Case report of segmental odontomaxillary dysplasia with cutaneous manifestations

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DOI 10.23804/ejpd.2020.21.03.14

Abstract

Backgroud Segmental odontomaxillary dysplasia is an uncommon nonhereditary growth disorder that affects the maxilla, gums and ipsilateral dentition. The disorder is diagnosed mainly based on dental (over-retention of primary teeth, dental agenesis and diastemas) and bone findings (bone sclerosis, irregular trabeculation of immature bone and reduced maxillary sinus). This paper provides a case report.

Case report A 5-year-old child with skin manifestations including hypertrichosis, facial erythema and pigmented nevus was diagnosed with type II segmental odontomaxillary dysplasia based on clinical, radiographic and histopathological analysis.

Conclusion The skin findings can help with the suspicion of segmental odontomaxillary dysplasia, although the definitive diagnosis is typically established by a paediatric dentist based on clinical and radiological findings.

KEYWORDS Segmental odontomaxillary dysplasia, dental agenesis, hypertrichosis.

Introduction

In 1987, Miles et al. reported on two patients with congenital facial asymmetry and unilateral enlargement of the maxillary bone and ipsilateral gingiva, which they called hemimaxillofacial dysplasia. To define this uncommon condition, the term segmental odontomaxillary dysplasia (SOD) was coined [Danforth et al., 1990]. The diagnosis is based mainly on dental (over-retention of primary teeth, agenesis of premolars, delayed tooth eruption and diastemas) and radiological findings (bone sclerosis, thick and irregular trabeculation of immature bone, reduced maxillary sinus and confirmation of dental agenesis) [Yassin and Rihani, 2008]. Although histopathology is typically unnecessary, it can help establish the differential diagnosis with fibrous dysplasia [Smith et al., 2018].

Although the aetiology of SOD remains unknown, it has been suggested that SOD is a nonhereditary anomalous development of the first branchial arch [Danforth et al., 1990], with a slight predominance in the male sex, with no preference for the left or right side [Smith et al., 2018].

To date, there have been 65 cases described in the English scientific literature; however, SOD is a disease that is often incorrectly diagnosed or goes unnoticed by medical and dental healthcare practitioners, including paediatric dentists [Smith et al., 2018].

This paper presents a case report of segmental odontomaxillary dysplasia (SOD) with skin manifestations including hypertrichosis, facial erythema and pigmented nevus.

Case report

A 5-year-10-month-old child with no relevant personal or family medical history was admitted to the paediatric dentist for pain of presumably dental origin. During the extraoral examination, pigmented nevus was observed on the left eyebrow, as well as mild ipsilateral facial erythema, skin hyperpigmentation and hypertrichosis on the upper left lip

(Fig. 1). During the intraoral examination, we observed thickening of the upper left maxilla from the primary maxillary left canine to the tuberosity. The thickened area was hard to the touch, asymptomatic, fully covered by mucosa and had a normal appearance. The clinical crowns of the teeth involved (primary maxillary left canine, primary maxillary left first molar and primary maxillary left second molar) were only partially visible (Fig. 1). The presence of several caries was also noted, which explained the onset of pain. Agenesis of the definitive ipsilateral premolars was radiologically confirmed, as was characteristic bone trabeculation with thick and vertical lines and a significantly reduced left paranasal sinus (Fig. 2).

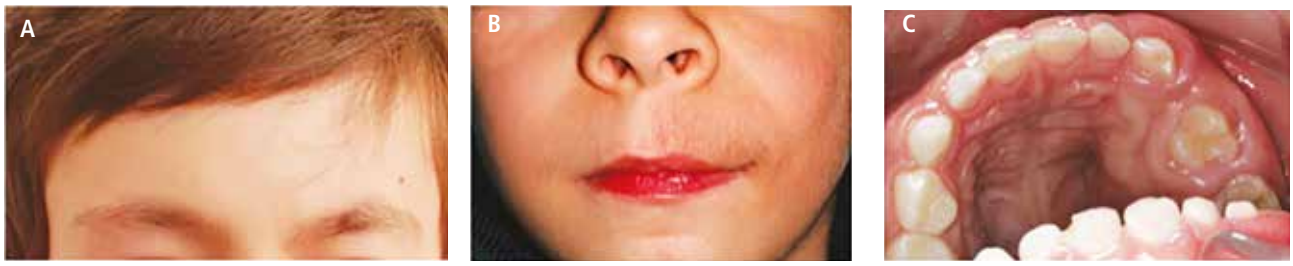

FIG. 1 Clinical findings of segmental odontomaxillary dysplasia. A) Pigmented nevus on the left eyebrow. B) Hypertrichosis and slight redness on the left cheek. C) Thickening of the left superior maxilla, with over-retention of the primary maxillary left first and second molars.

