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CASE REPORT

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PHACES syndrome and multi-regional odontodysplasia: a case report

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Abstract

PHACES syndrome is an acronym for the syndromic presentation of Posterior fossa malformation, Hemangioma, Arterial anomalies, Coarctation of aorta/cardiac defects, Eye abnormalities and Sternal malformations. Infantile hemangiomas are the most common tumors of infancy. Regional odontodysplasia, commonly referred to as "ghost teeth", is a rare localized developmental malformation of enamel and dentin with varying levels of severity that results in unusual clinical and radiographic appearances of affected teeth. This report describes a rare case of a two-year-old Caucasian male diagnosed with PHACES syndrome also presenting with multi-regional odontodysplasia. Ten of twenty teeth were dysplastic. The patient was treated under general anesthesia in a hospital setting. All affected primary teeth were extracted due to sensitivity, abscess and extremely poor long-term prognosis. Moving forward, a long-term interdisciplinary approach will be necessary to address this child's dentition as it develops.

Keywords

Odontodysplasia; PHACES syndrome; Dental anomaly

1. Introduction

PHACES syndrome was first described in 1996 as an acronym for the syndromic presentation of posterior fossa malformation, hemangioma, arterial anomalies, coarctation of aorta/cardiac defects, and eye abnormalities with the acronym later updated to include sternal malformations [1, 2]. The diagnostic criteria for PHACES syndrome includes a large or segmental infantile hemangioma which usually involves the face, scalp or cervical region, and one or more of the above diagnostic features [3]. PHACES syndrome occurs in about two to three percent of children with an infantile hemangioma [4]. PHACES syndrome can have effects on individuals for the duration of their life. Induvials with PHACES syndrome have been shown to suffer neurovascular complications, cardiac concerns and dental issues throughout their life span [5].

Recent studies have demonstrated the presence of enamel hypoplasia and root malformation in primary and permanent dentition of individuals with PHACES syndrome [6, 7]. Intraoral hemangiomas have more commonly been associated with enamel hypoplasia [6]. PHACES syndrome has also been associated with severe deformity or failure of development of the roots of permanent first molars [8]. The prevalence of dental involvement for patients with PHACES syndrome is currently unknown.

Infantile hemangiomas are the most common tumors of infancy [9]. Most infantile hemangiomas are visible at birth or shortly thereafter presenting in three phases—proliferation, stabilization and spontaneous involution [10]. Infantile hemangiomas can be present in multiple segments or regions of facial development [9].

Regional odontodysplasia, commonly referred to as "ghost teeth", is a rare localized developmental malformation of enamel and dentin with varying levels of severity resulting in unusual clinical and radiographic appearances of affected teeth [11, 12]. The condition can affect the primary and permanent dentition and additionally presents with large pulp chambers, short roots and open apices. Teeth effected by regional odontodysplasia can be soft and discolored, slow to erupt and have associated soft tissue swelling. Due to the poorly formed tooth structure these teeth are prone to fistula or abscess even in the absence of dental caries [13, 14]. This can result in the premature loss of primary teeth and the loss of permanent dentition which require prosthetic replacement. The condition is usually unilateral or located in one region or sextant of the mouth [15, 16].

The purpose of this case report is to describe the management and treatment of a patient presenting concurrently with PHACES syndrome and Regional Odontodysplasia requiring significant dental care.

2. Case report

A two-year old Caucasian male, referred from a private practice pediatric dentist and oral surgeon, presented for consultation to the pediatric dental clinical at the University of California San Francisco (UCSF) with complaints of tooth pain and poorly developed teeth. The child was previously managed

at the referring pediatric dental office but due to progressive deterioration of teeth, the child was referred to a hospital based dental clinic. The mother was particularly concerned with the child's extreme sensitivity to toothbrushing.

The patient was born by caesarian section at gestational age of 40 w 1 d. At birth, the patient presented with a hemangioma of the right face and chin. A magnetic resonance imaging and angiography (MRI/MRA) was completed and a work-up for PHACES syndrome was initiated by the primary care provider and a dermatologist. The diagnosis of PHACES syndrome was made based on presences of a hemangioma greater than five centimeters of the cervicofacial and mandibular region of the right face and neck, narrowing of the distal cervical internal carotid arteries, and a hypoplastic transverse arch. No ocular, brain or sternal anomalies were identified. The patient was started on propanolol three days after birth to gradually reduce the visual appearance of the hemangioma. The child had been diagnosed with speech delay. He started receiving speech therapy at twenty months old and was still receiving speech therapy at the time of dental treatment.

