Oral Manifestations of Hurler Syndrome

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University Hospitals Rainbow Babies & Children's
The Science of Health. *The Art of Compassion.*

Introduction

Hurler syndrome (HS), is a rare genetic lysosomal storage disorder. Lysosomes contain enzymes that break down molecules such as fats and sugars in the body. HS is caused by a variation of the gene for the protein that helps lysosomes break down complex sugars. Consequently, undigested sugar molecules accumulate and cause systemic damage across the brain, heart, and other vital organs. HS is inherited in an autosomal recessive pattern and is considered a pediatric syndrome because the life expectancy of these patients only extends into the late teens and early twenties. The craniofacial manifestations of HS are significant, necessitating a multidisciplinary approach with significant emphasis on treatment of dental alveolar anomalies.

Case Description

A thirteen year old female presented to University Hospitals Rainbow Babies & Children's Hospital dental clinic in January 2024 with a chief complaint of dental pain. Patient reports pain upon palpation near #9. Mother states the child's nutritional intake has decreased over the last couple weeks. Past medical history is significant for Hurler syndrome, developmental and speech delay, autism, ADHD, hypothyroidism, mitral valve regurgitation, hip dysplasia, scoliosis, patent foramen ovale and sensorineural hearing loss. Patient (pt) is also status post stem cell transplant for enzymes**.** Pt is currently taking Levothyroxine and Vyvanse for medications. Pt is allergic to Omnicef, fruit juice and milk.
Mother reported that pt has been overdue for dental care since 2018. Pt started experiencing dental discomfort since 2020. Pt completed her first comprehensive oral rehabilitation (OR) under general anesthesia (GA) in 2021, however, has not had routine recalls with the pediatric dentist since. After seeing the pt in clinic, we planned for OR under GA to complete restorative work for March 2024.

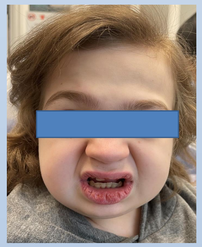
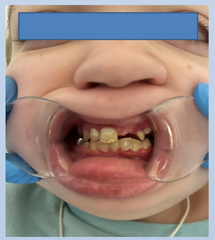


Fig. 1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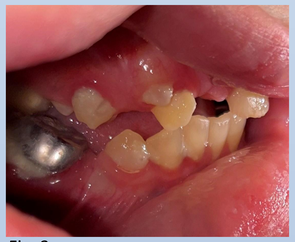
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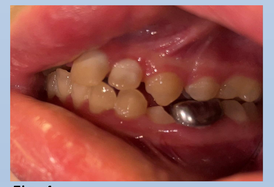
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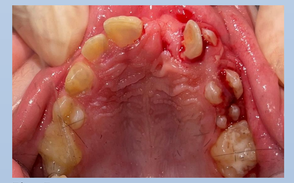
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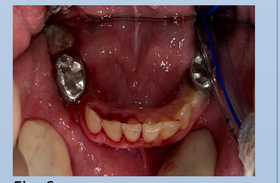
Fig. 6

Findings

- Extra oral exam: No facial asymmetry, widely spaced eyes, large forehead, flat nasal bridge, enlarged lips, no swelling or lymphadenopathy present, and midlines on center (Fig. 1-2)
- Intraoral exam: Generalized plaque, calculus and gingival inflammation. Macroglossia. Patient in late mixed dentition with retained K,T; #4,9 are impacted. #10 is distally rotated..Bilateral posterior cross bite. Multiple microdonts noted. Enamel defects noted on #7,8 (Fig. 3-6)
- Radiographic exam: Panoramic radiograph and a CBCT was acquired. Periapical radiolucency was noted around #22 and 26. Microdonts #2,18 and 31 noted (Fig. 8, 11-12) . #11 is superimposed on #10. Bilaterally missing premolars #21 & #29. Third-molars #1,16 were noted to be developing (Fig.7)