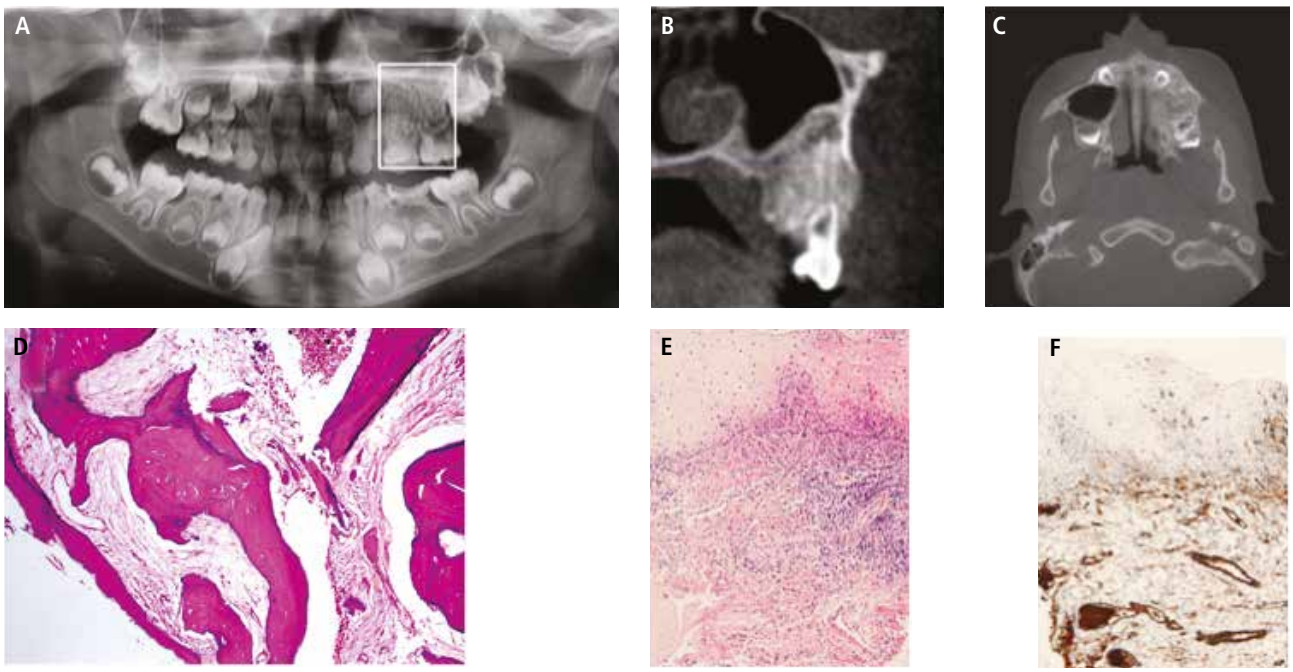

FIG. 2 Radiological and histopathological findings. A) agenesis of maxillary left premolars; B) abnormal bone trabeculation; C) reduced left maxillary sinus; D) large bone trabeculae with no osteoblastic or osteoclastic activity, nor inflammation, with occasional small and irregular bone spicules in a fibromixoid stroma (HE, x10); E) gingival mucosa with epithelial hyperplasia, subepithelial fibrosis and vascular structures surrounded by a moderate lymphocytic inflammatory infiltrate (HE, x10); F) high density of blood vessels confirmed with immunohistochemistry using an endothelial marker (CD31, x10).

