

Case Report

Segmental odontomaxillary dysplasia: report of a case emphasizing histopathological, immunohistochemical and scanning electron microscopic features

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Abstract: Segmental odontomaxillary dysplasia (SOD) is a rare developmental disorder of the maxilla, and there is little information on its morphological features. Thus, the present article describes a case of SOD focusing on its histopathological, immunohistochemical and scanning electron microscopic features. Several dental abnormalities were present, including numerous dentin and pulp defects, altered composition of hard tissue, and proliferation of myofibroblasts in the pulp and the soft tissue surrounding affected teeth. This myofibroblastic proliferation was identified for the first time in SOD and may be involved in both bone and tooth resorption mechanisms. (J Oral Sci 55, 259-262, 2013)

Keywords: segmental odontomaxillary dysplasia; immunohistochemistry; myofibroblast; scanning electron microscopy; teeth.

Introduction

First described as hemimaxillofacial dysplasia in 1987, segmental odontomaxillary dysplasia (SOD) is a rare

developmental disorder involving the posterior maxilla. It was later renamed SOD due to the presence of dental and bone abnormalities, even with a lack of constant facial abnormalities (1,2). There are around 50 well-documented cases of SOD reported in the literature (3), and most of these report the clinical, radiological, diagnostic and treatment features of the condition. This paper reports a case of SOD and describes the microscopic features, via light microscopic, immunohistochemical and scanning electron microscopic (SEM) studies, of teeth, bone and soft tissue affected by SOD.

Case Report

A 6-year-old Caucasian girl was referred to the Pediatric Dentistry Clinic, School of Dentistry, State University of Rio de Janeiro, Brazil, complaining of a painless swelling on the right maxilla lasting for months. Intraoral clinical examination revealed a poorly defined bone swelling covered by normal mucosa located on the posterior segment of the upper right alveolar process in the region of both primary molars (Fig. 1a). Extraoral clinical examination revealed no abnormalities. Panoramic and periapical radiographs showed an altered bone pattern with a mottled ill-defined radiopacity, exhibiting a vertical orientation of trabecular bone, superimposed over primary molar roots. Radiographically, both primary molars had an irregular morphological aspect or external resorption, with both premolars in the region being absent (Fig. 1b),

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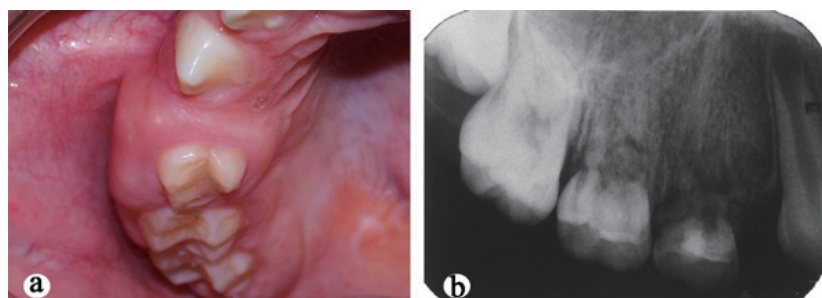


Fig. 1 Clinical and radiological features in segmental odontomaxillary dysplasia. **a** Right maxillary diffuse swelling covered by normal oral mucosa. **b** Periapical radiograph of the same region revealing diffuse radiopacity with vertical trabeculae orientation and missing premolars. Note the irregular root resorption of both primary molars.

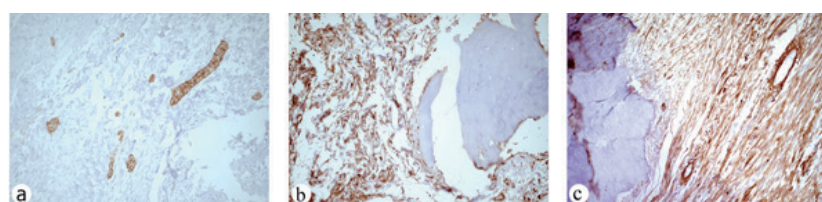


Fig. 3 Immunohistochemical features in segmental odontomaxillary dysplasia (immunoperoxidase, original magnification, $\times 200$). **a** Cytokeratin 19 highlighting odontogenic epithelia in bone. **b** Smooth muscle actin characterizing myofibroblasts surrounding the bone tissue. **c** And inside the pulp chamber and root canal of the affected tooth.

and the maxillary sinus had normal appearance. Based on clinical and radiological characteristics, a diagnosis of SOD was rendered and the patient was managed through extraction of primary molars and removal of excessive adjacent bone and gingival tissues, allowing a regular contour for the maxillary process.

