MORQUIO SYNDROME: A CASE REPORT

Palwasha Gul,[1] Brig Imran Masaud,[1] Zeenat Gul[2]

[1] Department of Radiology, Combined Military Hospital (CMH), Quetta, Pakistan.
[2] Resident, Department of General Medicine, Bolan Medical College Hospital (BMCH), Quetta, Pakistan.

PJR January - March 2014; 24(1): 32-35

ABSTRACT

Morquio syndrome is an autosomal recessive condition and is mucopolysacchroidosis (MPS), type IV. It is a rare clinical condition and results from an excess of keratin sulphate in cells from a deficit in its degradation pathway. Clinical features include severe dwarfism, joint laxity, deafness, short neck, kyphoscoliosis, corneal clouding, prominent mandible and lower face and lymphadenopathy. Radiographic features show platyspondyly, atlantoaxial subluxation, os odontoideum, vertebral body beaking, hypertelorism, dolicocephaly, multiple epiphyseal centres and wide metacarpals. Life expectancy ranges from 30 - 40 years, but depends on severity of abnormalities present as some may die as early as 2 or 3 years, while others can live upto 60 or 70 years. Most common cause of death is cervical myelopathy from C2 abnormality. Patients are vulnerable to respiratory infections. We are presenting a case of Morquio Syndrome and to the best of our search this is the first reported case in a national journal.

Key words: Morquios Syndrome (MS), Mucopolysacchroidosis (MPS), Autosomal recessaive

Introduction

Morquios Syndrome was first described in 1929 by Luis Morquio, a Uruguayan physician and James Frederick Brailsford, an English radiographer.[1] Diagnosis is based on clinical examination, radiographs, urinary GAG tests, and the enzymatic activity of N- acetylgalactosamine-6 sulfatase in blood cells or fibroblasts. Multidisciplinary approach is required for the care of the patient.

At birth the patient appears healthy but as adulthood starts, skeletal and extraskeletal manifestations begin to emerge. Skeletal features include severe dwarfism, joint laxity, short neck, kyphoscoliosis, prominent mandible and lower face, platyspondyly, atlantoaxial subluxation, os odontoideum, vertebral body beaking, dolicocephaly, multiple epiphyseal centres and wide metacarpals, knock knees, bell shaped chest, duck waddling gait.[2]

Extraskeletal features include coarse facial features, prognathism, a broad mouth, a short nose with anteverted nares and a flat nasal bridge, widely spaced teeth with thinness of tooth enamel and carries and macrocephally.[3] Other features include aortic valve incompetence with regurgitation with hepatosplenomegaly, abdominal hernias, neuro-sensory deafness, clouding of cornea, pigmented degenerative retinal lesions or glaucoma. Pulmonary complications includes restrictive disease due to kyphoscoliosis resulting in decreased lung volumes, ventilation - perfusion mismatching and obstructive sleep apnea, which can result in pulmonary hypertension and cor pulmonale. Characteristic abnormalities include anterior hypoplasia of T12, L1 or L2 which may give

Correspondence : Dr. Palwasha Gul,
Post graduate resident, Diagnostic radiology,
Combined Military Hospital, Quetta, Pakistan.
Cell: 03003884472
E-mail: gul_l123@yahoo.com

Submitted 01 February 2015, Accepted 12 February 2015

rise to kyphosis. Intelligence is normal and the trunk is short with proportionately long limbs.[3,4]

Case Report

We report a case of 20 years old male who started developing characteristic skeletal features at the age of one and a half year. He was born normally with no antenatal, intrapartum and postnatal complications. Parents and other brothers and sisters are normal. With growing age, patient developed short stature, abnormal gait, pectus carinatum and kyphosis. He was attending school normally with no learning difficulties. Then slowly there was worsening of gait and neck pain and over a period of last three years he developed quadriplegia. Imaging findings showed pectus carinatum, platyspondyly (Fig. 1), os odontoideum (Fig. 2), widening of foramen magnum, basilar invagination with atlantoaxial subluxation and cervical spinal stenosis at the level of C2, causing kinking and compression of cervicomedullary junction, metacarpals with proximal pointing, irregular carpal bones and distal end of radius and ulna pointing towards each other (Fig. 3). The patient had no surgical history but was on symptomatic treatment since childhood. Due to quadriplegia, the patient has been under consideration for surgery and a discussion has been going on among neurosurgeons, anaesthetists, radiologists and family is being counseled about the associated complications of Morquio Syndrome. At the time of reporting this report, patient is on conservative support.

