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Case Report (Open Access)

HURLERS SYNDROME-A RARE CASE REPORT

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ABSTRACT
Mucopolysaccharidosis type I (MPS I, Hurler syndrome) is a rare inherited disorder caused by the deficiency of a specific lysosomal hydrolase alpha L- iduronidase enzyme. It results in the accumulation of incompletely degraded glycosaminoglycans into various organs of body, which leads to impairment of organs and body functions. Mucopolysaccharidosis I is classified into severe MPS I (Hurlers syndrome) and attenuated MPS I (Hurler Scheie and Scheie syndrome). It is characterized by oral manifestations like widely spaced dentition, poorly formed enamel, short and broadening of mandible, multiple unerupted teeth, macroglossia and gingival hyperplasia. Early diagnosis, genetic counselling and regular follow up with recent modalities of treatment can decrease the mortality significantly. In this case report, a rare case of a 9-year-old child affected with Mucopolysaccharidosis I is reported.

Keywords: *Mucopolysaccharidosis I, Iduronidase, Macroglossia, Maxilla, Mandible*

INTRODUCTION
Mucopolysaccharidosis is an inborn heterogeneous group of rare metabolic disorders inherent as autosomal recessive traits (Ballıkaya *et al.,* 2018). Though clinical manifestations may vary with the type of mucopolysaccharidosis, the main features include: short stature, macrocephaly, mental retardation, coarse facies, umbilical/inguinal hernias, developmental delay, skeletal dysplasia with dysostosis multiplex, limited joint mobility or joint laxity, hearing loss, ocular involvement, neurodegeneration, dementia, cardiovascular diseases, respiratory distress and hepatosplenomegaly (Makhela *et al.,* 2015). Oral findings include thick lips, short/broad mandible, macroglossia, gingival hyperplasia, flattened alveolar ridges, high arched palate with pronounced rugae, delayed eruption of teeth, spaced dentition and anterior open bite (Makhela *et al.,* 2015, McGovern *et al.,* 2010). Radiographically, in infancy, bone trabeculation is coarse. In late infancy and early childhood, a pattern of skeletal changes called "dysostosis multiplex" emerges: the skull becomes large and deformed, the sphenoidal plane is depressed, and the sella is J-shaped (from arachnoid cysts). Joint stiffness and progressive lumbar Gibbus or kyphosis were also present (Ballıkaya *et al.,* 2018, Muenzer *et al.,* 2009). Herewith we report a rare case of a 9-year-old child affected with Mucopolysaccharidosis I .

CASE
A nine year-old female child reported to the department of oral medicine and radiology with the chief complaint of pain in right lower back tooth region. Past medical history revealed refractive anaemia, gradual hearing loss, history of multiple blood transfusions 5 years back (for the correction of anaemia). She was born to the non-consanguineous couple without any relevant family history. Clinically she had a short stature, coarse facial features like large head, prominent forehead, depressed and broad nasal bridge, wide upturned nostrils, bushy eyebrows, prominent supra orbital rim bilaterally, short neck, broad feets with the toes curved. Enlarged abdomen and stiffness of joints were present. Lateral view of the patient head revealed hypertelorism of fronto-occipital area.

On extra oral examination thick lips was present (Figure 1a). Intra oral examination revealed wide maxillary and mandibular arch (Figure 1b), multiple unerupted teeth, grossly decayed 84 and 85 and large broad tongue were present. The existing primary teeth were hypoplastic. Lateral and PA view of the skull radiographs were taken, showed features of dysostosis multiplex which included a large skull with thickened sclerotic calvarium, frontal, occipital hyperostosis, sellaturcica with J sign. Patient was referred to department of pediatrics for further investigations and management. Hand and wrist x rays were taken and it showed bullet shaped phalanges with proximal pointing of metacarpals. X-ray D-L spine lateral view was also taken which showed thick ribs and ovoid vertebral bodies were present (Figure 1d,e,f).

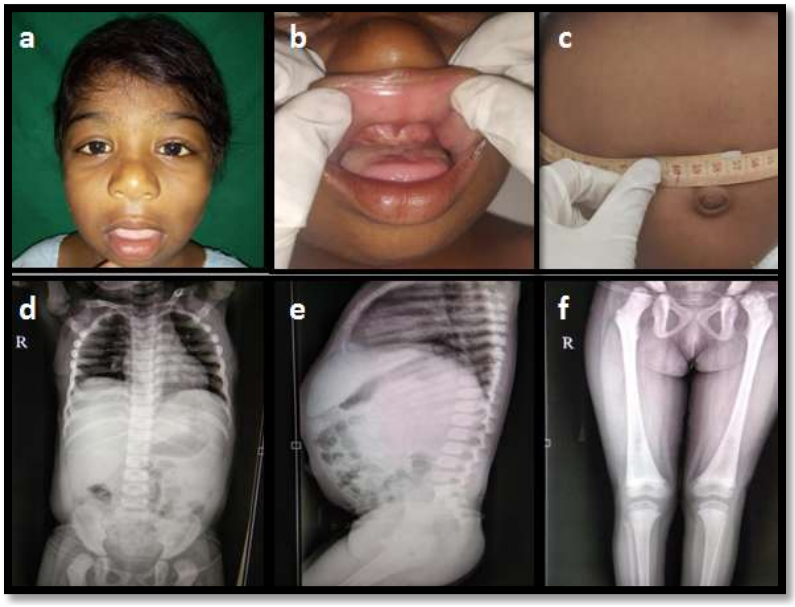

Figure 1

Based on the clinical and radiological findings, provisionally diagnosis of Mucopolysaccharidosis was given. Differential diagnosis of Hunter's syndrome and Morquio syndrome was considered. Patient was referred back to the department of oral medicine and radiology to address the chief complaint with multidisciplinary approach. Extraction of 84 was done under local anaestheia, oral hygiene instructions were given. Further, importance of proper oral hygiene and follow ups were stressed to the parents.

