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Case Report

Metatarsalgia in Morquio A Syndrome

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Abstract

Morquio A Syndrome, also known as Mucopolysaccharidosis type IV (MPS IV), is a lysosomal storage disease of autosomal recessive pattern. The disease is rare and features musculoskeletal and cardiopulmonary complications. We present a case study of Morquio A Syndrome with pes planus and metatarsus elevatus. The patient had a long history of metatarsalgia and a severe antalgic gait. Upon investigation, it was discovered that the deformity was caused by ligamentous deforming forces of the foot, linked to ligamentous restrictions seen in Morquio A Syndrome. An overload of the metatarsal heads leads to subsequent 1st metatarsal base subluxation and 2nd and 3rd rigid hammer toes. Surgical correction was done by a Lapidus procedure and simultaneous correction of the metatarsus elevatus deformity of the 1st ray, as well as a flexor hallucis longus transfer to the extensor hallucis longus. The procedure was successful in terms of pain relief and return of functional level.

Keywords: Metatarsalgia; Morquio A Syndrome

Background

Morquio A Syndrome is a genetic disorder of polymucopolysaccharide catabolism. The disorder is of the autosomal recessive mucopolysaccharidosis variant (MPS), including the IVA type and the IV B type. The type IVA variant is due to N-acetylgalactosamine-6-sulfatase deficiency, while IV B type is attributed to deficiency of beta galactosidase, both leading to degradation of keratin sulphate [1].

An independent description of Morquio-Brailsford Syndrome was given by A Uruguayan pediatrician, Morquio, and an English radiologist, Brailsford, in 1929 [2]. Husler in the 1930s came up with the name Dysostosis Multiplex, a term that described skeletal findings that were characteristic of MPS patients and patients with other disorders of lysosomal storage [3]. The skeletal manifestations present in the form of, genu valgum, pigeon or barrel chest featuring pectus carinatum, joint laxity, and kyphoscoliosis [3].

Other major characteristics of Morquio A Syndrome include the extreme changes to skeletal structure, including odontoid process hypoplasia, short neck and dwarfism [4]. Other features of this disorder include joint laxity, acoustic deafness, abnormalities of the dentition, respiratory disorders, and cardiac abnormalities. X-ray examination of Morquio A Syndrome shows shallow acetabula, ilium flaring, flattened femoral heads, Dysostosis Multiplex, genu and coxa valga. Spinal skeletal disorders include kyphosis, absence of the odontoid process, and platyspondyly with central beaking [5]. Clinical manifestations of the type IVA variant was believed to be more severe than type IV B. Patients with the type IVA variant have a life expectancy of no more than three decades. However, there have been records of isolated cases of prolonged survival [6]. Patients who make it into adulthood, mainly have a C1 ring and odontoid peg ossification [7].

Diagnosis is based on radiological and physical features, skin biopsy, and enzyme blood levels of N-acetylgalactosamine-6-sulfatase (GALNS) [7]. Confirmation is done when high keratin sulphate levels are seen in the urine. With the exception of Allman and Kitzing, whose report highlighted scintigraphy features showing anthropomorphic skeletal uptake patterns consistent with a mucopolysaccharide storage disease in a Morquio A Syndrome case, there have been no other recent publications on the systematic collection of imaging of this rare disorder [8].

Orthopedic care might be a vital component of management of Morquio A Syndrome, but treatment and surgical guidelines remain scarce. Guidelines for the diagnosis and management of spinal involvement in this disorder were recently published [9]. However, there is very little evidence in the literature to direct the management of the extremity abnormalities and their consequences, in

patients with this disorder. With the goal of improving patient prognosis, this article reviews a case in which we surgically correct a forefoot driven flatfoot deformity with a metatarsus elevatus, which was incidentally discovered in a Morquio A Syndrome patient. With regards to the patient's tibialis posterior tendon in the patient's effected foot, she did have a hindfoot valgus with the single heel raise test.

Case Presentation

A 29-year-old lady presented to the foot clinic with right foot metatarsalgia for over 2 years. The patient has been diagnosed with Morquio A Syndrome as a young adult. She had otherwise been fit and well. The patient was the 1st child of a consanguineous marriage. The family of the patient had no history of Morquio A Syndrome. Physically, she had a short stature with thoracic kyphosis. She also had a severe antalgic gait. The sagittal plane showed a forward pelvic tilt with both knees and hips in flexion. The right foot had an obvious flatfoot deformity with plantar subluxation of the 1st MTP joint (Figure 1). Upon further examination, overlapping of the 5th toe was noticed in the same foot but it was not bothering the patient (Figure 2). She had a Beighton Hypermobility Score of 5. She fulfilled two major criteria of the benign joint hypermobility syndrome. The patient had completed a Foot and the American Orthopedic Foot and Ankle Score (AOFAS) [6] on 3 separate occasions, her average score was 26.5. The silfverskiold test was negative.

