Medicine & Clinical Science

Case Report

Mucopolysaccharidosis Type IVa (Morquio Syndrome) Clinical Features and Management: Case Report

Fouad Althobaiti[1], Shatha Alkhulaisi[2], Muaath Alzahrani[3]

[1]Consultant, Pediatric Dentistry, Ministry of Health, Taif Specialized Dental Center, Taif, Saudi Arabia.
[2]Resident, Saudi Board of Pediatric Dentistry, Ministry of Health, Taif Specialized Dental Center, Taif, Saudi Arabia.
[3]General Dentist, Department of Pediatric Dentistry, Ministry of Health, Taif Specialized Dental Center, Taif, Saudi Arabia.

Correspondence

Fouad Althobaiti

Consultant, Pediatric Dentistry, Ministry of Health, Taif Specialized Dental Center, Taif, Saudi Arabia.

Abstract

Mucopolysaccharidosis type IV A (MPS IV A), often referred to as Morquio syndrome, is a rare genetic disease characterised by an autosomal recessive deficiency in N-acetylgalactosamine-6-sulfatase (GALNS), which results in the accumulation of glycosaminoglycans within cells. Treating individuals with this condition with enzyme replacement therapy has proven to be essential. Radiographic imaging, clinical evaluation, and biochemical testing are used to diagnose MPS IV A. Notable skeletal anomalies, aberrant joint mobility, severe growth impairment, dental misalignment, and defects in the enamel are the characteristics of the disorder. This clinical case report details the dental care provided to a seven-year-old Saudi Arabian girl with many bone anomalies.

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Keywords

Mucopolysaccharidosis, Morquio syndrome, Skeletal abnormalities, Lysosomal storage disorder, Glycosaminoglycans.

Introduction

Mucopolysaccharidoses (MPS) are a subclass of rare lysosomal storage diseases that are genetically inherited. Gene diseases cause deficiencies in or absence of the enzymes necessary for the breakdown of glycosaminoglycans (GAGs). The primary cause of MPS is believed to be undegraded GAGs, and GAG accumulation in cells can have secondary and tertiary effects such as autophagy, mitochondrial dysfunction, and apoptosis [1]. Key features include skeletal difficulties such as short-trunk, dwarfism, dental problems, and possibly heart concerns [2].

Morquio syndrome is made up of two conditions: MPS IV B, which is caused by a deficiency in beta-galactosidase, and MPS IV A, which is caused by mutations in the galactosamine-6-sulfatase genes [3]. Seven clinical categories are among the several subtypes of MPS [4].

The intralysosomal accumulation of two GAGs, chondroitin-6-sulfate and keratan sulphate, which impairs chondrocyte function in the cartilage, bone, and ligaments, is the characteristic that distinguishes MPS type IV A. This accumulation is caused by mutations in the gene encoding N-acetylgalactosamine-6-sulfate sulfatase (GALNS) [5]. Unlike other MPS, affected children frequently have normal IQ [6].

While the front teeth are widely separated and flared, the maxillary posterior teeth have sharp cusp ends and are tapering. In other circumstances, the enamel may be thinner and have pits even though the hardness may be normal [7]. Maintaining good oral health may be more challenging in cases of upper extremity malformations as well as teeth [8].

The purpose of this case report was to report the systemic and orofacial features of Morquio A syndrome in a 7-year-old female pediatric patient, along with detailing the dental care she received with follow-up.

Case presentation

A seven-year-old Saudi female patient was referred to the Taif dentistry Center's paediatric dentistry clinic for a routine oral examination. a documented instance of type IV A mucopolysaccharidosis. Since the patient's older sister also suffers from the same skeletal disorder, there is a family history of the same skeletal disorder. Genetic studies verified the diagnosis; the recessive gene accounts for 25% of all pregnancies with reassurance.