	Diagnosis	Natural history	Etiology	Asymmetry	Hypertrichosis	Facial pigmentation	Gingival enlargement	Topography
Segmental odontomaxillary dysplasia	Childhood-adolescence	Not progressive	Growth disorder of unknown cause	Unilateral maxillary and facial	Present in some cases	Present in some cases	+	Unilateral maxillofacial
Regional odontodysplasia	Childhood	Not progressive	Growth disorder of unknown cause	-	-	-	-	Teeth; more common in a single quadrant
Monostotic fibrous dysplasia	First or second decade	Progressive til growth is completed	Growth disorder of unknown cause	Unilateral maxillary and mandibular	-	-	-	Maxillary and mandibular

TABLE 1 Differential diagnosis of segmental odontomaxillary dysplasia.

Histopathology of the gingival mucosa showed discrete epithelial hyperplasia, subepithelial fibrosis and numerous vascular structures. The bone biopsy showed a predominance of lamellar bone, with no peripheral osteoblast activity (Fig. 2). Based on these findings, the diagnosis of SOD was established.

Discussion

Approximately half of patients with SOD have reported skin manifestations, including hypertrichosis, facial erythema, lip hypopigmentation, Becker's nevus, pigmented nevus, discontinuity of the vermilion border and cheek depression [Shah et al., 2012; Becktor et al., 2002; Jones and Ford, 1999; DeSalvo et al., 1996; Packota et al., 1996]. Consequently, the abbreviation HATS (hemimaxillary enlargement, asymmetry of the face, tooth abnormalities and skin findings) has been proposed to describe the spectrum of abnormalities associated with SOD [Welsch and Stein, 2004]. Based on the presence and type of facial skin lesions, a new clinical classification for SOD has been proposed [Yassin and Rihani, 2008], which recognises 2 main types: type 1 or classical, with gingival-dental-alveolar manifestations but no extraoral involvement, and type 2 or cutaneous, with dermato-gingival-dental-alveolar involvement. Four subtypes are differentiated in type 2 SOD: IIa hypertrichotic, IIb pigmentary, IIc erythematous and IId commissural defect. Combinations of the subtypes are possible, such as IIab (hypertrichotic-pigmentary), IIabd (hypertrichotic-pigmentary-commissural) and IIbc (pigmentary-erythematous).

The present patient met the clinical and radiographic criteria for SOD and presented cutaneous signs that helped establish the diagnosis of type 2 SOD. The differential diagnosis should be established mainly with regional odontodysplasia and monostotic fibrous dysplasia (Table 1) [DeSalvo et al., 1996]. When faced with the presence of the previously mentioned skin manifestations, the patient should be referred to a dentist for a clinical and radiological assessment. In this case, the dermatologist should also participate in the patient follow-up, given that ipsilateral skin changes can occur over time [Minett and Daley, 2012].

When performing an early diagnosis of SOD, the primary objective is to maintain the deciduous teeth and facilitate occlusion. If the molars of the primary dentition are completely covered by the gum, ulectomy might be indicated to facilitate their eruption [González-Arriagada et al., 2012; Kuklani and Nair, 2010]. There are cases in the literature of children with SOD with successful orthodontic treatments in the short and long term [Whitt et al., 2011; Drake, 2003]. There have also been published cases of adults satisfactorily rehabilitated with tooth-supported [Ozpinar et al., 2009] or implant-supported prostheses [Becktor et al., 2002; Whitt et al., 2011].

SOD is an uncommon disorder that can be incorrectly diagnosed or go unnoticed. Its prevalence is therefore likely much higher than that reported in the literature. Generally, SOD is diagnosed by pediatric dentists, who should understand its functional and aesthetic implications and reassure the patient's family when faced with the onset of the intraoral and, eventually, skin signs of SOD at a very early age.

Bullet points

SOD prevalence is likely much higher than that reported in the literature, as it can be incorrectly diagnosed or go unnoticed.

SOD is diagnosed mainly based on dental and bone findings, but some skin manifestations can help with the suspicion of this disorder.

Paediatric dentists should know the functional and aesthetic implications of SOD and reassure the patient's family when faced with this disorder at an early age.

Acknowledgements

Dr. Márcio Adjudarte Lopes provided the histopathologic image of the bone biopsy.

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