Figure 1: Simulated weight-bearing of the foot showing a mild hindfoot valgus, collapsed medial arch and a 1st Metatarsal bone in dorsiflexion.

Figure 2: Forefoot adduction with the 1st metatarsal phalangeal joint in plantar subluxation.

Investigations

At the time of referral to clinic the patient was already diagnosed with Morquio A Syndrome and had already been subjected to genetic studies. Therefore anteroposterior (AP), oblique, lateral and Saltzman hindfoot radiographical views of the foot were obtained for clinical correlation. From the AP view (Figure 3), the talocalcaneal angle was 17°, the talonavicular angle was 9°. In the lateral radiograph (Figure 4), Meary's angle was 36° and the calcaneal pitch was about 15°. Calcaneonavicular coalitions were not observed in the oblique view.

The radiological investigations were in keeping with a flat foot deformity associated with 1st MTP joint planter subluxation as well as 2nd and 3rd rigid hammer toes.

Treatment

The symptoms persisted with minimal improvement on non-surgical management. Surgical intervention was indicated at this point, so she underwent a 1st metatarsal Lapidus procedure with an attempt to correct the metatarsus elevatus (Figures 5-8) using a direct medial approach and fixed with a plate and screws. The patient also underwent a Flexor Hallucis Longus to the Extensor Hallucis Longus transfer (Figure 9). The patient was kept in a below knee back slab for 6 - 8 weeks then shifted to an aircast boot with gradual weightbearing, starting at 25% in the first week, proceeding to 100% by week 4.

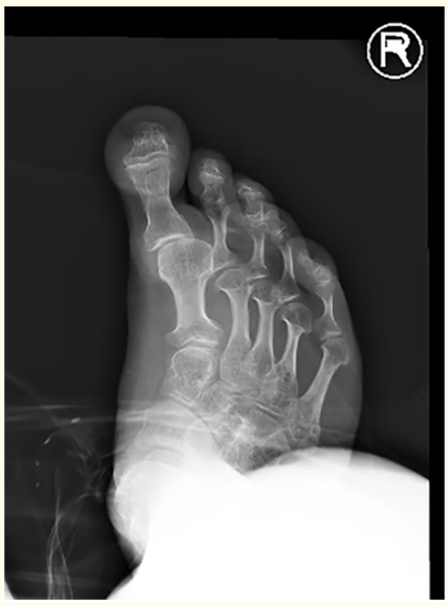

Figure 3: The weight-bearing view on an AP radiograph of the foot.

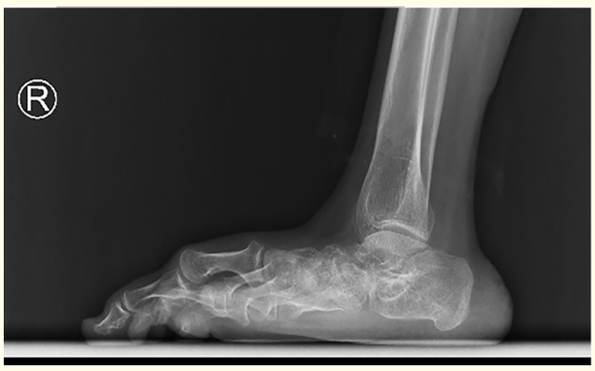

Figure 4: The lateral weight-bearing view seen on the lateral radiograph.

Figure 5: 1st Metatarsal Lapidus procedure with correction of the metatarsus elevatus.

Outcome and follow-up

The patient filled the AOFAS pain score form on each post-operative follow-up visit. The patient reported feeling no pain at rest.

Figure 6: AP intra-operative image after correction and fixation of the 1st metatarsal phalangeal joint with a plate and screws.

Figure 7: Lateral intra-operative image after correction and fixation of the 1st metatarsal phalangeal joint with a plate and screws.

Figure 8: Intra-operative clinical photograph with simulated weight-bearing to check for the 1st metatarsal head touching the floor.

Figure 9: Harvested Flexor Hallucis Longus tendon.

However, the patient complained of some pain after walking for long hours. Her gait has improved significantly. She assists with household chores and has better control over her activities of daily living.

At 18 months post-deformity correction, the patient had scored an average of 76.4 on the AOFAS pain score. She is compliant with her ankle strengthening exercises at home, and she will not require any further surgical treatment for this deformity (Figure 10).

Figure 10: Simulated weight-bearing view post-operatively at 18 months. Reduction of the 1st metatarsal phalangeal joint is noted, with the head in contact with the floor and the restoration of the medial arch.