The patient has a normal IQ and has been diagnosed with complicated medical conditions. Upon clinical examination, the endocrinologist recorded that the patient had hepatosplenomegaly, extending 2 cm below the costal border. The orthopedist's examination indicated that she had bone deformities in the ribs, chest, hips, and wrists, which contributed to her low stature and other skeletal abnormalities. The cardiologist's echocardiogram revealed a bicuspid aortic valve. She had flexible joints but other anomalies included stubby hands, kyphosis, twisted legs, knock-knees, and a

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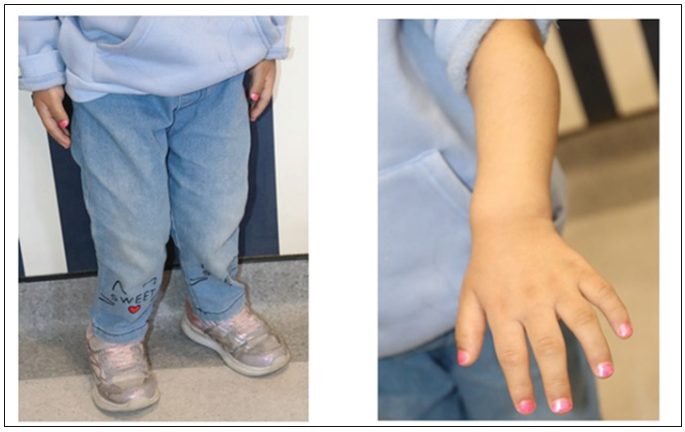

Figure 1. *Knock-knees, stubby hands, short forearm of the presented case*

waddling gate (Figure 1). Ophthalmologists also recorded issues with eyesight and corneal opacity.

The patient exhibits characteristic craniofacial traits, such as a small nose, broad mouth, coarse face with a brachycephalic convex profile, and a flat nasal bridge, in addition to these skeletal anomalies (Figure 2). The patient's weight was recorded at 15.6 kilograms, and her height measured 90 centimeters. Both weight and height bellow 50 percentile of her age (Figure 3).

The radiograph of the hand and wrist shows a number of interesting features, including small and irregular carpal bones, short and widened metacarpal bones with pointed proximal ends of the second to fifth digits, and short ulna and radius with bowing deformity associated with metaphyseal flaring and mild tilting of the distal radius and ulna towards each other. Furthermore, the proximal and middle phalanges have a distal rounded tapering, and there is irregularity in the metacarpal epiphyses (Figure 4).

Regarding the intraoral examination, radiographs showed that the teeth's enamel layer was quite thin. She also had open bite, spaced teeth, sharp cusps, hypoplastic pitted thin enamel, and a rough texture. She had mesial step molar relation on both sides, mixed dentition, and caries in every primary molar (Figure 5).