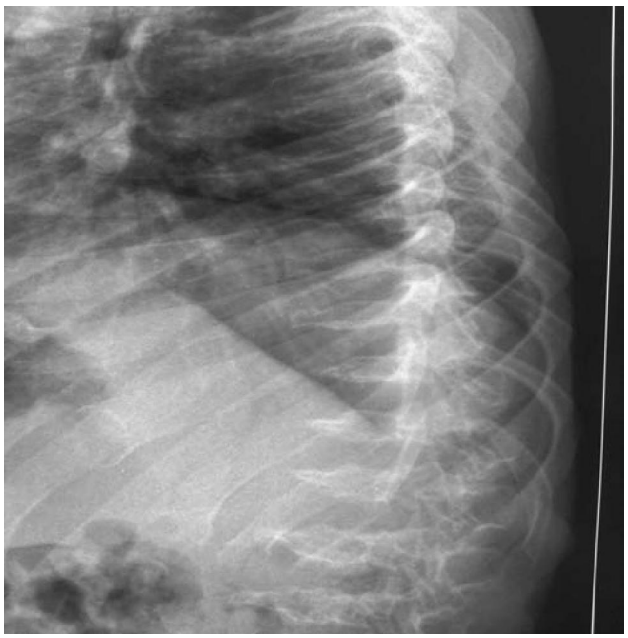

Figure 1: X-ray dorsolumbar spine showing platyspondyly with anterior beaking.

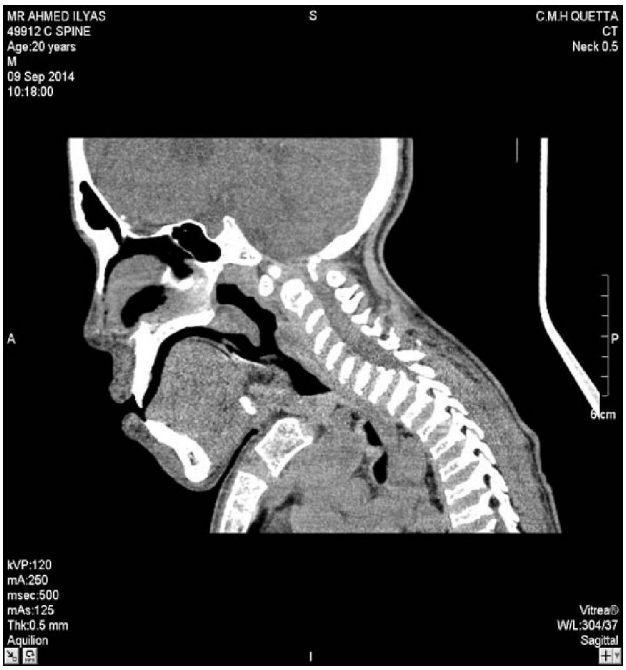

Figure 2: CT (sagital image) cervical spine shows unfused and hypoplastic odontoid process of C2. Visualized cervical and thoracic vertebra shows platyspondyly.

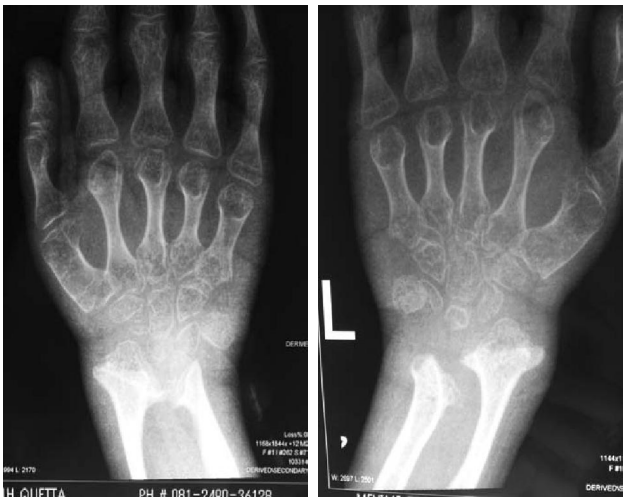

Figure 3: X-ray wrists show metacarpals with proximal pointing (4th metacarpal), irregular carpal bones and distal end of radius and ulna pointing towards each other.

