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Oral Manifestations of Morquio-Brailsford Syndrome with Trigeminal Neuralgia: A Rare Presentation of Mucopolysaccharidosis Type IV

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Abstract

Morquio syndrome (mucopolysaccharidosis type IVA) is an autosomal recessive disorder caused by the accumulation of mucopolysaccharides in lysosomes because of the deficiency of N-acetylgalactosamine- 6-sulphate sulphatase. Patients with MS(Morquio syndrome) can be clinically distinguished from other forms of MPS in that their intelligence is unimpaired.The present case deals with a known case of MS assosciated with trigeminal neuralgia having peculiar oral mucoasal manifestation which are rare in current literature.

Keywords: Mucopolysaccharidosis, Trigeminal neuralgia, Benign migratory glossitis,,Macroglossia.

Introduction

Mucopolysaccharidosis type IV also known as Morquio syndrome is an autosomal recessive lysosomal storage disorder characterized by intracellular accumulation of keratan sulfate and chondroitin-6-sulphate. The deposition of Glycosaminoglycans (mucopolysaccharides) in the lysosomes can results in damage to various organs in the body. The key clinical features include a normally intelligent child with short stature, skeletal dysplasia, dental anomalies and corneal clouding. Children will have no distinctive clinical finding at birth. In this article we aim to present Morquio syndrome having a rare manifestation of

trigeminal neuralgia with extensive bone involvement and peculiar dental features.

Case report

A fourteen-year-old boy was reported to the Department of Oral Medicine and Radiology, Government Dental college Kottayam, Kerala with an initial complaint of pain over multiple tooth and intermittent headache over the left side of the face. He has been diagnosed with mucopolysaccharidosis IV and trigeminal neuralgia at birth for which amitriptyline was taken for 5 years stopped due to financial problems. He was referred to the department to rule out dental foci. The parents did not belong to different ethnicity, yet they were not consanguineously related (first cousins, double first cousins, second cousins, double second cousin, or uncle niece relationship). The parents gave a history of recurrent upper respiratory tract infection since birth. The clinical examination revealed short stature, short neck, protuberant chest, scoliosis and unable stand or sit without the help from his parent. The craniofacial examination revealed a brachycephalic skull, prominent forehead with frontal bossing, hypertelorism, mid face deficiency, prominent chin area, a flat nasal bridge, flared alae nasae, broad mouth [Fig 1]. The absence of odontoid process made the head unstable and drooping [Fig 2]. On oral examination, oral hygiene was found to be satisfactory. Tongue tie and macroglossia were noted. The dorsum of tongue showed multiple areas of depapillation lined with white serpiginous line suggesting benign migratory glossitis. The anterior dentition was found to be healthy and surprisingly free of caries. Even though abundant plaque, calculus and stains were noted over the posterior tooth no caries were observed. Labially inclined maxillary and mandibular incisors, spaced arch, thin enamel, and pits of the enamel were noted. High arched palate, tongue thrusting and mouth breathing was evident. The premolars were missing and deciduous second molars found to be intact without any evidence of physiological mobility in relation to 54,55,64,65 region [Fig 3,4]. The radiographic examination was not undertaken due to the restless uncooperative jerky nature of the child. Fractured root stump of 45 was noted and removed from oral surgery department under local anesthesia. Patient was placed under periodic review for every 3 months to record dental status and to rule out orofacial pathologies.

Discussion

Mucopolysaccharidosis are rare heterogenous metabolic disorders due to the deficiency in the enzymatic degradation of complex carbohydrates: glycosaminoglycans. In 1929, Morquio, a pediatrician in Uruguay, and Brailsford, a Radiologist in England, independently described cases and later named as Morquio syndrome [1]. There are 7 types of mucopolysaccharidosis caused by 11 distinct single lysosomal enzyme deficiencies [2.3]. Patients with MPS IV can usually be clinically distinguished from patients with other forms of MPS as their intelligence is unimpaired as seen in this case. The exact incidence of this disease in India is not known. The estimated incidence of MS covers a wide range including 1 case per 200,000 births in British Columbia [4] and 1 case per 75,000 births in Northern Ireland.[5]

The child with Morquio syndrome will be normal at birth and later develop spondylo-epiphyso-metaphyseal dysplasia, skeletal deformities (platyspondyly, kyphosis, scoliosis, pectus carinatum, genu valgum, long bone deformities) ,joint hyperlaxity ,growth arrest at around 8 years of age and a definitive size of 1m to 1.50m, depending on the severity of the disease. Potential nervous complications are secondary to

skeletal deformations. Hypoplasia of the odontoid vertebra combined with joint hyperlaxity leads to an instability at the level of the first two cervical vertebra, with a risk of spinal cord compression. Extra-skeletal manifestations include respiratory problems, hepatomegaly, valvulopathies, hearing loss and corneal clouding. The lack of adequate ossification causes atlanto-axial loosening and odontoid dysplasia resulting from subluxation [6]. This situation shows up in patients as fatigue and progressive muscle weakness due to slow cervical cord compression. Sudden respiratory arrest with small traumas in severe forms may happen.Patients lose their gait function in the second or third decades if no enzyme replacement is done.