Macroscopically, the affected teeth showed normal crown morphology, except for the presence of some fissures, but abnormal root morphology with a prominent splayed architecture and an irregular resorption pattern was observed in both teeth. Bone and gingival tissues were histologically processed, the former being previously decalcified in 5% nitric acid. Half of each tooth was decalcified for microscopic evaluation and the other half was maintained in 10% formaldehyde for further SEM evaluation.

Microscopically, the 5- μ m hematoxylin and eosin stained sections confirmed that gingival tissue was otherwise normal, whereas bone tissue from the area revealed a high cellular trabecular bone with a number of irregular basophilic reversal lines (Fig. 2a) surrounded by a spindle cell fibrous tissue proliferation, blood vessels and several epithelial cords and nests. Osteoblasts and osteoclasts were scarce in the specimens. The tooth had both coronal and root tubular dentin defects, particularly near the pulp chamber, which had an irregular interface with dentin

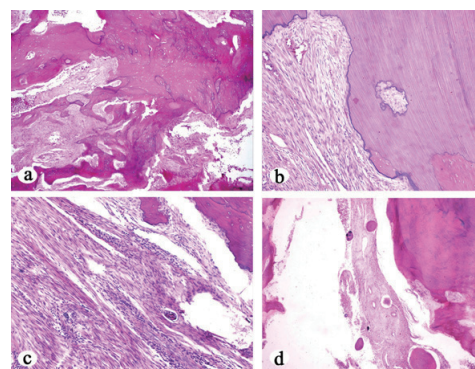


Fig. 2 Light microscopic features in segmental odontomaxillary dysplasia (hematoxylin and eosin, original magnification, $\times 100$). **a** Trabecular bone with numerous basophilic reversal lines among cellular fibrous tissue. **b** Tooth specimen showing dentin defects, irregular outline of the pulp chamber without an odontoblast layer. **c** Spindle cell proliferation inside the pulp tissue, associated to an inflammatory infiltrate. **d** Numerous pulp stones of various sizes.

and absence of the odontoblast layer (Fig. 2b). Fibrosis with numerous spindle cells, an inflammatory infiltrate predominantly composed of plasma cells and several variable sized pulp stones were also observed in the pulp tissue (Figs. 2c and 2d).

Immunohistochemistry was performed on 3- μ m sections mounted on glass silanized histological slides stained with antibodies against high molecular weight pan-cytokeratin (clone 34 β E12, dilution 1:200; Dako A/S, Glostrup, Denmark), cytokeratin 19 (clone RCK108, dilution 1:400; Dako A/S), alpha-smooth-muscle actin (clone 1A4, dilution 1:400; Dako A/S), muscle-specific actin (clone HHF35, dilution 1:800; Dako A/S), desmin (clone D33, dilution 1:1000; Dako A/S) and h-caldesmon (clone N-CD, dilution 1:400; Dako A/S). For the reactions, pressure cooker antigen retrieval using citrate buffer and overnight incubation with primary monoclonal antibodies and conjugated secondary antibodies (LSAB system; Dako A/S) were followed by the use of diaminobenzidine as the chromogen and Mayer's hematoxylin as the counterstain. Bone-tissue immunohistochemical analysis highlighted epithelial remnants being positive for high molecular weight pan-cytokeratin and cytokeratin 19 (Fig. 3a) and spindle cells being positive for smooth-muscle actin (Fig. 3b) but negative for muscle-specific actin (HHF35), desmin and H-caldesmon.

Table 1 Mean Calcium (Ca) and Phosphorus (P) content values (weight percent) and Ca/P ratio identified through X-ray microanalysis in different dentinal areas from affected tooth in segmental odontomaxillary dysplasia (SOD) and from healthy first primary molar

Dentin areas	X-ray microanalysis		
	Ca (%)	P (%)	Ca/P
Visible healthy dentin in SOD	70.5	28.9	2.4
Dentin defects in SOD	67.2	26.3	2.6
Visible healthy dentin in a first primary molar	68.8	23.3	3.0

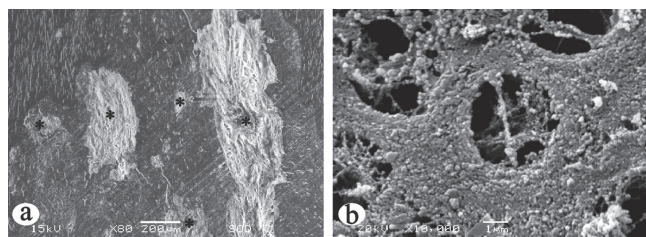


Fig. 4 Scanning electron microscopic features in segmental odontomaxillary dysplasia. **a** Longitudinal section showing tubular dentin defects in coronal area (black asterisks) (original magnification, $\times 80$). **b** Transverse section showing peritubular and intertubular dentin with minor structural differences (original magnification, $\times 10,000$).

