Dental Manifestations of Trisomy 8 Mosaicism: A Case Report

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INTRODUCTION

Trisomy 8 mosaicism, also known as Warkany Syndrome 2, is an autosomal genetic disorder in which some cells of the body contain a third copy of chromosome 8. It occurs in approximately 1:25,000-50,000 births with a strong male predilection. Trisomy 8 mosaicism can manifest in a variety of structural and systemic abnormalities ranging from mild to severe, including distinct facial features, deep palmar and plantar creases, cardiac and renal anomalies, joint and vertebral abnormalities, and intellectual disability. The few reports detailing oral manifestations of Trisomy 8 mosaicism have highlighted findings including cleft palate, high arched palate, and micrognathia, though there are limited descriptions of dental anomalies.

This case report discusses a 10 year old male with Trisomy 8 mosaicism who presented to University Hospitals Rainbow Babies and Children's Hospital for comprehensive oral rehabilitation under general anesthesia. The patient displayed many characteristic features of Trisomy 8 mosaicism including cardiac, renal, and vertebral malformations. Notably, his dental exam revealed unerupted permanent first molars. This report describes dental anomalies associated with a case of Trisomy 8 mosaicism as well as the dental treatment rendered to improve patient outcomes.

CASE REPORT

A 10 year old male presented to Prentiss operating room at University Hospitals Rainbow Babies and Children's Hospital in May 2024 for comprehensive oral rehabilitation under general anesthesia. His medical history includes Trisomy 8 mosaicism, prematurity, autism spectrum disorder, mild intermittent asthma, resolved atrial septal defect, Still's murmur, occult spinal dysraphism sequence, congenital hydrocele, spina bifida, congenital hypothyroidism, developmental delay, Duane's syndrome, eustachian tube dysfunction, intestinal malrotation, oropharyngeal dysphagia, feeding by g-tube, gastroesophageal reflux disease, hydronephrosis, hypotonic cerebral palsy, seizure disorder, undescended testis, and torticollis.

The patient was previously seen under general anesthesia in May 2023 during which no caries were identified and a cleaning was performed along with extractions of two mobile primary teeth.

At his 2024 visit, the patient's soft tissue exam revealed generalized gingival hyperplasia and a high arched palate. It was noted that #20 was congenitally missing. The dental exam revealed permanent incisors with primary canines and molars. #14 was partially erupted with a small distal operculum. #3 presented with soft tissue impaction, and #19 and 30 with partial bony impaction. No caries were identified. #I was mobile and near exfoliation.

The patient had #I extracted and sealants placed on #K and 14. A gingivectomy was subsequently performed on #3, 19, and 30 using the Coltene PerFect TCS II high frequency electrosurgery system. The excised gingival specimen were about 5mm in thickness as measured pre-operatively with a dental probe. A thin layer of alveolar bone was removed from #19 and 30 using rongeurs. The dental follicles were disrupted. An operculectomy was completed on #14. The hypertrophic gingiva was preserved in formalin and sent to pathology for assessment.

The pathology report yielded squamous mucosa with scant chronic inflammatory infiltrate, along with fibrous tissue consistent with the dental follicle and fragments of bone with reactive changes.

CLINICAL AND RADIOGRAPHIC PRESENTATION

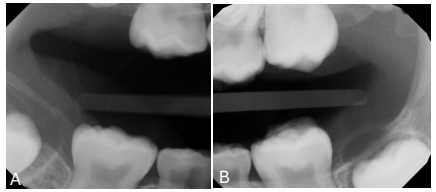

Figure 1, A-B: Initial radiographs [May 2023]
Please note the unerupted first permanent molars. The patient was age 9 at this visit.

