Segmental Odontomaxillary Dysplasia: Report of 3 Cases and Literature Review

Wilfredo Alejandro González-Arriagada ·
Pablo Agustin Vargas · Rodrigo Fuentes-Cortés ·
Marco Antonio Nasi-Toso · Marcio Ajudarte Lopes

Received: 5 September 2011 / Accepted: 17 November 2011 / Published online: 3 December 2011
© Springer Science+Business Media, LLC 2011

Abstract Segmental Odontomaxillary Dysplasia (SOD) is an uncommon developmental disorder of unknown etiology that causes a unilateral alteration of the maxilla associated with an abnormal growth and maturation of bone, lack of one or both premolars and delayed tooth eruption on the affected side, alteration of adjacent gingival tissue with or without facial cutaneous lesions. Radiographically is observed an irregular trabecular pattern and reduction of the maxillary sinus. There are 50 cases reported in the English-language literature. A literature review is herein presented emphasizing the clinical, radiographic, and histological features, and three additional cases of SOD affecting a 22 years-old woman, a 18 years-old man, and a 5 years-old boy, respectively are described.

Keywords Segmental odontomaxillary dysplasia · Hemimaxillofacial dysplasia · Facial asymmetry · Maxillary diseases

Introduction

Segmental odontomaxillary dysplasia (SOD) is an uncommon disorder that causes a unilateral development of disturbance of the maxilla associated with an abnormal growth and maturation of bone, teeth, and adjacent gingival tissue, with or without facial cutaneous lesions [1–3]. The etiopathogenesis is unknown [1, 3, 4]. This disease was initially reported in 1987, by Miles et al. as hemimaxillofacial dysplasia (HMD) [5]. Danforth et al. [6] subsequently proposed the term SOD instead of HMD. In this disorder, the maxilla is affected unilaterally causing enlargement and, in some cases, facial asymmetry, accompanied by delayed eruption, absent premolars and tooth spacing [2]. The age of onset ranges from 5 to 27 years but is commonly diagnosed in the first decade of life. There is no gender predilection, although some studies report a male predominance [3, 4]. Any side can be equally affected [3]. Hypertrichosis and commissural lip clefting has been reported in the affected side [2, 4]. Welsch and Stein in 2004 used the acronym HATS (hemimaxillary enlargement, asymmetry of the face, teeth abnormalities, and skin findings) when skin hyperpigmentation is also observed [7]. Radiographically, an ill-defined radiopacity with a ground glass appearance is observed [2–4, 8]. Reduction of the maxillary sinus has also been reported [1]. Histologic examination of the bone described thick bone trabeculae formed by an irregular and immature bone with absence of lamellar bone and osteoblastic/osteoclastic rimming [1, 3]. Mitosis and cellular pleomorphism are not observed [3]. Histologic examination also showed alterations of teeth. Armstrong et al. [9] reported fibrous enlargement of the pulp of deciduous teeth, an irregular pulp/dentin interface, pseudoinclusions, and pulp stones. Irregular tubular dentin and deficient odontoblast layer was also reported.

W. A. González-Arriagada · P. A. Vargas · M. A. Lopes
Oral Diagnosis Department, Semiology and Oral Pathology,
Piracicaba Dental School, State University of Campinas
(UNICAMP), Piracicaba, Sao Paulo, Brazil

W. A. González-Arriagada · R. Fuentes-Cortés ·
M. A. Nasi-Toso
Oral Pathology and Diagnosis, Facultad de Odontología,
Universidad de Valparaíso, Valparaíso, Chile

M. A. Lopes (✉)
Área de Semiologia, Faculdade de Odontologia de Piracicaba-
UNICAMP, Av. Limeira, 901, Bairro Areão, Piracicaba,
São Paulo CEP: 13.414-903, Brazil
e-mail: malopes@fop.unicamp.br

Report of Cases

Case 1

A 22-year-old female was referred to the Orocentro (Oral Diagnosis Clinic, Piracicaba Dental School-UNICAMP) complaining of a painless expansion of the right side of the maxilla and absence of premolars on the affected side, which led her to seek information about having implants in the area. There was no significant past medical history and family history was noncontributory. Extraorally, there was a slight facial asymmetry, hypertrichosis, and commissural lip clefting on the right side (Fig. 1a). Intraoral examination revealed a buccal and palatal expansion of the right maxilla, midline deviation and absence of the right maxillary premolars (Fig. 1b).

