

Maroteaux-Lamy Syndrome : A rare case of Mucopolysaccharidosis

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Abstract:

Maroteux-lamy syndrome is one of the genetic disorder involving disturbances in mucopolysaccharide metabolism, due to deficiency of aryl sulfatase-B which leads to accumulation of dermatan sulfate in tissues and their excretion in urine. The disease has several oral and dental manifestations, is first diagnosed on the basis of clinical findings. It is characterized by coarse facial features, normal intelligence, organomegaly, enlarged head, short neck, corneal clouding, enlarged tongue and prominent metachromatic inclusions in leukocytes. Death is usually a result of either respiratory tract infection or cardiac disease, which are caused by the deposition of mucopolysaccharides. An 6- year-old with Maroteux-Lamy syndrome is described in this article, with special emphasis on the oral manifestations.

Keywords: Maroteux-lamy syndrome, Genetic disorder

Introduction:

The mucopolysaccharidosis (MPSs) are a group of inherited disorders that results from the deficiency of one or more of the lysosomal enzymes required for glycosaminoglycans (GAG) catabolism^{1,6}. Mucopolysaccharidosis type VI or Maroteaux-Lamy syndrome, is a rare, autosomal recessively inherited glycosaminoglycans storage disease caused by deficiency of enzyme aryl sulfatase B (ASB)⁵.

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Fig 1 : Shows Characteristic facies of Maroteux-lamy Syndrome



Fig 4 : Shows Corneal Clouding, Hypertelorism, Flattening of nose



Fig 2 : Large Head, Short Neck, Open Mouth, Enlargement of Skull and Antero-Posterior dimension are typical of Maroteux-lamy Syndrome



Fig 5: Macroglossia, spacing of teeth, open bite



Fig 3: Shows Hypertrichosis



Fig 6: Short Strature

Aryl sulfatase B is required for the degradation of the glycosaminoglycans dermatan sulfate and chondroitin-4-sulfate. Aryl sulfate B deficiency causes intralysosomal accumulation and urinary excretion of large amounts of partially degraded dermatan sulfate¹⁵. Disease symptoms include growth retardation, coarse facial features, organomegaly, corneal clouding, prominent metachromatic inclusions in peripheral blood leukocytes¹⁶. In contrast to most other mucopolysaccharidosis, mental development in MPS type VI is normal⁷. Diagnosis and management are often challenging because of the considerable variability in symptom presentation and rate of progression.

Case report:

A 6 year old male patient with Maroteaux Lamy syndrome or MPS type VI disorder, was brought by his father to the Department of Oral medicine and Radiology, V S Dental college, Bangalore, with a chief complaint of decayed tooth in the upper anterior region. Past medical history of the patient revealed that he was suffering from severe obstructive sleep apnoea since 1 ½ years of age, mitral valve prolapse with mitral regurgitation and history of umbilical hernia operation done at 1 year of age. Family history of consanguinous marriage of parents and first sibling died at 1 ½ years of age with respiratory failure. Extraorally, he had frontal bossing, enlarged head, brachycephalic shape, euryprosopic face with convex profile, short neck, flattened nasal bridge, hypertrichosis, saddle nose, hypertelorism, clouding of cornea, and incompetent lips. Intraorally, macrognathia, macroglossia, with relative microdontia, spacing of teeth, and delayed eruption of teeth was observed. Dental caries was present in 54,64,65,74,75,84,85 and root stumps was noticed in 51,61,62. A panoramic radiograph was taken under difficult circumstances (the patient was uncooperative and anxious) and indicated several unerupted teeth. Laboratory diagnosis revealed abnormally high levels of Glycosaminoglycans concentration in urine which was 624.7173 mg GAG/g creatinine (normal range- 19.97-110.53). Enzyme assay revealed abnormally low levels of Aryl Sulfatase B that was 13.62 nmol/mg Proton/hr (normal value > 121 nmol/mg

Proton/hr) which is the pathognomic sign of MPS type VI.