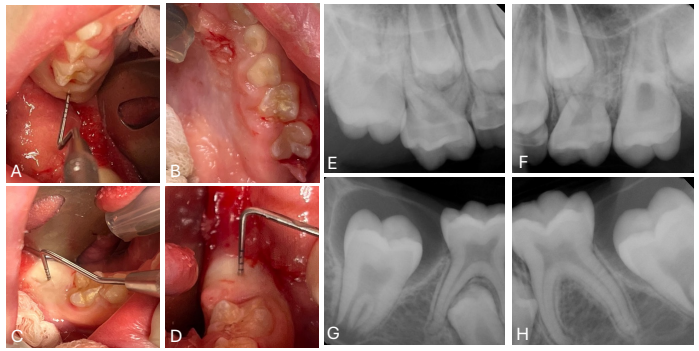

Figure 2, A-H: Pre-operative photographs and radiographs [May 2024]
Pre-operative photographs were taken to demonstrate unerupted first permanent molars and 5mm probing depth of overlying gingiva. Radiographs reinforce the clinical findings, showing soft tissue impaction of #3 and bony impactions of #19 and 30.

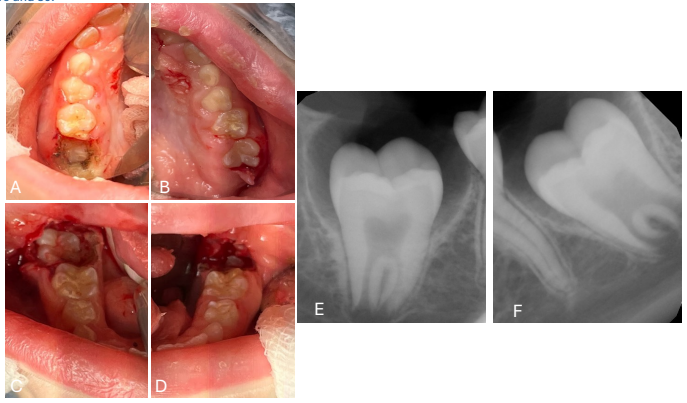

Figure 3, A-F: Post-operative photographs and radiographs [May 2024]
Post-operative photographs were taken to demonstrate exposure of impacted first permanent molars. Radiographs were taken to verify adequate removal of bone to allow for eruption of mandibular first permanent molars.

DISCUSSION

Trisomy 8 mosaicism presents with significant variability in terms of systemic and orofacial manifestations. Facial features may include hypertelorism, deeply set eyes, micrognathia, upturned nose, and large ears. There is scant literature available describing intraoral anomalies, though cleft palate has been noted in many cases. This patient did not present with a cleft palate, but his unerupted first permanent molars were notable.

In normal conditions, a tooth is expected to erupt when root formation is less than ¾ of the expected final length. Delayed eruption is defined as an unerupted tooth with more than ¾ root formation. Primary failure of eruption is evident when there is no detectable obstruction preventing eruption. Common etiologies for delayed eruption and primary failure of eruption can be syndromic, such as in the case of Apert syndrome, or non-syndromic. Physical barriers such as tumors, cysts, scar tissue, thick mucosa, and supernumerary teeth may impede eruption. Localized cases of primary failure of eruption may result from an idiopathic disturbance of the dental follicle which subsequently interferes with the metabolic pathways that facilitate eruption. Some studies have shown that systemic conditions such as endocrine and renal disorders may delay eruption, as growth can be impaired in these cases.

The clinical, radiographic, and histologic findings in this case do not indicate a clear etiology for the delayed eruption of permanent first molars. It is possible that the dense connective tissue in gingival hyperplasia acted as a physical impediment to tooth eruption. This patient also has hypothyroidism, a history of anemia in infancy, as well as renal disease which may be contributing to his dental presentation.

Clinical guidelines recommend active intervention of unerupted teeth when more than 2/3 of the root has developed. In this case, gingivectomy was completed to facilitate an obstructed path of eruption for the permanent first molars, with the ultimate intention of improving function and preventing supra-eruption of opposing teeth. Given that this patient is unable to cooperate for visits in the outpatient dental setting, he will return annually for recalls in the operating room to monitor eruption progress.

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