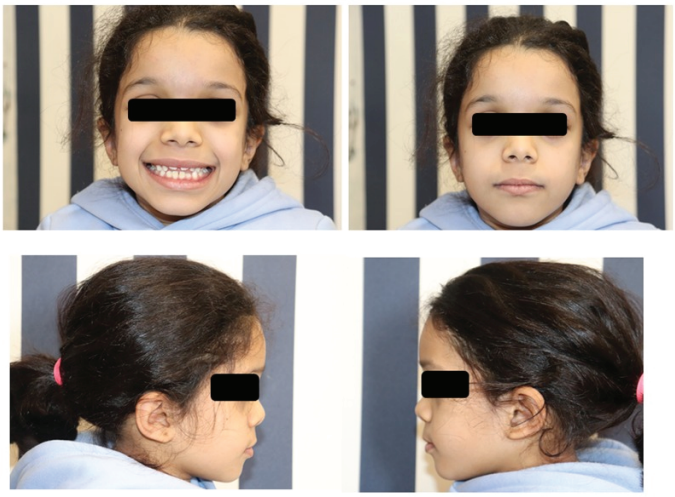

Figure 2. *Craniofacial traits of the presented case*

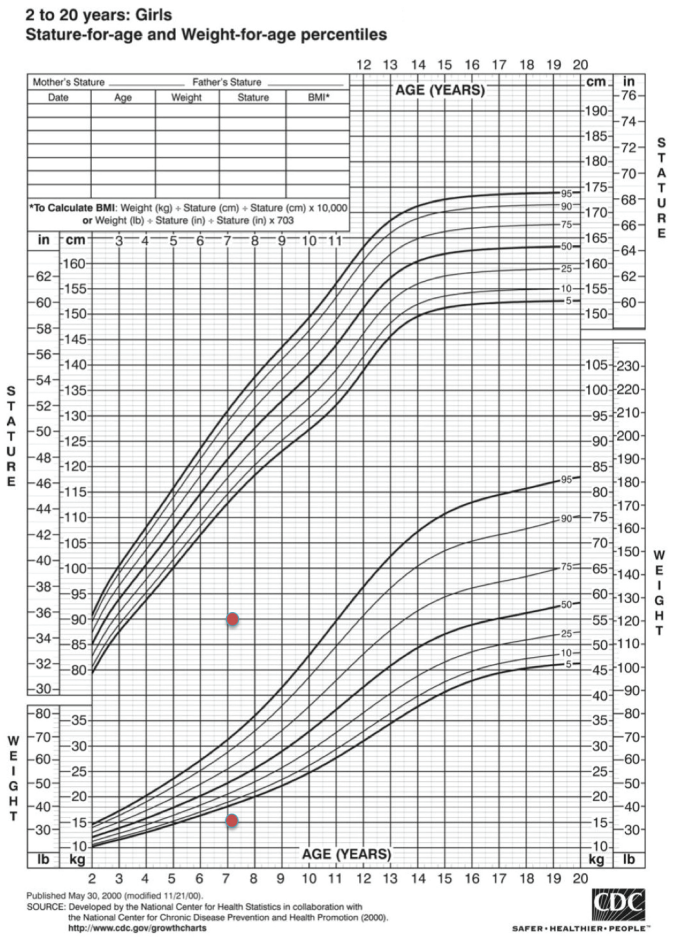

Figure 3. *Stature for age and weight for age percentile*

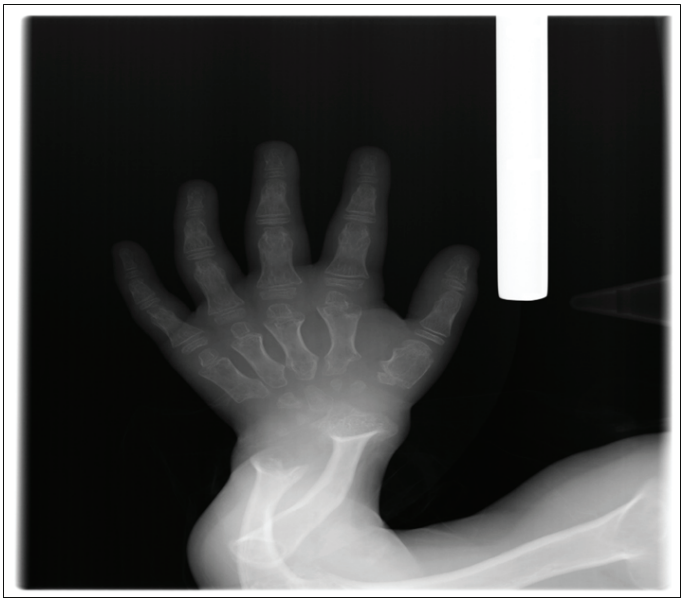

Figure 4. *Hand wrist radiograph showing distal rounded tapering of the proximal and middle phalanges and irregularity of the metacarpal epiphyses*

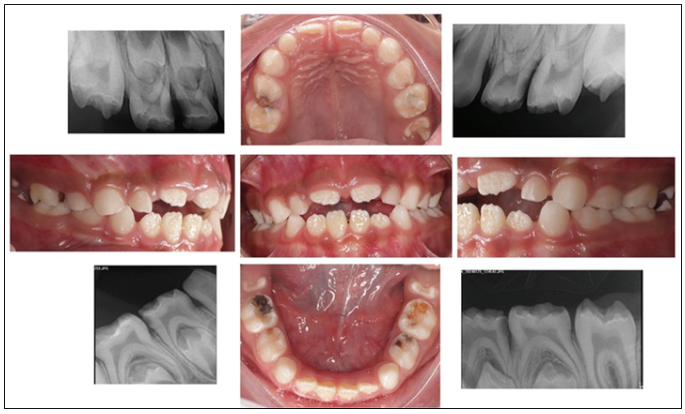

Figure 5. *Preoperative photographs and radiographs showing mesial step molar relation on both sides, mixed dentition, and caries in every primary molar*