Similarly, teeth were positive for smooth-muscle actin only in the pulp chamber (Fig. 3c), and negative for other mesenchymal immunomarkers.

For SEM analysis (JSM 5600 LV; JEOL, Tokyo, Japan), non-decalcified teeth were cleaned with diamond paste, polished and ultrasonicated in water for 5 min, treated with 37% phosphoric acid for 5 min, and covered with a thin layer of carbon. This process was repeated after transverse sectioning. A normal healthy primary molar was used as a control for SEM study at 15 and 20 kV using a backscattered electron detector to determine surface morphology and X-ray microanalysis to determine the mineral composition. Ultrastructurally, the enamel surface showed normal morphology, but tubular dentin showed several changes. Numerous structural tubular dentin defects were observed, including a decreased concentration of collagen fibers and increased similarity among intratubular, peritubular and intertubular dentin (Fig. 4). In addition, X-ray microanalysis showed a similar calcium (Ca)/phosphorus (P) ratio inside and outside of dentin defects in affected teeth, but a minor ratio in comparison with healthy deciduous dentin (Table 1).

After surgery, healing was uneventful and a removable provisional appliance was inserted onto the affected region. The patient has been clinically and radiographically followed for six years, without any signs of recurrence, and has been subjected to annual replacement

of the removable appliance, waiting for definitive prosthetic rehabilitation.

Discussion

SOD is an unusual developmental disorder presenting well-recognized clinical and radiological characteristics (1,2). It is characterized by unilateral enlargement of the posterior segment of the maxilla that may cause facial enlargement and, as a result, gingival fibrous hyperplasia, bony expansion or both (3,4). Extension of maxillary swelling is variable and can include a solely alveolar process from the canine to the tuberosity or extend up to the floor of the maxillary sinus and orbit (4,5). Skin facial findings such as hypertrichosis, erythema and hypopigmentation may also be present (5); however, in this case, the patient showed only intraoral swelling caused by osseous growth without any facial alteration.

Gingival/alveolar mucosal soft tissue in SOD is thick and presents slight to dense fibrosis without inflammation (1,2,5), but may also exhibit a loosely mucoid material similar to perifollicular tissues (2,4), a feature not observed in our case. Radiographically, the affected area shows an ill-defined radiopacity resembling fibrous dysplasia (2,5), and bone changes were described as an increased sclerosis with mediolateral expansion, and thickened and vertically oriented trabeculae appearing indistinguishable from the root images (2,5).

Histologically, bone features in SOD include a large amount of dense or loosely arranged collagen fibers surrounded by immature cancellous bone with prominent reversal lines in a hypocellular stroma (1,3-5). Despite the aforementioned characteristics, affected bone in the present case showed spindle cell proliferation and a large number of odontogenic epithelial cords and nests.

Teeth abnormalities in SOD include agenesis of one or both premolars in the affected area, delayed eruption of the primary molars or the adjacent permanent molars, malformation of both deciduous and permanent teeth including hypoplasia, enlarged crowns and roots, splayed roots, pulp stones, irregular root resorption, root abnormalities of the primary molars and abnormal

spacing between erupted teeth (2,4,5). One study has been conducted analyzing microscopic features of three primary teeth affected by SOD; its major findings included the presence of tubular dentin defects in the circumpulpal area, enlarged pulp chamber and root canals, moderate cellular and vascular pulp fibrous tissue, a number of pulp stones, irregular outline dentin, absence of a peripheral odontoblast layer, and apical external resorption (6). These findings were also observed in our case. In the present study, spindle cell proliferation inside the pulp and bone-associated fibrous tissue was observed. Immunohistochemically, these cells were thought to be myofibroblasts. These cells have already been described to be associated with different pathologic processes such as reactive, benign and malignant lesions (7-9), and one study reported a correlation between the prominent presence of myofibroblasts in the stroma and the rupture of cortical bone in ameloblastomas (10). Myofibroblast occurrence in SOD may be justified, as these cells are probably involved in the tooth and bone resorption observed in this condition.

Our electron microscopic findings reproduced the microscopic structural dentin defects observed by Armstrong et al. in 2004 (6). An interesting observation was the similar Ca/P ratios identified in both visibly healthy and defective dentin from the affected teeth, which differed from the visibly healthy dentin in similar, normal teeth, thus suggesting a compositional modification through all the dentin in SOD. Enamel imperfections have previously been described in some cases (1,2), but this SEM study was unable to detect any variations in enamel structure in the affected teeth.

Despite some typical clinical and radiological characteristics, SOD may be misdiagnosed as fibrous dysplasia, segmental hemifacial hypertrophy, regional odontodysplasia, focal osseous dysplasia and gingival fibromatosis (2,4-6), thus reinforcing the importance of describing the clinicopathologic features of such a rare disorder.

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