The patient brought past radiographs taken when she was 4 years old. These radiographs revealed agenesis of the maxillary premolars and advanced eruption of the first definitive molar on the right side (Fig. 2a). Two years later, when she was 6 years old, all the first definitive molars had erupted and bone of the affected side revealed a slight

ground glass appearance (Fig. 2b). Between the ages of 6 and 18, the patient was under close follow-up by her dentist, and tooth eruption occurred normally in the non-affected side, whereas the affected side revealed alteration in normal teeth eruption and maxillary growth. The patient was under orthodontic treatment to improve the midline deviation. When she was 18 years old, the orthopantomography showed absence of the third molars and tooth buds on the affected side, in the maxilla, and in the mandible. Radiopaque bone trabeculae in the premolar area of the right maxilla and a decrease of the maxillary sinus were also observed (Fig. 2c). The current panoramic radiography showed an ill-defined radiopacity in the affected area and an almost imperceptible maxillary sinus (Fig. 2d). The computed tomography confirmed the presence of a poorly-defined lesion and decrease in the size of the maxillary sinus on the right. An alteration in the anatomy of the nasal osseous structures with deviation of the nasal septum was also observed (Fig. 3).

An incisional biopsy of affected bone and gingival tissue was performed. Histologic examination revealed large bone trabeculae without osteoblastic or osteoclastic

Fig. 1 Clinical pictures of case 1. **a** Extraoral photograph showing a mild commissural lip cleft and hypertrichosis on the affected side. **b** Intraoral view showing vestibule-palatine expansion on the posterior *right* side and the absence of premolars

Fig. 2 Panoramic radiographs of case 1. Involvement of the maxillary right sinus, changes in the radiographic pattern and absence of the *upper right* premolars and superior and inferior third molars. **a** 4 years old, **b** 6 years old, **c** 18 years old, **d** 22 years old

Fig. 3 Computed tomographic imaging of case 1. **a** Axial CT view showing the altered trabeculae and osseous expansion inside the maxillary sinus. **b** Axial CT view showing the unilateral maxillary enlargement and altered bone of the *right side*. **c** In the frontal

(coronal) view, the maxillary lesion is noted and seen to encroach upon the right maxillary sinus, diminishing its size, as well as marked deviation of the nasal septum

rimming, absence of inflammation, and a fibrous and myxoid stroma containing few small caliber vessels, compatible with the diagnosis of SOD (Fig. 4). The gingival mucosa was hyperplastic and focally inflamed, but without cellular pleomorphism, atypical mitoses or necrosis.

The patient and her parents were educated about the diagnosis and informed that there were only two case reports in the English language literature of patients with SOD who underwent implants. Although success was achieved in these two patients, osseointegration could not be guaranteed and other options to rehabilitate the edentulous area needed to be considered.

Fig. 4 Photomicrograph obtained from the biopsy of case 1. Note the altered large bone trabeculae with reversal lines, and lack of inflammatory cells and osteoblastic or osteoclastic rimming. The stroma is composed of a fibrous and myxoid tissue (hematoxylin eosin, original magnification $\times 200$)

Case 2

A 17-year-old male was referred to a private consult of Viña del Mar, Chile, complaining of a painless enlargement of the right maxilla and tooth absence on the same side. The past medical and family histories were not contributory. Extraorally, facial asymmetry without cutaneous findings was observed. On intraoral examination, features similar to case 1 were seen, including vestibular and palatine bone expansion of the right maxilla, a slight midline deviation, and absence of premolars at the affected side (Fig. 5).