There was no known family history of dental anomalies or PHACES syndrome.

2.1 Examination

A comprehensive examination was completed in the dental clinic. Extra-oral examination revealed a facial hemangioma of the right check, chin and neck area (Fig. 1). Intra-oral clinical examination revealed right mandibular, right maxillary and mandibular anterior dysplastic teeth. An abscess was noted buccal to the right primary first molar at the time of clinical exam. All primary teeth were erupted except for second primary molars. Due to odontogenic pain, presence of a dental abscess, severity of disease and inability to cooperate for dental treatment due to young age, the child was scheduled to be treated by pediatric dentistry with general anesthesia at the UCSF Benioff Children's Hospital San Francisco. At time of referral for treatment the child was also prescribed an oral antibiotic, Amoxicillin.

At the time of the procedure approximately one month after the initial consultation, a thorough examination, radiographs and intra-oral photographs were completed in the operating room. At the time of treatment, the upper right first molar (#54), lower left canine (#73), lower anterior primary incisors (#71, 72, 81, 82), and lower right canine (#83), first and second primary molars (#84, 85) appeared hypoplastic (Figs. 2,3). Some of these primary teeth were only partially erupted or broken down. There was a buccal abscess present adjacent to the lower left incisor and lower right first primary molar. Radiographically, upper right primary molars, lower left canine, lower anterior and lower right primary molars appeared dysplastic (Figs. 4,5,6). These teeth had enlarged pulp chambers, very thin enamel and dentin, and wide-open apices. The tooth bud of the upper right and lower right first permanent molars appeared dysplastic (Figs. 4,5). The lower anterior permanent teeth did not yet appear present radiographically (Fig. 6).

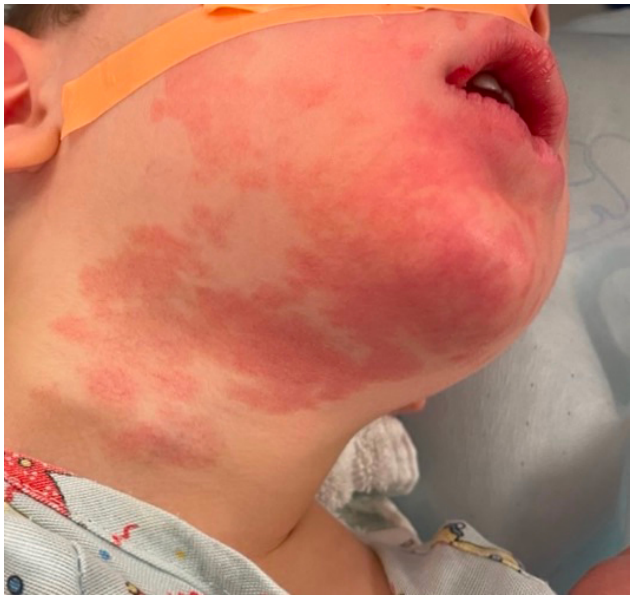

FIGURE 1. Extra-oral image depicting distribution of hemangioma of right cheek, chin and neck.

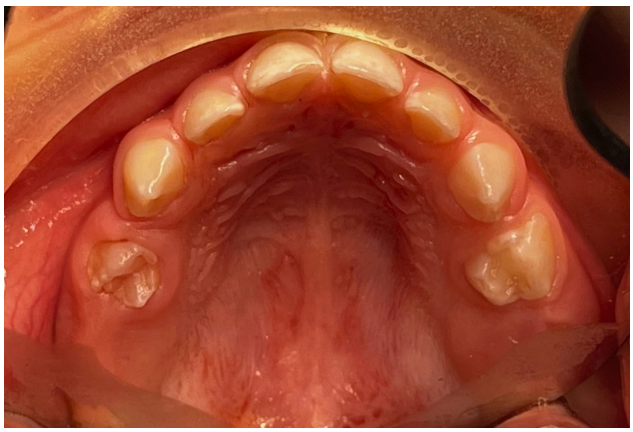

FIGURE 2. Intra-oral photo of maxillary arch prior to treatment. Dysplastic upper right primary first molar is visible in the upper right region, primary second molars on the upper right and upper left quadrants are unerupted.