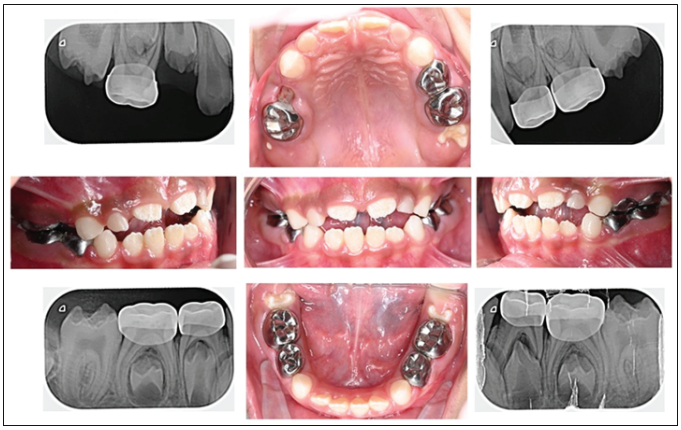

Figure 6. *Postoperative photographs and radiographs*

As determined by the medical staff, the patient began enzyme replacement treatment. The patient experienced an adverse reaction to the treatment; therefore, the doctors were forced to stop the enzyme replacement therapy. Because of her extensive dental treatment history and her refusal to cooperate, the patient requires full-mouth dental rehabilitation under general anesthesia. All features and results of the treatment were explained to the parents, along with the possible dangers and benefits of general anesthesia. The consent form was signed and discussed with the parents. In addition to laboratory testing, consultations with a pediatrician, anesthesiologist, and cardiologist were undertaken. The patient was scheduled for general anesthesia on the day of operation after receiving approval from the doctors. The patient had complete mouth dental rehabilitation in the operating room. Following dental treatments performed in the operating room, the patient is moved to the post-anesthesia care unit (PACU) for observation and recuperation. This method guarantees the safety of the necessary dental procedures and the preservation of the patient's well-being at all times. During the follow-up visit, radiographs and postoperative photos were acquired (Figure 6).

Discussion

Patients with mucopolysaccharidosis type IV (MPS type IV) have several musculoskeletal problems. This is a rare condition. The Morquio A syndrome (MAS) is an uncommon autosomal recessive condition that is multisystemic and progressive in nature. There are variants A and B in this scenario. Unlike type A, type B, which is linked to β-galactosidase deficiency, frequently lacks enamel hypoplasia [9].

The majority of individuals with Morquio syndrome do not show any symptoms until the last quarter of their first year of life; however, as they become older, they have atrophy of the muscles, pectus carinatum, and knock knees [10].

Dental manifestations include defects in the enamel of primary and permanent teeth. Enamel is characterized by its thinness, fragility, and flakiness. Gardner reported that although the enamel only amounted for approximately one-third of its regular thickness and its radiodensity was normal [11].

Enzyme replacement therapy is one method of treating patients with Morquio syndrome. [12] However, the cost of this treatment plan is high. Hematopoietic stem cell transplantation offers many benefits for physical activity. Therefore, HSCT improves the quality of life for MPS patients; the therapeutic impact on abnormalities of the bones is yet unclear [13].

The majority of severe MPS IVa cases end in death from cardiac or pulmonary problems within 25 years [14].

Conclusion

Currently, MPS is a significant medical disorder that has to be swiftly evaluated and treated. The relevant physical, craniofacial, and dental characteristics unique to MPS must be understood by all dentists in order to facilitate early detection and proper oral management. To guarantee accurate diagnosis, a multidisciplinary healthcare team needs to be included. To avoid this syndrome developing in subsequent pregnancies, genetic counselling is crucial.

Authors' contributions

All authors have materially participated in the study and manuscript preparation. All authors shared in proposal writing, data collection, manuscript writing, reviewing and drafting. All authors have approved the final article.

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Nil.

Conflicts of interest

The authors declared that there are no conflicts of interest.

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