Case Report

Multiple Dentigerous Cysts as a Rare Presentation of Maroteaux–Lamy Syndrome

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Abstract

Maroteaux–Lamy syndrome is one of the genetic disorders involving disturbances in mucopolysaccaride metabolism, due to deficiency of aryl sulfatase-B which leads to accumulation of dermatan sulfate in tissues and their excretion in urine. The diseases 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 is caused by the deposition of mucopolysaccharides. An 18-year-old with Maroteaux–Lamy syndrome is described in this article with multiple dentigerous cysts as the first presentation.

Keywords: Genetic disorder, Maroteaux–Lamy syndrome, multiple dentigerous cysts

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.[2,3] MPSs Type VI or Maroteaux–Lamy syndrome, is a rare, autosomal recessively inherited GAG storage disease caused by deficiency of enzyme aryl sulfatase-B (ARSB). ARSB is required for the degradation of the GAG dermatan sulfate and chondroitin-4-sulfate. Aryl sulfate B deficiency causes intralysosomal accumulation and urinary excretion of large amounts of partially degraded dermatan sulfate.[4] Disease symptoms include growth retardation, coarse facial features, organomegaly, corneal clouding, multiple odontogenic cysts, and prominent metachromatic inclusions in peripheral blood leukocytes.[5] In contrast to most other MPSs, mental development in MPS Type VI is normal.[6] Diagnosis and management are often challenging because of the considerable variability in symptom presentation and rate of progression.

Case Report

An 18-year-old male patient presented to the Department of Oral and Maxillofacial Surgery, Government Dental College and Hospital Srinagar, Jammu and Kashmir with a chief complaint of nasal obstruction on the right side for the past 3 months. Medical history of the patient revealed that he was suffering from severe obstructive sleep apnea from 1½ years of age, with a history of delayed milestones and cardiac abnormalities in childhood. Past dental history revealed gingival overgrowth that was operated at the age of 15 years and family history of consanguinous marriage of parents. Extraorally, he had frontal bossing, enlarged head, brachycephalic shape, euryproscopic face with convex profile, short neck, flattened nasal bridge, hypertrichosis, saddle nose, hypertelorism, and clouding of cornea. Intraorally, macroglossia, spacing of teeth and unerupted third molars. A panoramic radiograph and a computed tomography (CT) scan were taken and it indicated unerupted third molars with cyst formation in all the four quadrants [Figures 1-6]. Laboratory diagnosis revealed abnormal levels of GAG concentration in urine which was 224 mg GAG/g creatinine (normal range - 19.97–110.53). Enzyme assay revealed slightly low levels of ARSB that was 91.62 nmol/mg proton/h (normal value >121 nmol/mg proton/h) which is the pathognomonic sign of MPS Type VI.

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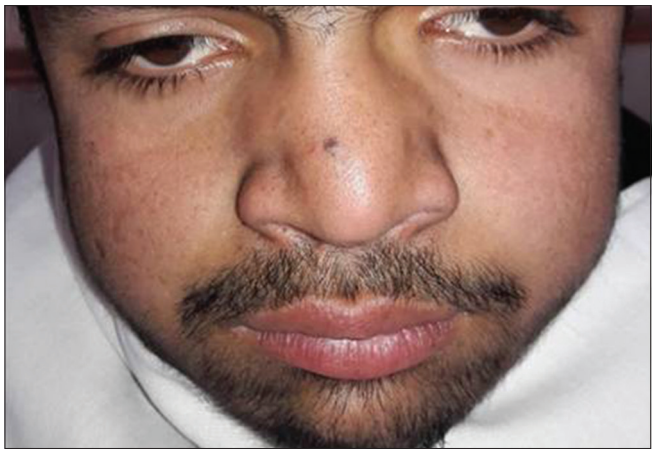