Discussion

Morquio syndrome is an autosomal recessive lysosomal storage disorder with type A and B subtypes caused by reduced activity of one of the two lysosomal hydrolases, N acetyle galactosamine - 6 - sulphatase and B - galactosidase respectively. Patient often die in late childhood or early adulthood from neurological disability or cardiac complications.[4,5] Due to the wide variation and subtleties of the radiographic findings, imaging of multiple body regions is recommended. The accurate diagnosis rests on a thorough clinical evaluation and multiple laboratory measures.

Dalive et al[6] reported four children with Morquio syndrome who had neurological deterioration related to a progressive thoracolumbar gibbus. X-ray in erect position showed kyphosis and canal compromise. Anterior decompression and correction of kyphosis was done and the patients survived the complications. Chen et al[7] reported three cases of Morquio syndrome over a period of ten years, they had distinctive bony changes, as vertebra plana and tongue like protrusion in the anterior part of the lower thoracic and upper lumbar vertebral bodies, particularly short distal deformed ulna and poor ossification of the proximal lateral tibial epiphysis. Definitive diagnosis depends on enzyme analysis.

According to Takeda et al,[8] compression of the spinal cord due to atlantoaxial subluxation was diagnosed in a patient with Morquio syndrome by cervical radiography and MRI. 15 year old patient had no neurological symptoms however spinal cord compression was noted at C1-C2. Hypoplasia of the dens is a common and severe manifestation causing atlantoaxial instability, compression of the cervical spinal cord and complications during endotracheal intubation. Tetraplagia of acute onset following atlantoaxial subluxation during general anesthesia. When an operation is to be undertaken, it is important that hypoplasia of the dens of the axis vertebra entails a high risk of vertebral disc dislocation and spinal cord damage especially during general anesthesia when excessive movements of the head may occur.[9,10]

The treatment of Morquio Syndrome consists of prenatal identification and enzyme replacement therapy. There are approximately 800 patients of this syndrome in United States. Federal Drug Administration (FDA) has approved Elosulfase Alfa (Vimizim) for treatment of the Moquio syndrome. Vimizim is intended to replace the missing GALNS enzyme involved in an important metabolic pathway. The treatment is more effective if started before five years of age.[11]

References

1. Morquio L. Sur une forme de dystrophie osseuse familial. Arch Med Enfentes 1929; **32:** 129-40.

2. Mopagar VS, Kathariya MD, Umapathy T, Premkishore K, Chandrashekar, Raurale A. Morquio's syndrome: A rare case report. Journal of Indian Academy of oral Medincine and Radiology, 2013; **25:** 63-5

3. Fitzgerald J, Verveniotis SJ. Morquio's syndrome. A case report and review of clinical findings. N Y State Dent J. 1998; **64:** 48-50.

4. Rekhi GS. Morquio Syndrome (MPV IV) _A case report. Indian J Ophthalmol 1991; **39:** 78-81

5. Wood TC, Harvey K, Beck M, Burin MG, Chien YH, Church HJ et al.Diagnosing mucopolysacchridosis IVA. J Inherit Metab Dis. 2013; **36:** 293-307.

6. Houten JK, Kinon MD, Goodrich JT. Morquio's syndrome and craniocervical instability. Pediatr Neuro Surg. 2011; **47:** 238-40.

7. Dalvie S, Skinner J, Vellodi A, Noorden MH. Mobile thoracolumbar gibbus in Morquio type A: the cause of paraparesis and its management. J Pediatr Orthop B. 2001; **10:** 328-30.

8. Chen SJ, Li YW, Wang TR, Hsu JC. Bony changes in common mucopolysacchroidosis. Zhogua Min Guo Xiao Er Ke Yi Xuo Hui Za Zhi. 1996; **37:** 178-84.

9. Takeda E, Hashimoto T, Tayama M, Miyazaki M, Shirakawa E, Shiino Y et al. Diagnosis of Atlantoaxial Subluxation in Morquios Syndrome and spondyloepiphyseal Dysplasia Congenita. Pediatric International 1991; **33:** 633-8.

10. Beighton P, Craig J. Atlantoaxial subluxation in the Morquio syndrome. Bone and Joint Surgery J 1973; **55:** 478-9.

11. http://www.fda.gov/NewsEvents/Newsroom/Pressannouncements/ucm 386008.htm