DISCUSSION

Hurler syndrome stands as the classic prototype of mucopolysaccharide disorder. It caused by deficiency of lysosomal enzyme alpha L- iduronidase has been mapped to the chromosome band 4p 16.3 leads to deposition of incompletely degraded mucopolysaccharide, which then interferes with the function of affected organs.[5] Severe MPS -1 patients die within first year of life due to cardio respiratory failure and progressive neurological disease.

Clinical findings include cardinal features, growth failure after infancy, marked mental retardation, characteristic craniofacial dysmorphism, physical habitus, dysostosis multiplex, corneal clouding, and deafness. These features were evident in the above case. Cardiovascular manifestations like cardiomyopathy, pulmonary arterial hypertension and heart failure are commonly noted which was not present in the discussed case. Frequent upper and lower respiratory tract infections are commonly encountered secondary to enlarged tonsils, adenoids (Neufeld and Muenzer, 2001; Sharma *et al.,* 2012).

Oral manifestations include hyperplastic alveolar ridges with spacing of teeth. Short mandibular rami with abnormal condyles, macroglossia, hyperplastic gingiva, high arched palate, spaced hypoplastic peg-shaped teeth with retarded eruption; and localized dentigerous cyst-like radiolucencies.[5] Radiological changes seen in the skull include macrocephaly, frontal bossing, calvarial thickening, and premature

closure of the sagittal and lambdoid sutures. The above case exhibited the classic radiological findings. Hydrocephalus is common. The sella turcica is enlarged and J shaped. The ribs are overly wide, with tapered ends, producing a paddle or spatulated appearance.

Often, the facial bones are the mandibular angle is widened. The gonions stand out very prominently, there is a wide intergonial distance, and the distance around the arch from the posterior border of one ramus to the posterior border of the other is greater than normal, counting for the spacing of the teeth. Often, the metacarpals and phalanges are short and wide, producing a trident hand which was observed in the above discussed case. Osteoporosis is a frequent finding (Srinivas *et al.,* 2015; Tatapudi *et al.,* 2011).

Hematopoietic stem cell transplantation is the treatment of choice for a child younger than 2 years of age suffering from Hurler syndrome with no or minimal central nervous system disease. Enzyme replacement therapy (alpha-Liduronidase, Aldurazyme) is now available to treat the intermediate and milder phenotypes (Hurler–Scheie and Scheie) as well as recently more severely affected patients .As a result of these improved therapies, patients with Hurler syndrome may increasingly need dental treatment. It is therefore useful for the dental practitioner to be aware of the clinical and radiographic features of Hurler's syndrome (Thakur *et al.,* 2015).

CONCLUSION

Mucopolysaccharidosis type I (MPS I, Hurler syndrome) is a rare inherited disorder with accumulation of incompletely degraded glycosaminoglycans into various organs of body, which leads to impairment of organs and body functions. This case report presents early diagnosis, genetic counselling and regular follow up with recent modalities of treatment of a patient using a multidisciplinary approach which can decrease the mortality significantly.

REFERENCES

Ballıkaya E, Eymirli PS, Yıldız Y, Avcu N, Sivri HS, Uzamış-Tekçiçek M (2018). Oral health status in patients with mucopolysaccharidoses. *Turkish Journal of Pediatrics*, **60**(4) 400-406. *doi: 10.24953/turkjped.2018.04.007. PMID: 30859764.*

Makhela J, Kaushal K, Choraria S, Ranka M, Patel S B (2015). *Report of three cases*. IAIM, **2**(6) 210-213.

McGovern E, Owens L, Nunn J, Bolas A, Meara AO, Fleming P (2010). Oral features and dental health in Hurler Syndrome following hematopoietic stem cell transplantation. *International Journal of Paediatric Dentistry,* **20**(5) 322-9. doi: 10.1111/j.1365-263X.2010.01055.x. Epub 2010 Jun 2. PMID: 20545789

Muenzer J, Wraith JE, Clarke LA (2009). International Consensus Panel on Management and Treatment of Mucopolysaccharidosis I. Mucopolysaccharidosis I: management and treatment guidelines. *Pediatrics,* **123**(1) 19-29. *doi: 10.1542/peds.2008-0416. PMID: 19117856.*

Neufeld EF,Muenzer J 2001). The mucopolysaccharidoses. In: Scriver CR, Beaudet AL, Sly WS, Valle D, editors. The metabolic & molecular bases of inherited disease, vol. III. 8th ed. New York: McGraw-Hill;.

Sharma S, Sabharwal JR, Datta P, Sood S (2012). Clinical manifestation of Hurler syndrome in a 7 year old child. *Contemporary Clinical Dentistry,* **3**(1) 86-89. *doi:10.4103/0976-237X.94554)*

Srinivas V, Ravikumar N, Srinivas V R, Vikramvadhan R (2015). Hurler's Syndrome: A Rare Case Report. *Scholars Journal of Medical Case Reports,* **3**. 568-570.

Tatapudi R, Gunashekhar M, Raju PS (2011). Mucopolysaccharidosis type I Hurler-Scheie syndrome: A rare case report. *Contemporary Clinical Dentistry,* **2**(1) 66-68. *doi:10.4103/0976-237X.79287*

Thakur AR, Naikmasur VG, Sattur A (2015). Hurler syndrome: orofacial, dental, and skeletal findings of a case. *Skeletal Radiology,* **44**(4) 579-86. *doi: 10.1007/s00256-014-1982-7. Epub 2014 Aug 20. PMID: 25134498.*