Discussion

Morquio A Syndrome belongs to a collection of metabolic disorders known collectively as Mucopolysaccharidoses. The underlying pathophysiology is lysosome deficiency necessary for glycosaminoglycans or mucopolysaccharide degradation. Presently, seven identified phenotypes of mucopolysaccharidoses have been linked to deficiency of eleven unique single lysosomal enzyme [10]. Inheritance of all mucopolysaccharidoses is autosomal recessive, with the exception of X-linked Hunter syndrome [11].

Progression of MPS's is characterized by multisystem involvement, laboratory findings, radiographic abnormalities, and several physical features [5].

Morquio A Syndrome patients are differentiated from other MPS's patients clinically, because they are not mentally retarded, nor do they lack coarse facial characteristics. As a plus, they have an extra skeletal structure originating from ligamentous laxity and spondyloepiphyseal dysplasia [4,12]. The manifestations on the skeletal system include genu valgus, a unique dwarfism (short trunk) and odontoid hypoplasia. Morquio A Syndrome patients have more spine involvement with kyphosis, scoliosis, and a severe form of gibbus, together with rib flaring, platyspondyly, ligamentous laxity, and pectus carinatum [13,14]. Odontoid hypoplasia is a sinister skeletal structure that should be identified in Morquio A Syndrome patients. It suffices to say that there is a primary relationship between morbidity, mortality, and atlantoaxial subluxation due to the unstable nature of the odontoid process [13].

The globally estimated incidence of Morquio A Syndrome has wide coverage: for Northern Ireland, the incidence occurs at 1 case in 75,000; for British Columbia, it is 1 case in 200,000; while Germany records 1 case in 263.157 [15]. The Italian incidence is not known although the Italian Association of Mucopolysaccharidosis estimates 1 case in 1250000 live births (non-published data). Screening strategies for new-borns are being developed. Very few researches have been done on the status of the skeletal structure of patients with type IV B Morquio Syndrome patients [5,15].

The major pathology in MPS's is the failure of glycosaminoglycans to be degraded. The major glycosaminoglycans that are present in tissues are Dermatan sulphate, keratan sulphate, heparin sulphate, and chondroitin sulphate. Degradation of keratin sulphate in Morquio syndrome IV B is defective due to deficiency of the beta-galactosidase (GLB1) gene. Keratin sulphate is mostly found in the cornea & cartilage, two organs that are the most affected in Morquio A Syndrome [10].

Tissue distribution of dermatan and Heparan sulphate is more generalized. Metabolism of these glycosaminoglycans in the patient spares him or her the trauma of mental retardation and the manifestation of disorders that are characteristic of other types of MPS's. We do not know the major reason why accumulation of keratin sulphate triggers the skeletal dysplasia that is characteristic of Morquio Syndrome [10].

Many investigations are needed to make a diagnosis of Morquio A Syndrome, such as a skin biopsy, blood enzymes, MRI, and radiographs assist with vital information about the characteristic of joint and skeletal changes. A critical recognizable feature is the odontoid hypoplasia and this explains why a cervical spine MRI must be used to ascertain whether or not the upper vertebrae are underde-

veloped, thus assisting in accurate diagnosis of neurological risks conditions. It also serves as a confirmatory for cord compression and cervical myelopathy.

In our case, MRI of the lower limb failed to identify any osteonecrosis of the distal and proximal femur as is characteristic of some patients with Morquio A Syndrome. Our patient had an improved gait and barely felt any pain post-operatively, an indication that her condition was generally satisfactory. At 18 months post deformity correction, the patient had scored an average of 76.4 on the AOFAS pain score. She is compliant with her ankle strengthening exercises at home, and she will not require any further surgical treatment for this deformity. Sustenance of functionality is a major challenge in Morquio A Syndrome therapy. Maintaining occupational performance should be a major aim of the therapies in use [16].

Pes planus is a deformity in the foot, seen as a combination of a collapsed arch, hindfoot valgus such as the deformities that were seen in this case [17]. To the authors knowledge a pes planus deformity has not been related among the clinical features of Morquio A Syndrome [18].

It is pertinent to state that assessments with MRI imaging can offer very vital information with respect to the progressive course of Morquio A Syndrome, making a substantial impact on the patients with respect to the evolution of its pathology. The rheumatologist can also play a role in ensuring that pain symptoms (acute and chronic) are controlled, while the physiatrist monitors joint and bone function, adopting the necessary rehabilitation techniques to be employed.

Conclusion

Morquio A syndrome has many soft tissue and skeletal manifestation. In this particular case, the mucopolysaccharide deposition in the patient's ligaments has caused a pes planus and a metatarsus elevatus deformity which needed surgical correction. The surgical intervention was successful in restoring the patient's quality of life as well as significantly reducing her pain levels.

Learning Points

- Syndromic clinical manifestations can vary, it is important to lower your suspicion threshold to find the culprit.
- Surgical Reconstruction of a neglected pes planus foot has been proven effective.
- A balance between soft tissue as well as bony reconstruction should be tailored for each individual's needs.

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