Severe Mandibular Crowding in a Child with Infantile Pompe Disease: A Case Report.

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Background

Pompe disease (PD) is a rare, multisystemic, hereditary disease which is caused by the mutation in GAA gene. Children with Infantile Pompe disease usually present with hypertrophic cardiomyopathy, extreme weakness, decreased muscle tone, and feeding/swallowing difficulties. Before recent advances in the Enzyme replacement therapy (ERT), death almost invariably occurred in the first year of life.
To date, only a few reports are available on orofacial manifestations in children with PD and no studies reported the involvement of pediatric dentistry in the long-term management of children affected by infantile Pompe disease.

Case Presentation

This case is a 4 year 2-month-old male with Pompe disease present with the chief concern of “Crowded lower front teeth that are pointing straight back, and he is grinding his teeth because of pain.” Parents noticed that patient’s lower teeth were increasingly crowded and is now in 3 different rows in the past 3 years. Parent provided previous dental radiograph to support the finding of gradual change of the occlusion (Fig 1). Father also mentioned that patient has a large open bite, which they believed leads to pain, excessive grinding and challenges in speech ability.

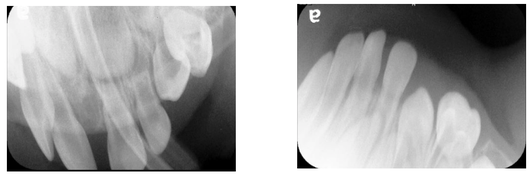

Fig.1 Parents provided occlusal films from June 2018 (3 years ago from this appointment).

Medical history: Pompe disease, dysplastic tricuspid valve with severe regurgitation and pulmonary hypertension, history of left ventricle hypertrophy, respiratory insufficiency, GERD, central sleep apnea, developmental delay, hypotonia and hearing loss
Current therapy: Enzyme replacement therapy (ERT), daily respiratory therapy with BiPAP (for 2.5 years; 12 hr/night)
Medications: colistimethate, tobramycin, alglucosidase alfa, famotidine, melatonin, acetylcysteine, diphenhydramine, levocetirizine and vitamin D3
Diet:G-tube fed, occasionally tasting on a 3 times/week basis.
Brushing habits: performed by parents twice daily

Clinical Findings

Dentition: full primary dentition
Occlusion:
- 15 mm anterior open bite, only occlude on the second primary molars.
- Maxillary dentition was well-aligned with adequate spacing while mandibular dentition was severely crowded (Fig 2).
- Tooth #Q and Tooth #N were aligned lingually to Tooth #O and Tooth #P and bilateral posterior alveolar ridges is inclined lingually.
- Tooth #M and Tooth #S were in infra-occlusion, suggesting possible ankylosis.

Oral hygiene and periodontal condition: No obvious gingival inflammation.
Caries: No clinical caries present.
Restorations:No restorations present.
Other findings: no significant attrition on primary molars.

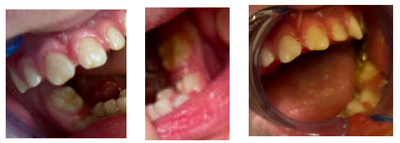

Fig.2 Clinical photo showing crowded mandibular dentition.

Treatment approach

The case is managed in a multidisciplinary approach with a team including otolaryngology, pediatric pulmonology, pediatric genetics, speech therapy, pediatric dentistry, oral surgery and orthodontics.

Concerns	Management	
Attrition	Pediatric Dentistry	No attrition sign observed in the clinical exam
Pain		Did not observe obvious wear on the primary molars related to grinding, pain was suspected to be related to TMJ discomfort resulting from the occlusion.
Speech	Speech therapy	Prefer more anterior dental contact.
Occlusion	Oral Surgery Orthodontic	• The occlusion appeared to have been initiated and caused by pressure from the BiPAP mask. • Suggest changing the direction of force on the BiPAP to prevent more incisor tipping • Upper incisor could be moved lingually with aligners • Correction of the open bite with orthodontic appliances would be too much risk for aspiration and anchorage is limited at this time.
	Pulmonology	Could try larger mask or fullface mask to alternate pressure on the face

Discussion

Common oraofacial features: hypotonia of facial and tongue muscles, lip incompetence, narrow palate with lateral and posterior crossbite, macroglossia, hypoplasia of maxillary-malar area and mandibular prognathism.
Common oral signs: gum swelling, gingival overgrowth, delayed tooth eruption, taurodontism and fusion of teeth.

The hypoplasia of maxillary-malar area
- Suspected to be associated with the absence of physiological nasal breathing and the negative effects of long-term use of nasal non-invasive ventilation (NIV) on the facial growth.
- Treatment in past literatures include orthodontic extraoral forces (e.g. chin cup, facial mask) /fixed orthodontic treatment combining the substitution of the NIV mask with a nasal pillow and orofacial myofunctional therapy (oral exercises to improve facial muscle weakness, swallowing and chewing).

Role of Dental professions: Early treatment of orofacial functions is important to limit the onset of severe deficits in swallowing, speech, chewing and breathing.
Other consideration includes the use of bisphosphonates as a treatment of reduced bone density in patient with Pompe disease

Conclusion

Treating children with Special Health Care Needs is often very challenging and seldom has straight-forward resolutions. In this case while we attempt to relieve the malocclusion, ventilator options that are available for the patient also has to be considered. Multidisciplinary medical and dental interaction is essential in the assessment and treatment of these children with complex needs. By presenting this case, we wish to add another case to the present limited literature of orofacial manifestation associated with Pompe disease.

References

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