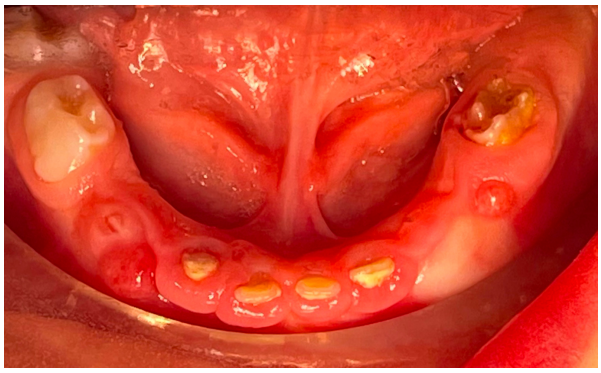

FIGURE 3. Intra-oral photo of mandibular arch prior to treatment. Dysplastic and partially erupted teeth in the anterior and lower right regions. Buccal abscesses are present on the left and right.

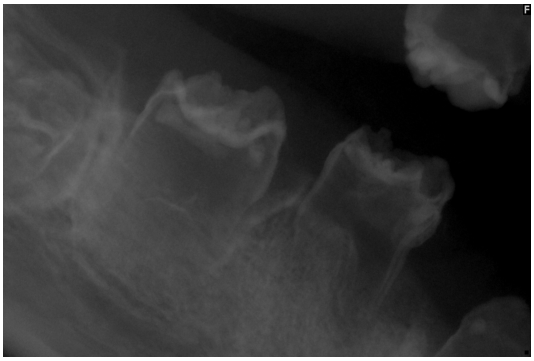

FIGURE 4. Lower right peri-apical radiograph. Lower right primary canine, first molar, and second molar, and first permanent molar are dysplastic. Primary teeth have enlarged pulp chambers and open apices.

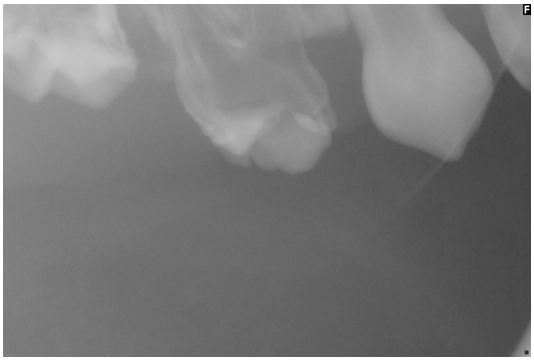

FIGURE 5. Right side maxillary primary teeth radiograph. Upper right primary first and second molar dysplastic. The upper right primary canine appears normally developed.

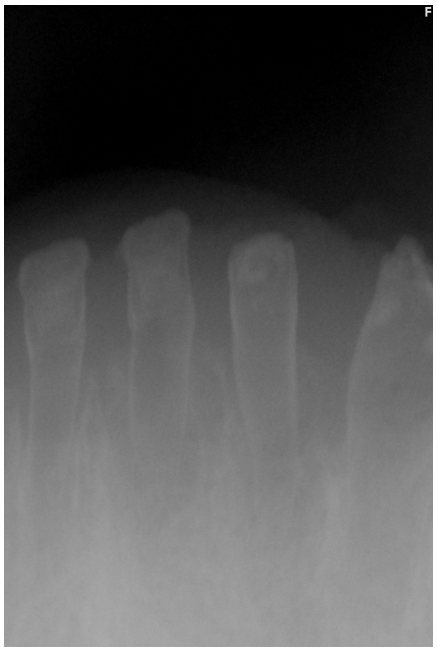

FIGURE 6. Lower anterior periodical radiograph. All lower anterior primary incisors are dysplastic. The permanent successors do not appear to be present.

