Dental Root Resorption in Cri-du-chat Syndrome

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Introduction to Cri du chat Syndrome

In the care of adults with special healthcare needs, there is a perpetual expectation for the unexpected. Frequently, cases will present with the most uncommon comorbidities and often in very atypical fashion. In this, we present a case of unique chromosomal abnormality and it's associated dental findings.

Cri-du-chat Syndrome is a rare genetic disorder (1 in 20,000-50,000) characterized by ID, microcephaly, weakened muscle tone and a characteristic high pitched cry in infancy that gives it, it's rather fanciful name. First identified in 1963, though rare in the general populous, it is one of the most common chromosomal abnormalities involving a deletion of the 'p' arm of chromosome 5. It is believed that the loss of multiple genes on this short arm is what contributes to the physiologic manifestations of cri-du-chat syndrome. While most of these remain unidentified; some – such as CTNND2 (believed to associate with more severe ID) – have been recognized.

It is not impossible to imagine that the dental presentation of these patients would have to suggest that one of these "yet-to-be-identified" deleted genes would have to have some impact on both the development and maintenance of teeth and bones. From the viewpoint of the dentist, a patient with cri-du-chat syndrome might be identified by distinct retrognathia, radiographic dental opacities, dental agenesis, and dental transposition among many other low-prevalence anomalies.

Of these anomalies, dental root resorption has been previously recognized, but not very well understood. In this poster, we present a case of a patient with cri-du-chat syndrome presenting multiple, atypical root resorptions. We will review the case's presentation, findings and management to the present moment. We will, further, reflect on the correlation of specific genetic disorders with unique, characteristic dental findings, and suggest possible next steps in the study and further understanding of the manifestations of these unique dental findings in general and in cri-du-chat patients in particular.

Other names
- Chromosome 5p deletion syndrome
- 5p– syndrome
- Lejeune's syndrome

Facial features of a person with Cri du chat syndrome at the age of 8 months (A), 2 years (B), 4 years (C) and 9 years (D)

Figure 1: Classic craniofacial features of Cri-du-chat Syndrome. Both images cited from Ritwik, P., & Patterson, K. K. (2018, December 21). *Diagnosis of tooth agenesis in childhood and risk for neoplasms in adulthood*. Ochsner Journal. https://www.ochsnerjournal.org/content/18/4/345

Unique Dental Findings in Cri-du-chat and other syndromes

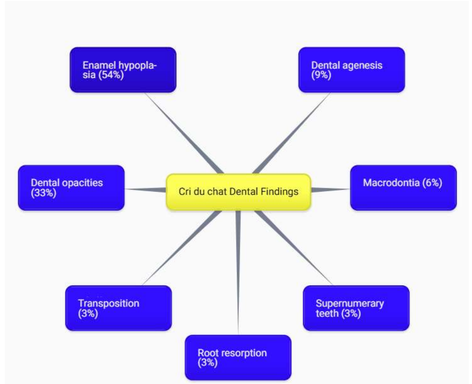

Figure 2: Atypical dental findings in Cri-du-chat patients

Other Syndromes and "Classic" Dental Findings

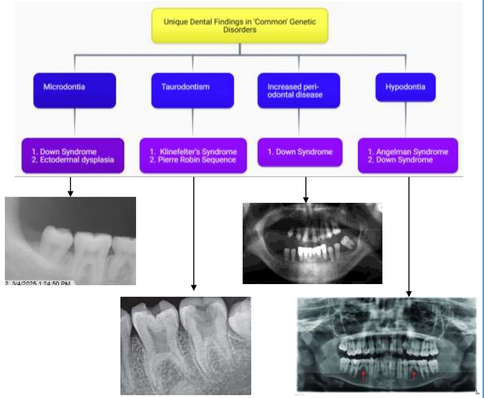

Figure 3: Other unique dental findings and syndromes that they tend to be associated with. (A) Pt from Pitt CPSN (B) Photo from Dentagama website (What is Taurodontism page) (C) Amano, Atsuo & Murakami, Jumpei & Akiyama, Shigehisa & Morisaki, Ichijiro. (2008). Etiologic factors of early-onset periodontal disease in Down syndrome. Japanese Dental Science Review. 44. 118-127. 10.1016/j.jdsr.2008.07.001. (D) Ritwik, P., & Patterson, K. K. (2018, December 21). *Diagnosis of tooth agenesis in childhood and risk for neoplasms in adulthood*. Ochsner Journal. https://www.ochsnerjournal.org/content/18/4/345

