MULTIDISCIPLINARY CARE FOR RARE CONDITIONS:DENTAL MANAGEMENT OF MUCOPOLYSACCHARIDOSIS TYPE-VI ;A CASE REPORT

by Jana Publication & Research

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Abstract

Mucopolysaccharidosis (MPS) is a set of metabolic illnesses characterized by abnormalities in lysosomal enzyme function that result in the buildup of glycosaminoglycans (mucopolysaccharides). Reporting a case of mucopolysaccharidosis in a 6-year-old child is the goal of this investigation with the oral manifestation and challenges that need to be addressed in the treatment. The involvement of several body organs necessitates consideration in dentistry for these patients. Treating these patients safely and effectively might be facilitated by the dentist's understanding of their oral-dental and systemic issues.

Keywords:Mucopolysaccharidosis,Arylsulfatase B,Chondroitin sulfate

Introduction

Mucopolysaccharidosis Type VI (MPS-VI), also known as Maroteaux-Lamy syndrome, is a genetic condition caused by the **absence of the enzyme arylsulfatase B (ARSB).**[1]

In 1963, Pierre Maroteaux and Maurice Lamy initially described MPS VI as a new dysostosis characterized by **elevated chondroitin sulfate** excretion in the urine.[2]

Worldwide, the birth prevalence ranges from 1 in 43,261 to 1 in 1,505,160 live births.[3] The incidence and prevalence of MPS are not adequately documented in India.[4]This case report has covered the consequences of dental management and related issues during treatment and observation.

Case Report

A 6-year-old female was diagnosed with Mucopolysaccharidosis Type VI at birth. She presented to the Department of Pedodontics with an asymptomatic grossly decayed upper front tooth following referral from the Department of Pediatric Neurology.

She is the first child with no reported family history of consanguinity and was a preterm baby with placenta previa born via cesarean section with a birth weight of 2.2 kg.

Genetic analysis confirmed a **homozygous variant c.260C>A** in **exon 1 of the ARSB gene**, confirming the diagnosis of Mucopolysaccharidosis type VI with autosomal recessive inheritance.

General examination findings

- Gross motor delay
- Chest deformity
- Mitral regurgitation

- Corneal clouding
- Umbilical hernia
- Mongolian spot
- Kyphosis
- Hepalosplenomegaly
- Astasia-abasia
- Stubby fingers
- Wrist deformity
- Dyosostosis multiplex
- Mucopolysaccharides in urine

The child had normal speech

Neurological defects :Craniovertebral junction(CVJ) anomalies,Dysplastic odontoid process with adjacent soft tissue thickening ,subluxation with ligamentous hypertrophy with anterior invagination of posterior arch of C1 severe spinal cord narrowing at CVJ with hyperintense signal at CVJ suggestive of compressive myelopathy.The child was planning for occipito-cervical decompression and fusion with wiring.

Airway: Difficult airway anticipated with history of pneumonia.

The child's behaviour during treatment was positive.

Extraoral facial findings

- Frontal bossing
- Flat nasal bridge
- Coarse face
- Facial profile was convex
- Hoarse voice
- Ala of nose was wide
- Philtrum was high and broad
- Antero-verted nostrils.

Intraoral findings

- Macroglossia
- Anterior open bite
- Tongue thrusting
- Diffficulty in mouth opening, and lateral movements due to cervical collar.
- Grossly decayed 51,52,61,62
- Dentinal caries in 54,64,74,84

Consent was obtained from the Pediatrician prior to all the procedures being done, and it clearly mentioned the instability of the neck and that no procedures should be done without

neck stabilization. Consent under general anesthesia was not given due to high risk.Oral hygiene instructions given. Restoration of 54,64,74,84 was done using GIC along with atraumatic extraction with proper neck stabilisation for extraction of 51,52,61,62was done.Monitoring all the primary teeth along with Casein Phosphopeptide-amorphous Calcium Phosphate (CPP-ACP) toothpaste was recommended.

There was difficulty in positioning the patient in the dental unit with the neck stabilized, which was challenging, but the patient was highly cooperative. Radiographs were not possible in this case due to difficulty in opening the mouth by the use of a collar and also due to gross motor delay.

DISCUSSION

Increased **accumulation of mucopolysaccharides**within intracellular **lysosomes** in various bodily tissues due to impaired metabolism of glycosaminoglycans (mucopolysaccharides)occur in this enzyme defect.[5,6,7].Types I, II, III, IV, VI, VII, and IX are the seven MPS types that have been identified. Eleven GAG-degrading enzyme deficiencies serve as the basis for additional classification[8,9].With the exception of MPS II, often known as "Hunter syndrome," which is X-linked, MPS show a recessive gene inheritance.[10]

Dermatan-sulphate and chondroitin-sulphate, which are naturally occurring substrates of arylsulfatase B activity, are undegraded glycosaminoglycans that accumulate pathologically as a result of the enzyme deficiency. A variety of clinical symptoms that increase with age are brought on by the build-up of partially degraded GAGs in tissues and organs as a result of this enzyme deficiency.