2.2 Treatment

The upper right first molar, lower left canine, lower anterior and lower right canine and first molar were extracted using periosteal elevators and forceps. Due the extremely thin enamel and dentin structure, the teeth were very fragile mostly removed in fragments. Due to their radiographic presentation and poor long-term prognosis when erupted, the upper right and lower right second primary molar were surgically extracted. Post-operative radiographs were taken to ensure no tooth fragments remained. Multiple simple interrupted gut sutures were placed in the extraction sites to reduce post-operative bleeding. Post-operative instructions were delivered in person and on paper to the family. The patient was discharged home on the same day of treatment.

The extracted teeth were submitted to the UCSF Dermatopathology & Oral Pathology Service for evaluation. The teeth were fixed in 10% neutral buffered formalin followed by decalcification in formic acid and then processed to paraffin to make microtome sections stained with hematoxylin and eosin (H&E). Several malformed teeth were present containing thin, hypoplastic dentin (Fig. 7). One of the teeth contained a pulpal abscess (Fig. 8). The clinical and histological findings are consistent with regional odontodysplasia in the area of the upper right, lower right and lower anterior. These areas all also coincided with the region of the facial hemangioma.

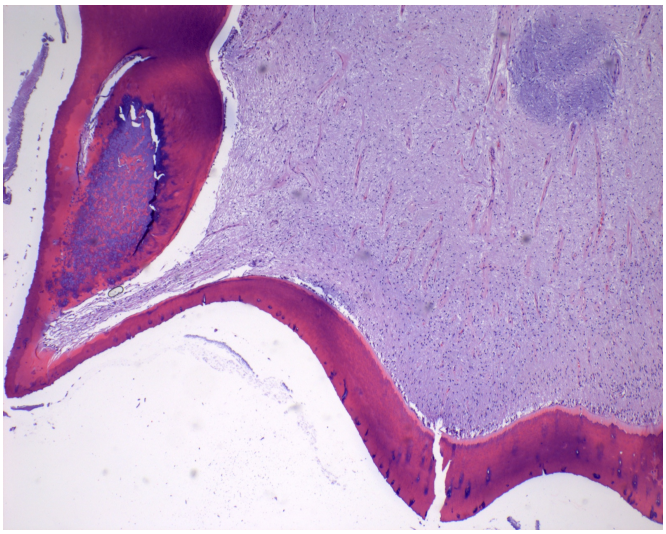

FIGURE 7. Histological section of primary tooth. The tooth has been decalcified and the enamel is not present. The dentin is thin, the tooth is irregularly shaped.

The patient and his mother returned to clinic for evaluation one-month after the procedure. The mother reported minimal post-operative bleeding. The child only required over the counter acetaminophen for post-operative pain management. She also reported that sensitivity to tooth brushing was much improved following removal of the dysplastic teeth.

3. Discussion

While regional odontodysplasia is widely known to be unilateral, there are other reported cases where regional odontodysplasia has crossed the midline, these cases included a thirteen-

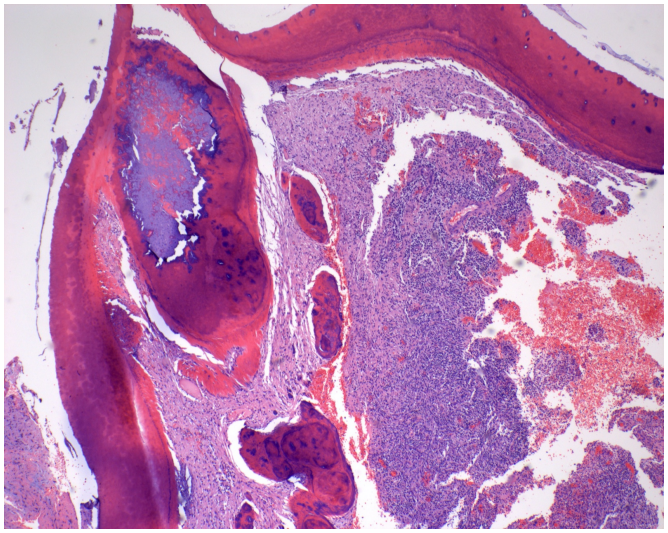

FIGURE 8. Histological section of extracted primary tooth. The tooth has been decalcified and the enamel is not present. The dentin is thin, malformed dentin is present under the cusp tip, and pulpal inflammation with abscess is present.

and twenty-year-old patient [17, 18]. This case was a very rare presentation of odontosyplasia presenting in half of all primary teeth and affecting the developing tooth buds in the upper right, lower right and lower anterior regions of the mouth. This case illustrates a rare example of a child with multi-regional odontodysplasia concurrently with the diagnosis of PHACES syndrome.