Case Presentation

Patient presents as a 23 year old female. Medical history significant for Cri du chat syndrome (limited verbal skills), autism spectrum disorder, Barrett's esophagitis, vitamin B-12 deficiency, self-injurious behavior, constipation and PTSD. Patient takes Nexium, probiotic, fiber gummies and magic swizzle (this is provided for frequent intraoral ulcerations). Patient is allergic to Doxylamine, Guanifenesin, Lactose, Tomato, Dextromethorphan. Patient has no significant surgical history except for dental care and adenoidectomy, and her social history is negative for smoking, drinking and use of any recreational drugs. She lives at home with her mother, who provides consent; and relies on others for some of her ADLs.

Treatment Timeline

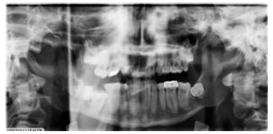

Figure 4: Presenting PAN from 2018

Jan 2025 - Pt presents as "walk-in" reporting pain on mand R

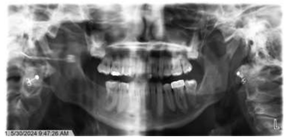

Figure 5: PAN from 2023 post ext of 3rds

Jan 2025 - Pt seen for GA case, and #30 extracted

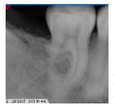

Figure 6: PA of #30 on date of first surgery

Feb 2025 - Pt presents for non-sedated follow up and exam

Mar 2025 - Pt reporting pain on mandibular R

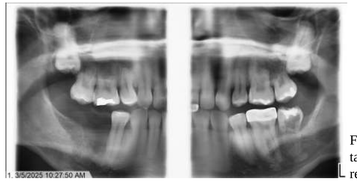

Figure 7: PAN BW taken when pt reported L sided pain

Mar 2025 - Pt sedated for extraction of #18 (pathologic fracture and caries). The patient is now considered dentally stable; her continued care will include monitoring and management of acute symptoms as needed. Patient is next due for recall in late June 2025.

Clinical Case in Photographs

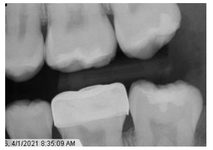

Figure 8: BW of Posterior L from April 2021

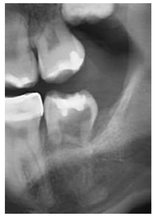

Figure 9: Caries and coronal fracture of #18 – March 2025

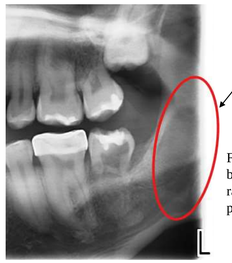

Figure 10: Concern for unilateral bone radiolucency – Follow up with radiology and CBCT confirmed, no pathology

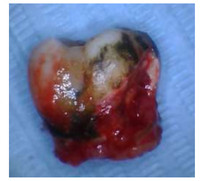

Figure 11: Immediate post-extraction #18

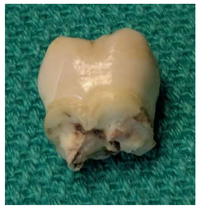

Figure 12: 2 months post-extraction – visualization of degree of resorption

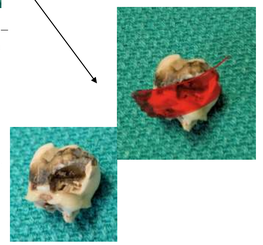

Figure 13: 2 month post-op images of #18 showing degree of caries. Note – red shading in figure shows external oblique ridge obscuring caries

Summary

As with any patient, the primary concern focuses on the outcome. The greatest ease a provider can feel is in the management of a well-defined, well-understood and predictably solved medical problem.

In the case of the presented 23 year-old female. While they are, at the time of this presentation, dentally stable, her case is reflective of an ill-defined problem with an as-yet-unclear solution. Figure 14 below reflects the additional, atypical and generalized recessions that we are starting to note with this patient.

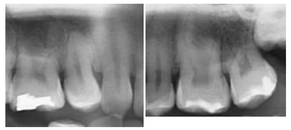

Figure 14: Atypical root resorption in the case study patient

It is the hope of these presenters that this case spark a degree of interest in these atypical findings associated with rare congenital disorders. We would highly support any collaborative literature review to search for a better understanding of the underlying genetic and biological mechanisms that might facilitate this finding. Any such effort would almost certainly constitute an admirable first step in the better understanding of this condition in the future. We might be able to offer a well-defined, well-understood solution for this 23 year-old female.

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