Orofacial manifestations of this syndrome is characterized by a flat nasal bridge, flared alae nasae, broad mouth, labially inclined maxillary and mandibular incisors, spaced arch, thin enamel, and pits of the enamel. The present case showed these characteristics. But there is no literature review regarding the oral mucosal changes as reported in this case[1,3,4].

There are two types of Morquio syndrome: type A caused by deficiency of N-acetyl-galactosamine-6-sulfatase (GALNS) and type B caused by deficiency of beta-galactosidase [4]. Both enzymes are required for degradation of keratan and chondroitin sulfate, but patients with Morquio type B usually lacks enamel hypoplasia. Therefore, it is likely that GALNS is more determinant for enamel hypoplasia in MPSIVA patients than the type with accumulated GAG. The enamel defects associated with the loss of GALNS activity in MPS IVA patients are likely to result from the pathological accumulation of keratan sulfate and chondroitin 6-sulfate in the lysosomes of ameloblasts in the secretory stage. In 1966, Langer and Carey found that eight of their patients with Morquio syndrome showed a thin enamel layer with sharp pointed cusps in both primary and permanent teeth [3]. Levin et al. described similar findings in 12 affected patients, and they concluded that these dental abnormalities were specific for the Morquio syndrome and were not seen in the other MPS [7].

The investigations for MPS IV includes Diagnostic methods available are blood and urine analysis to quantify the keratan sulfate level, direct enzyme assay in leukocytes or fibroblasts, and the wide range of radiographic views to demonstrate the skeletal abnormalities. Although the elevated urinary keratan sulfate is diagnostic of MPS, mucopolysaccharide excretion reduces with age and cases of Morquio with absence of excessive keratan sulfate in the urine have also been reported. Odontoid hypoplasia or dysplasia and spinal cord compression are the most consistent features in Morquio’s syndrome. A prenatal diagnosis for the disorder is possible by performing amniocentesis or chorionic villi sampling when there is a family history of Morquio’s syndrome. The diagnosis has to be confirmed with GALNS enzyme activity and molecular genetic testing of GALNS gene [8].

In 2014, the FDA approved a recombinant human GALNS enzyme replacement therapy for the treatment of MPS IVA [9]. The other treatment of MPS IV is symptomatic and supportive. Surgery to decompress and fuse the bones of the upper neck to the base of the skull can prevent destabilization of the cervical vertebrae and potential damage to the spinal cord. Management of affected individuals with MPS IV is best undertaken by multiple specialists, including: a physical therapist for physical rehabilitation, a psychiatrist for psychological support, educational

professionals for learning optimization, and home care professionals for affected individuals with medical equipment dependence. Surgeons may also play a crucial role in treating affected individuals. The placement of a bioprosthetic or prosthetic valve may be required for affected induvial with ventricular hypertrophy (overgrowth). Enlarged tonsils and adenoids may need to be removed in order to relieve upper-airway obstruction and sleep apnea [2,8]]. Additionally, ventilation tubes and hearing aids may be needed for individuals with hearing loss. Penetrating keratoplasty (corneal replacement) may be needed to treat corneal opacification (scarring or clouding of the cornea), which causes impaired vision.Since children with MPS IVA are of normal intelligence, they usually attend regular classes.Genetic counseling is recommended for affected individuals and their families.

Conclusion

In conclusion, MPS IVA is a rare type of mucopolysaccharidosis associated with highly specific dental abnormalities. As there is no cure for Morquio's syndrome, periodic monitoring and intervention are mandatory. Bone marrow transplant and enzyme replacement therapy (ERT) have been used with some success. This case illustrates the importance of systemic evaluation and inclusion of MPS type IVA in the differential diagnosis of enamel hypoplasia. As both the primary and permanent dentitions are affected, an early diagnosis is possible even in the clinically atypical cases. Early systemic evaluation and follow-up will help in improving the quality of life of the patients.

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Legends Figure

Figure 1: Extra oral photograph

Figure 2: Absence of odontoid process cause unstable head position.

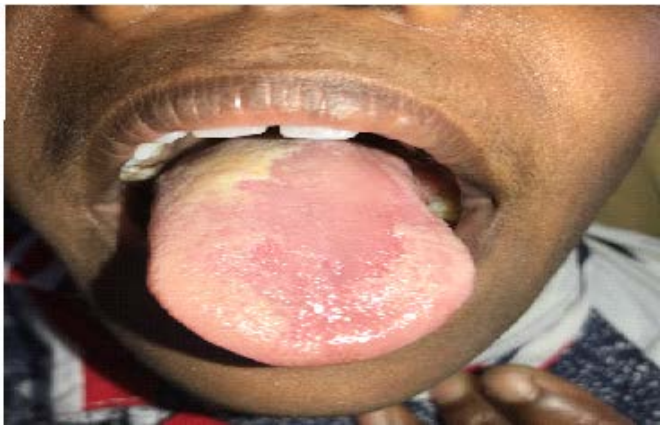

Figure 3: Macroglossia with benign migratory glossitis.