Figure 1: Coarse facial features

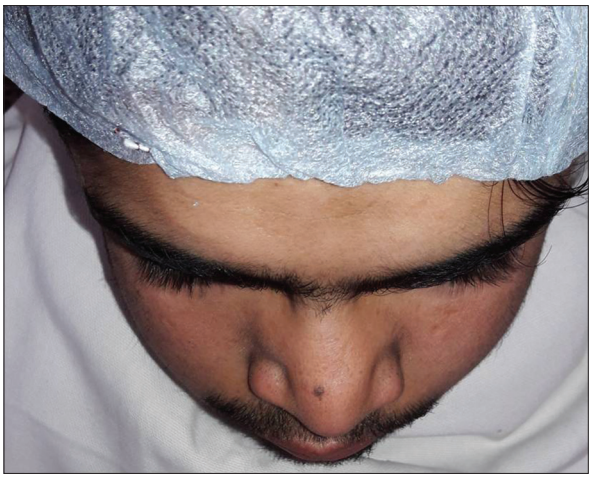

Figure 2: Broad nasal bridge

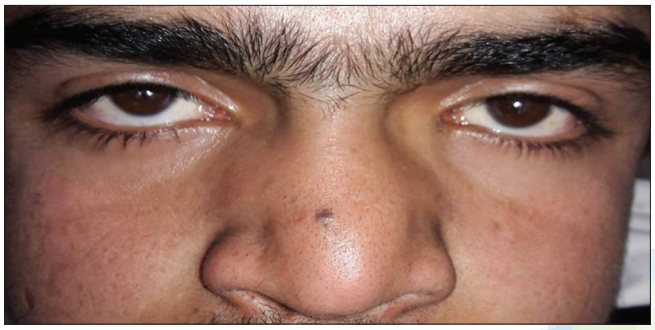

Figure 3: Hypertelorism and corneal clouding

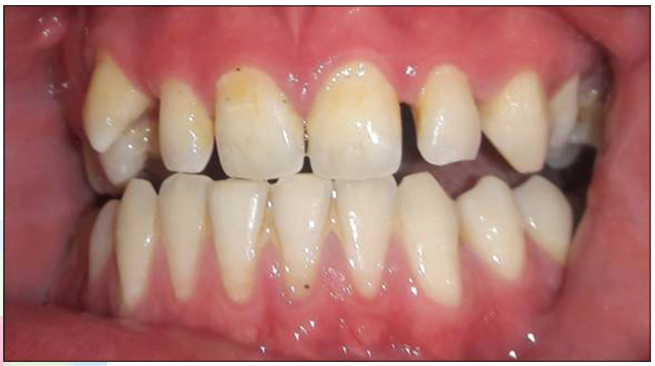

Figure 4: Spacing

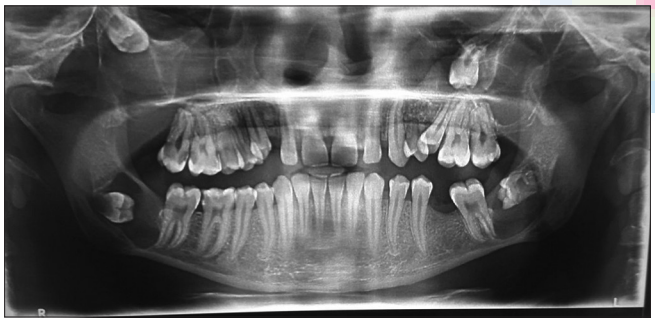

Figure 5: Multiple dentigerous cysts

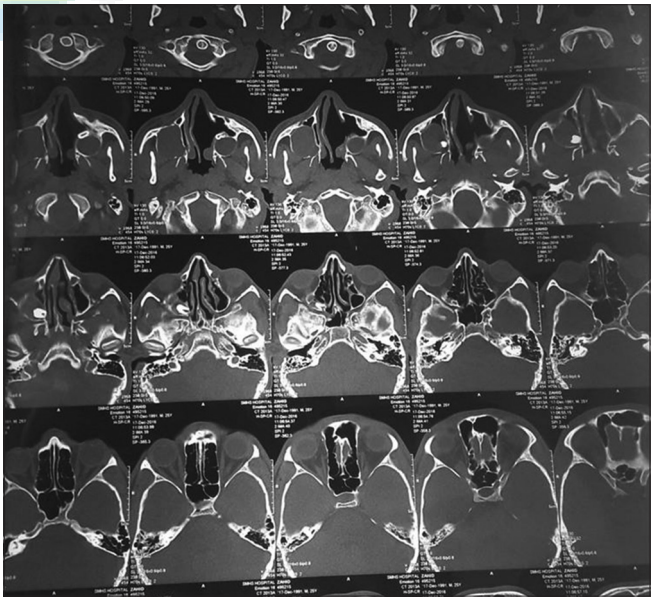

Figure 6: Computed tomography scan showing maxillary cysts

Treatment plan

A cardiology and general physician checkup was done for the patient which did not reveal any major abnormality at present except slight cardiac anomaly in the form of ST segment elevation and right bundle branch block. MPS VI presents a significant anesthesia risk because of instability of the atlanto axial 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. After thorough evaluation patient was cleared for surgical intervention. A CT scan was done and the extentions of the cysts were noted. Right maxillary cyst measured 3.5 cm × 4 cm that pushed the right maxillary antrum medially and left maxillary cyst measured 2.9 cm × 3 cm. Right mandibular cyst measured 2.4 cm × 2 cm and left mandibular cyst measured 2 cm × 2.3 cm. Aspiration of the cysts revealed dirty white fluid with protein content of 7.1 g/dl. A treatment plan was decided and the patient was operated

under local anesthesia. Marsupalization with opening into right antrum was done for right maxillary [Figure 7] cyst and marsupalization with secondary packing was done for left maxillary cyst [Figure 8]. Enucleation with primary closure was done for mandibular cysts [Figure 9] and the patient was treated with regular follow ups [Figure 10].

Discussion