Treatment plan:

General:

- Prompt treatment of all respiratory infection. [CPAP (Continuous positive air way pressure) use regularly at night with 13 cm of H₂O Pressure, with oxygen 3-4 litres/min]
- Prophylaxis against infective endocarditis if undergoing any dental or surgical procedures.
- Annual checkup with the cardiologist
- CT scan of the brain to rule out hydrocephalus.
- Extraction of 54, 51, 54, 55, 84 under antibiotic prophylaxis.
- Restoration of 74, 75.

Special concerns for dental treatment:

Anesthesia risk: MPS VI presents a significant anesthesia risk because of instability of the atlantoaxial joint. In particular, induction of anesthesia can be difficult because of problems maintaining the airway. According to the American Heart Association guidelines, patient should receive bacterial endocarditis prophylaxis before dental or surgical procedure.

Dental care:

- ▶ Preventive care – Fluoride application
- ▶ Maintenance of oral hygiene – use of battery operated tooth brush
- ▶ Regular dental check up

Discussion:

Maroteaux-Lamy Syndrome is also known as Mucopolysaccharidosis type VI. First described by Maroteaux and coworkers in 1965¹⁵. The estimated birth incidence of MPS VI ranges from 1 in 100,000 to 1 in 1,300,000 in various population⁴. Race is panethnic and it appears equally in males and females. It is a rare, autosomal recessively inherited glycosaminoglycan [GAGS] storage disease caused by deficiency of enzyme arylsulfatase B. ArylSulphatase B – required for degradation of glycosaminoglycans like dermatan sulfate and chondroitin-4-sulfate. ArylSulphatase B deficiency results in the accumulation of dermatan sulfate in tissues and their excretion in urine⁸. The deposition of mucopolysaccharides leads to a

progressive disorder involving multiple organs that often results in death in the second decade of life. However, in MPS VI which is characterized by somatic features but not by mental retardation, the patients are able to lead a relatively normal life when compared to other types of the disease. MPS VI patients appear healthy at birth and have accelerated growth in the first year, followed by deceleration and short stature later in childhood¹⁴. The disease can be diagnosed on the basis of clinical findings like: A enlarged head, short neck, corneal opacity, saddle nose, open mouth associated with macroglossia, widely spaced teeth with relative microdontia, unerupted dentition, dentigerous cyst like follicles^{1,16}, malocclusions, condylar defects, gingival hyperplasia, hepatomegaly and splenomegaly, umbilical and inguinal hernias are common. Growth may be normal for several years and may then stop, resulting in a final stature of 90-140 cm. A short trunk with lumbar lordosis is typically present. Restricted joint movement, including claw-hand deformities, appears in the first few years of life. Examination of the skin frequently reveals hirsutism.

Patients with MPS VI require ongoing medical care from numerous subspecialists. In addition, patients should receive routine pediatric care⁹, including immunizations. Obstructive airway disease can result from narrowing of the trachea, enlarged tongue, and redundant tissue. Tracheostomy has been performed in some patients. Tonsillectomy and adenoidectomy are also frequently performed to relieve obstruction. Many patients develop carpal tunnel syndrome, which may require nerve decompression. Enzyme replacement therapy with galsulfase (Naglazyme)^{2,10} has been shown to improve walking and stair-climbing capacity and to decrease urine glycosaminoglycan (GAG) levels in patients with MPS VI. Bone Marrow Transplantation has been attempted in a number of patients with MPS. Although BMT has been of particular interest in treating patients with MPS who are at risk for neurologic disease (MPS IH)^{6,14}, BMT has been limited by the associated mortality risk and the need for an appropriately matched donor.

Prognosis:

MPS VI is a progressive disorder with significant morbidity and early mortality, as with

many genetic inborn errors of metabolism, considerable variation exists among individual patients. Therefore, the prognosis of a particular patient must be determined after consideration of the presentation and complication.

Conclusion:

The dentist has a very limited role in treating patients with Maroteaux-Lamy Syndrome because dental care is primarily symptomatic with emphasis also on oral hygiene and caries prevention. Although Maroteaux-Lamy Syndrome is not encountered routinely in dental practice, but if properly managed, will maintain their oral and general health for the remainder of their lives.

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