Prior cases have recommended a conservative management of regional odontosdyplasia. However, due to the severe clinical presentation of this case, a more extensive approach was needed as multiple non-carious teeth had abscessed at the age of two due to extremely thin enamel and dentin. The extremely thin enamel and dentin that presented in this case made the teeth non-restorable (Fig. 4). The teeth were unable to be prepared for full coverage restorations, as there would not have been sufficient tooth structure left to support the restorations. Furthermore, the irregular and very large pulp chambers did not allow for good prognosis for any pulpal treatments. However, premature removal of primary teeth will likely impact the eruption of the second primary molars, the permanent molars and pre-molars and the overall occlusion. Several teeth may erupt ectopically or be delayed in eruption. Future management as this child grows will require possible space management and orthodontic, intervention.

The etiology of regional odontodysplasia is not well known. Possible etiologies suggest trauma, infection, genetic mutation and disturbances in blood supply to developing teeth [17].

Prior studies have demonstrated that PHACES can impact tooth structure, with published cases involving mostly malformed roots and enamel hypoplasia [18, 19]. Another published case report similarly described a patient with bilateral mandibular regional odontosyplasia co-occurring with the presence of vascular nevus of the face and neck [20]. No other reported examples of regional odontodysplasia and PHACES syndrome could be identified. Given that the areas of the mouth that had dysplastic tooth structures also aligned with the areas of the facial hemangioma it might be hypothesized that the hemangioma contributed to an irregular blood supply to the areas of tooth bud development and resulted in subsequent abnormal tooth development. This would conform to previously described etiologies of disturbances in blood supply resulting in regional odontodysplasia [9].

One major concern in the treatment of this child was the potential for uncontrollable bleeding at the time of tooth extraction. Consultation with pediatric dermatology and review of prior imaging (MRI/MRA) in this case indicated that there was a low risk for high flow underlying vascular malformations in the region of the teeth to be extracted. There was no unusual bleeding observed intra- or post-operatively in this case, however, this case highlights the need for interprofessional consultation when planning for treating higher risk patients with complex and rare medical conditions.

The treatment and interventions needed for this child's dental condition will continue into adulthood. In most cases of regional odontodysplasia impact both the permanent and primary dentition. A long-term interdisciplinary approach will be necessary to address this child's dentition as it develops. This will likely include the need for prosthetic tooth replacement including removal appliances and implants [21]. After this initial phase of treatment, the child's odontogenic pain and dental abscess have been treated, and the child's oral health related quality of life has improved [22]. The pediatric dentist has a crucial role in the early diagnosis and management of young patients with PHACES syndrome. This case demonstrates how the pediatric dentist serves as part of the interdisciplinary team in managing rare and complex syndromes and diseases impacting the craniofacial complex.

4. Conclusions

PHACES syndrome with concurrent with Regional Odontodysplasia affects multiple aspects of the craniofacial complex and requires an interdisciplinary approach to comprehensive management and treatment including appropriate and safely rendered dental care.

AVAILABILITY OF DATA AND MATERIALS

Not applicable.

AUTHOR CONTRIBUTIONS

JMS—completed the clinical case, and initiated the writing of the manuscript. RES and RCJ—provided consultation on treatment, and contributed to and revised the manuscript. RCJ—completed histological evaluation of specimens. All authors read and approved the final manuscript.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

Informed consent was completed with the parent of the patient prior to the procedure following hospital guidelines. Written informed parental consent was also obtained for specimen

collection for analysis and reporting of the clinical case after treatment. Case reports presenting less than three de-identified cases are deemed exempt by the University of California San Francisco Institutional Review Board.

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CONFLICT OF INTEREST

The authors declare no conflict of interest.

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