The estimated birth prevalence is 1 in 320,000 live births in Europe. There is no current worldwide incidence rate and numbers may range according to country or specific ethnic populations studied. There are between 50 and 300 patients in the USA and approximately 1100 patients in the developed world with MPS VI.[7] Rapidly progressive forms usually present before 2 years of age with severe dysostosis multiplex and coarse facial features. Without proper treatment patients succumb before the second or third decade. A more slowly progressive (attenuated) form has been described with later onset, clinical symptoms in fewer systems, less pronounced dysostosis multiplex, and longer survival. The symptoms, onset and rate of progression of Maroteaux–Lamy syndrome vary greatly from one person to another. Review of literature shows different presentations of Maroteaux–Lamy syndrome [Table 1]. Some individuals may only have a few symptoms and others may have serious symptoms affecting several different organ systems simultaneously. Maroteaux–Lamy syndrome can potentially cause life-threatening

Table 1: Review of literature: Different presentations of maroteaux lamy syndrome

Author	Presentation	Journal
Gardner DG. 1971	Dental caries lower than average patients	Oral Surg oral med oral pathol oral radiol endod 1971;32:46-57
Donal l. Peterson 1975	Myelopathy	Arch neurol. 1975;32(2):127-129
Michael W. Roberts, 1984	Multiple dentigerous cysts	Oral surg. 58:169-175,1984
Ali Riza Alpoz 2006	Several unerupted teeth with pericoronal radiolucencies Resembling dentigerous cysts	Oral surg oral med oral pathol oral radiol endod 2006;101:632-7)
Deepak TA 2010	Macrognathia, Macroglossia, with relative microdontia, spacing Of teeth, and delayed eruption of teeth	J. Int oral health 2010
Agnieszka Jurecka 2012	Spinal cord compression	Pediatr neurosurg 2012;48:191-198

