetabula, flattening of femoral heads, coxa and genu valgum deformity. Radiographs are crucial to provide substantial information about evolution of the skeletal and joints changes, and the rehabilitation strategies to be followed. From detailed history, physical examination, investigations, and treatment, different clinical, radiographic and biochemical studies it is clear that Morquio’s disease is an inherited error of MPS metabolism characterized by deficiency of galactose 6-sulfate sulfatase activity.

Case Report

DOI: 10.17354/cr/2015/123

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Access this article online

www.ijsscr.com

Month of Submission : 06-2015
Month of Peer Review : 07-2015
Month of Acceptance : 07-2015
Month of Publishing : 08-2015

Male twins of 10-year-old children who are a product of a consanguineous marriage. Both parents were of normal height and without features of MPSs. Pregnancy and delivery were without complications. According to the history, the early growth and development were normal. By the age of 5 years, their parents noticed deformity of knee joints and took him to a general practitioner who prescribed
some vitamins. Both of them were admitted in the hospital now at the age of 10 years with deformity of both knees (Figure 1a and b).

Physical examination revealed increases dorsal kyphosis, short neck, bilateral genu valgum.

No hypermobility of all the joints was noted. There was no hepatosplenomegaly. Ear, nose and throat examination was normal, cardiovascular examination: Mild systolic murmer and ophthalmological examination was normal. Anthropometry examination was also carried out which is given in Table 1.

**Radiological Examination of Elder Patient**

Skull was normal. Definite flattening of vertebral bodies with beak formation posterior scalloping in the lumbar area was seen. The inter-vertebral disc spaces were wide. Deficient ossification of the superior acetabulae, defective femoral capital epiphyses and marked coxa valga deformity were present. The proximal bases of metacarpals 3-5 were conical with normal construction of metacarpal shafts. The ossified carpal bones were small and reduced in number as compared with the patient’s age. Broadening of the anterior portion of the ribs was also seen (Figures 2-5).

**Biochemical Analysis**

Accumulation of keratan sulfate and chondroitin 6-sulfate in Morquio syndrome is due to a deficiency of galactose 6-sulfate sulfatase and N-acetyl galactosamine 6-sulfate sulfatase activity, which are necessary for the degradation of these two MPS. We found there was a deficiency of beta galactose 6-sulfate sulfatase.

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<th>Table 1: Anthropometry examination</th>
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**DISCUSSION**

Morquio syndrome is a member of a group of inherited metabolic disorders collectively termed MPS. The MPS are caused by a deficiency of lysosomal enzymes required for
the degradation of MPS or glycosaminoglycans. Currently, eleven distinct single lysosomal enzyme deficiencies are known to cause seven recognized phenotypes. MPS inherited in an autosomal recessive fashion, except Hunter syndrome which is X-linked.\(^4\)

The MPSs share a chronic progressive course with multisystem involvement, several physical features, laboratory findings, and radiographic abnormalities.\(^5\)

Patients with Morquio syndrome usually can be clinically distinguished from patients with other MPSs because they do not have coarse facial features or mental retardation and they have additional skeletal manifestations derived from a unique spondyloepiphyseal dysplasia and ligamentous laxity.\(^6\) These skeletal manifestations include odontoid hypoplasia, a striking short trunk dwarfism, and genu valgus. The patients with Morquio syndrome tend to have greater spine involvement with scoliosis, kyphosis, and severe gibbus, as well as platyspondyly, rib flaring, pectus carinatum, and ligamentous laxity. Odontoid hypoplasia is the most critical skeletal feature to be recognized in any patient with Morquio syndrome.\(^1\)

The exact incidence is unknown. Development of newborn screening strategies is underway. A very limited number of studies have been performed through the years regarding the skeletal status of patients with Morquio syndrome and only in very young people. The underlying defect in the MPSs is inability to degrade glycosaminoglycans. Dermatan sulfate, heparan sulfate, keratan sulfate, and chondroitin sulfate are recognized as the main glycosaminoglycans present in the tissues. Keratin sulphate is predominantly found in the cartilage and cornea, the major organs affected in Morquio syndrome.\(^7\) Heparan and dermatan sulfate have a more generalized tissue distribution. Their normal metabolism in patients with Morquio syndrome spares these patients from mental retardation and disease manifestations observed in other types. The specific mechanism by which excess storage of keratin sulphate results in the skeletal dysplasia unique to Morquio syndrome remains unknown.\(^2\)

Although the diagnosis of Morquio syndrome is based on physical findings, blood enzymes, radiographs and magnetic resonance imaging provide useful information about the gravity of characteristic of skeletal and joint changes.

The commonest cause of death which usually occurs around the third to fourth decade is due to cor pulmonale, valvular heart disease or myelopathy. Genetic counseling is advisable.\(^8\)

The preservation of functionality is an increasing challenge in the treatment of patients with Morquio syndrome and maintenance of occupational performance should be defined as one of the main goals to be reached by the therapies used.\(^5\)

**CONCLUSION**

We would like to stress the uniqueness of our imaging collection. We believe that for the study of chronic progressive course with multiple joint involvement of the Morquio disease, subsequent radiographic assessments can provide useful information, giving the patients a substantial impact about the evolution of this pathologic condition and planning the required treatment modality.

**ACKNOWLEDGMENT**

All the contributors would like to thank the entire Department of Orthopedics which worked as a team in making the diagnosis and planning the treatment procedures done for the patient.

**REFERENCES**


Source of Support: Nil. Conflict of Interest: None declared.