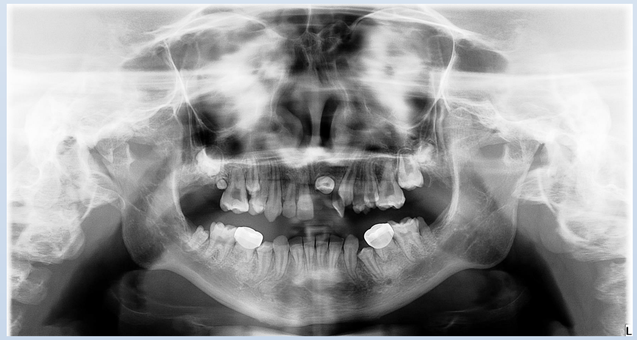
Fig. 7

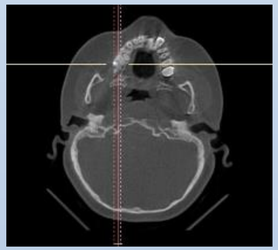
Fig. 8 Microdont #2

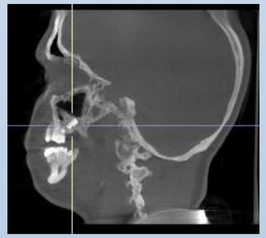
Fig. 9 Impacted #4

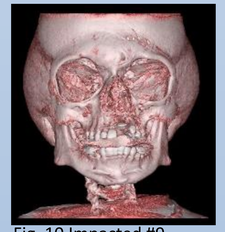
Fig. 10 Impacted #9

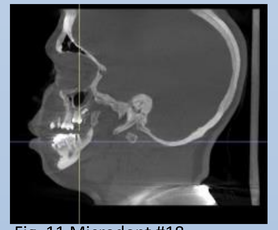
Fig. 11 Microdont #18

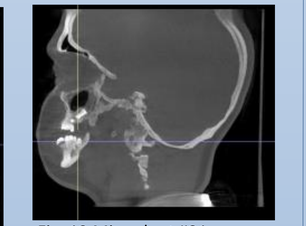
Fig. 12 Microdont #31

Dental and Craniofacial Manifestations

Following dental and craniofacial anomalies were noted in our patient which is typical in a patient with Hurler syndrome:

- Microdontia
- Frontal bossing
- Enamel defects
- Tooth impaction
- Delayed eruption
- Hyperplastic gingiva
- Malocclusion and cross bite
- High arched palate and macroglossia

Treatment

The department of Pediatric dentistry, the Craniofacial and Oral surgery (OS) team at Case Western Reserve University implemented a treatment plan including:

- Pediatric dentistry planned comprehensive OR under GA to complete restorative work
- Craniofacial consult was established for patient to determine the outcome of #9
- Upon recommendation of craniofacial team, extractions of #2,4,9,18 and 31 were planned by OS. Periapical radiolucencies noted around #22 and 26 were determined to be artifacts by OS (Fig. 7)

Patient will be followed by pediatric dentistry for routine care

Discussion

A CBCT was needed to obtain accurate diagnostics of the location, shape and root development of impacted #4, #9 and microdonts #2, 18, 31 (Fig. 8-12). #4 was impacted towards buccal, #9 was impacted apically (Fig. 9-10). Many considerations were taken into account with this case:

- Craniofacial Orthodontics: Upon consultation, patient was offered to expose and ligate #9, place in active orthodontic. During the consultation appointment the pt was not cooperative. The treatment plan changed to extract #9 since there was pain associated and the mother agreed.
- Oral Surgery: Full thickness flap was laid at site #2,4,9,18,31. Extractions were completed on 2,4,9,18,31. Chromic gut 3.0 sutures were placed on sites #2,4,9,18,31
- Pediatric Dentistry: #7,8 and 12 were restored with glass ionomer. Previously placed stainless steel crown on #T had wear facets, crown was replaced. Sealants were placed on 3,14,19,30. Pt was prescribed chlorohexidine rinse twice daily for 4 weeks for gingival inflammation

References

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3. McGovern E, Owens L, Nunn J, Bolas A, Meara AO, Fleming P. Oral features and dental health in Hurler Syndrome following hematopoietic stem cell transplantation. Int J Paediatr Dent. 2010 Sep 1;20(5):322-9. doi: 10.1111/j.1365-263X.2010.01055.x. Epub 2010 Jun 2. PMID: 20545789.