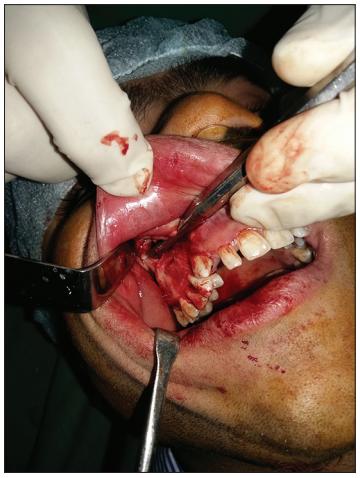

Figure 7: Marsupalization 1

Figure 8: Marsupalization 2

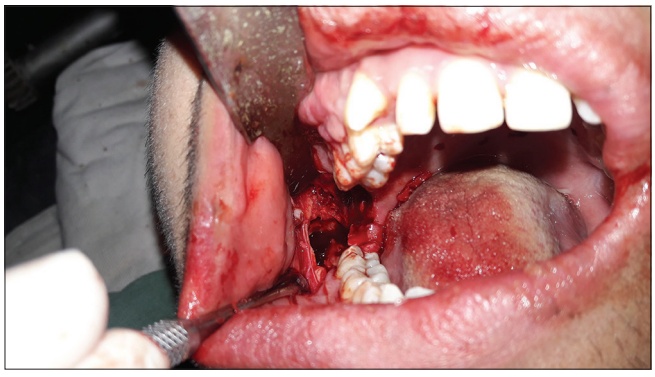

Figure 9: Enucleation of mandibular cyst

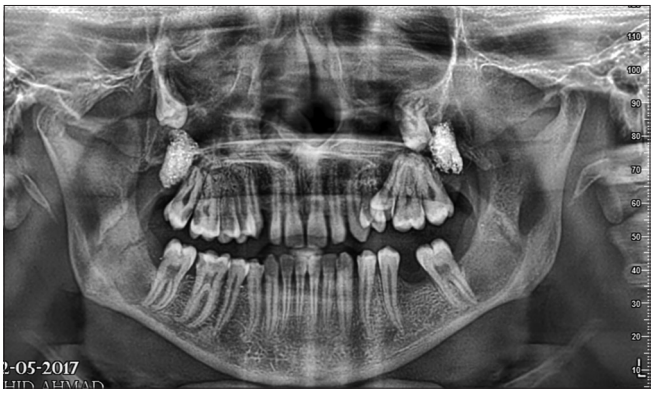

Figure 10: Postoperative orthopantomogram

Table 2: Reported cases of multiple dentigerous cysts till year 2000

Authors	Year	Sex	Race	Age (Years)	Location	Treatment
Sands and Tocchio	1998	F	N/A	3	Md. Central incisors and first molars	Enucleation
Banderas and others	1996	M	C	38	Md. Third molars	Enucleation
O'neil and others[6]	1989	M	Bl	5	Md. First molars	Enucleation
Eidinger[7]	1989	M	C	15	Md. First molars	Enucleation
Mcdonnell[8]	1988	M	N/A	15	Md. Second premolar and second molar teeth	Enucleation
Crinzi[9]	1982	F	Bl	15	Md. Third molars	Enucleation
Swerdloff and others[10]	1980	F	C	7	Md. First molars	Enucleation
Burton and others[11]	1980	F	Bl	57	Md. Third molars	Enucleation
Callaghan[12]	1973	M	C	38	Md. Third molars	Enucleation
Stanback[13]	1970	M	N/A	9	Md. First molars	Enucleation
Myers[1]	1943	F	N/A	19	Md. Third molars	Enucleation

N/A=Not Available; M=Male; C=Caucasian; F=Female; bl=Black; md.=Mandibular, credit- j can dent assoc 1999; 65:49

complications. The variable nature of Maroteaux–Lamy syndrome means that most affected individuals will not have all the symptoms potentially associated with the disorder. Individuals with this disorder can differ from one another dramatically. 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 1st year, followed by deceleration and short stature later in childhood.[8] The disease can be diagnosed on the basis of clinical findings like: an 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, 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. Multiple dentigerous cysts can be seen in a few conditions apart from this syndrome, although there have been various reports of multiple dentigerous cysts in literature [Table 2]. Maroteaux–Lamy syndrome is caused by mutations in the ARSB gene. Genes provide instructions for making proteins that play a critical role in many functions of the body. When a mutation of a gene occurs, the protein product may be faulty, inefficient, or absent. Depending on the functions of the particular protein, this can affect many organ systems of the body. Patients with MPS VI require on-going medical care from numerous subspecialists.

In addition, patients should receive routine pediatric care, 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)[9] 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 (BMT) 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),[3,8] BMT has been limited by the associated mortality risk and the need for an appropriately matched donor.

Conclusion

Although Maroteaux–Lamy syndrome is not encountered routinely in dental practice, unerupted dentition with dentigerous cyst formation can be rare presentation of the syndrome which if thoroughly investigated can help in diagnosing this rare syndrome with varied clinical features.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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