Extracellular and intracellular deposits gradually develop into a pathogenic situation that typically involves the **osteoarticular apparatus and most systems**.[11]

The respiratory system, spleen, central nervous system, blood, and bone marrow may all exhibit certain accumulations that over time cause harm to various physiological systems, tissues, and cells.[12,13]
The **IDVA (alpha-L-Idosiduronose) gene,** which contains the instructions needed to make the enzyme that hydrolyzes significant sugars known as glycosaminoglycans (GAGs), is mutated in the disorder.[14]Severe physical and neurological developmental issues, such as aberrant upper airways, restrictive lung illness, skeletal abnormalities, cervical spine deformities, and behavioral issues, may be present in the affected individuals.[15]Clinical signs

- Skeletal deformity
- Lumbar kyphosis or a hump
- Corneal opacity (with significant visual impairment and possibly loss of visual ability)
- Hepatosplenomegaly
- Heart valve problems

- Coarse facial characteristics
- Elevated urine mucopolysaccharide levels.[16]

Hunter and Dorfman claim that severe, recurring respiratory infections are caused by tonsillar and adenoid difficulties. After the patient is 2.1–3 years old, their hirsutism typically increases. Usually, cardiac or respiratory arrest is the cause of death.[17,18]

Varying degrees of orthodontic and paediatric issues are linked to the seven different forms of MPS.[19,20]

- Dental anomalies
- Malocclusions
- Tooth eruption deviations
- TMJ pathoses
- High caries index
- Periodontal diseases

According to the MPS literature, MPS types I and IV are associated with changes in the structure of the enamel and dentin, particularly in the DEJ.[21,22] Their scope and importance are unknown, though. It should be noted that in this instance, there were no alterations to the clinically healthy dentin and enamel structures of the existing teeth.

The symptoms that affected people may experience, according to Tyagi[23] and Scarpa et al.[24]

- Macrocephaly
- Hydrocephaly
- Heart valve abnormalities
- Short stature
- Mental retardation
- Dysostosis multiplex
- Cardiovascular anomalies
- Indigestion
- Skin thickening
- Large vocal cords

Additionally, some people may have narrow airways, which can result in repeated upper respiratory tract infections and sleep apnea. Typically, the skin curves inward, and when the knees are touched, the feet separate. These children have an odd stride and frequently **fall when walking**.Those who have been affected may have repeated ear infections and diminished hearing.[25]

Oral manifestations include

- Flattened TMJs
- Macroglossia
- Radiolucent jaw lesions
- Short and broad mandibles.

Macroglossia results from the **precipitation of GAG** in the tongue structure, which typically causes a large compressive force on the maxilla and an anterior open bite.[26] A broad tongue

has been linked to **anterior open bite** in these patients.[27] The palatal rugae are noticeable and have deep grooves in the midsagittal plane; the palate is often high-arched.[28]Patients with MPS require therapy in speciality centers due to their numerous oral manifestations, behavioral issues, and mental difficulties.[29]

GAG surrounding unerupted teeth, hyperplastic dental follicles from collagen precipitation, and dentigerous cysts with distinct borders may all contribute to delayed tooth eruption.[30] Since treatment is only successful in patients under the age of 2.5, a prompt diagnosis iscrucial.[31,32]

CONCLUSION

Considering the severity of MPS, children would require specialized medical and dental team. Because MPS has several impacts, children frequently need a multidisciplinary treatment approach from specialized pediatric teams that include Cardiologists, Neurologists, Psychiatrists, Orthopedic surgeons,Ophthalmologists,Pedodontists and Physiotherapists.

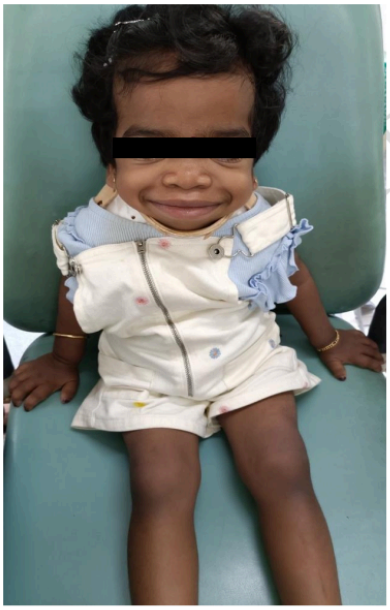

Fig 1:Neck stabilized with the help of cervical collar

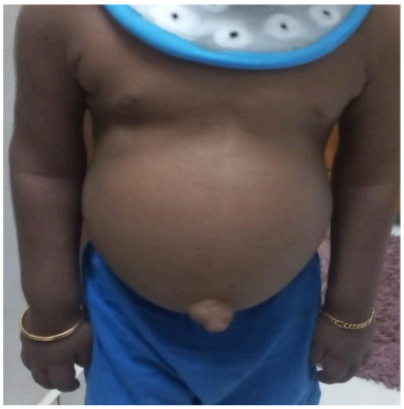

Fig 2:Umbilical hernia present

Fig 3:Corneal clouding

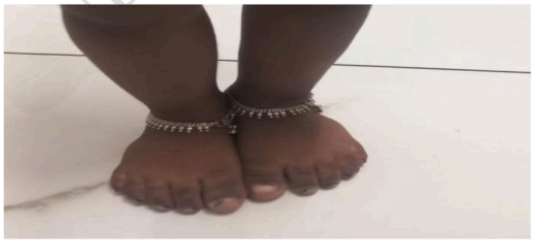

Fig4:Flat feet

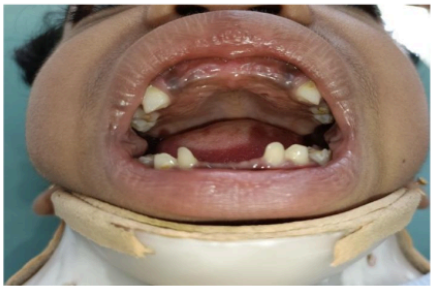

Fig 5 :Intraoral view of upper arch

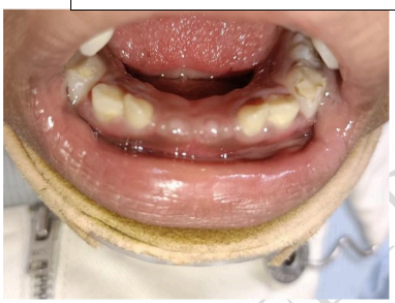

Fig 6 :Intraoral view of lower arch

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