The orthopantomography showed a poorly-defined lesion, less radiopaque than the normal bone in the right maxilla, and absence of the maxillary premolars in the affected area. The molars of the right maxilla showed partial root formation (Fig. 6). The eruption of these

Fig. 5 Intraoral view of case 2. Unilateral expansion of the right maxilla with absence of premolars

Fig. 6 Panoramic radiograph of case 2. Altered bone pattern, root formation and eruption of the *right upper* molars

molars was also altered when compared to other quadrants of the mouth.

With a clinical working diagnosis of a “benign fibro-osseous lesion”, an incisional biopsy was performed. Histologically, large and irregular bone trabeculae, with scarce osteoblasts and osteoclasts were observed. There was no inflammation and the stroma was composed of adipose and fibro-myxoid tissue, compatible with SOD.

Case 3

A 5-year-old male was referred to Orocentro (Oral Diagnosis Clinic, Piracicaba Dental School-UNICAMP) with a painless unilateral enlargement affecting the right maxilla. His parents did not share any medical or family history information that could contribute to the diagnosis. The extraoral examination revealed facial asymmetry and cutaneous findings, including unilateral lip clefting and hypertrichosis on the affected side. On intraoral examination, similar features to the previous cases were observed, including bony expansion of the right maxilla, midline deviation with a diastema between the canine and the first deciduous molar and absence of the second deciduous molar on the affected side (Fig. 7).

Radiographically, an ill-defined radiodensity with altered bone trabeculae and a corresponding smaller maxillary sinus of the right side when compared with the non-affected side was found. The altered formation of the tooth

Fig. 8 Panoramic radiograph of case 3. The altered trabeculae of the affected side, spacing between lateral and canine, and between the canine and the first deciduous molar are observed. The second deciduous molar, not visible in the clinical examination, is observed. There is alteration of the formation of the tooth bud (tooth germ) of the *right* premolars. The other quadrants have a normal eruption pattern

bud of the first premolar, absence of second maxillary premolar formation at the affected side, and the presence of the non-erupted second deciduous molar could also be observed. The root formation of the deciduous teeth was normal and the dental replacement in the non-affected sides was also without alterations (Fig. 8). The diagnosis of SOD and the need for a rigorous follow-up were explained to the patient and her parents.

Discussion

SOD is a rare, poorly recognized condition of unknown etiology that affects the maxillary bone [1]. The diagnosis is based on the clinical and radiographic features and it has particular histopathologic findings. The reported cases represent sporadic occurrence and the condition seems not to be inherited [1, 3]. Both genders can be equally affected and there is no predilection for any side. Unilateral maxillary enlargement, premolar absence, and decreased size of the maxillary sinus are the most common characteristics in the reported cases [1, 4, 5]. This condition is generally diagnosed in the first decade of life. However, it can be unrecognized or misdiagnosed and in some cases the diagnosis may be established in adulthood [1, 4, 5].

Fig. 7 Clinical pictures of case 3. **a** Extraoral view, showing the *right lip cleft*. **b** Intraoral photograph showing the right maxillary enlargement accompanied with hypodontia of the affected side and significant spacing between teeth of the affected side

Differential diagnosis includes monostotic fibrous dysplasia, regional odontodysplasia, gingival fibromatosis, segmental hemifacial hypertrophy, and benign odontogenic tumors [1, 3, 4, 6, 10, 11]. For this reason, in some cases, the histopathologic examination can be useful to establish the right diagnosis and exclude other possibilities [3]. In this paper, we present three additional cases of SOD with typical clinicopathologic, radiographic, and histologic features.

The negative, or noncontributory, past medical and family histories are reported in almost all the cases [1, 5, 10]. The presence of lip clefting in the affected side is a defect that has been associated with SOD [2, 4]; and the ipsilateral facial hypertrichosis is another characteristic reported in the majority of cases [1, 2, 5, 10]. Other alterations have been associated with this condition. Koenig et al. [2] reported palmar hyperlinearity with slight edema in the hands in a 26-year-old woman. Jones and Ford [12] and Welsch and Stein [7] reported the occurrence of SOD associated with Becker's nevus.

Some authors described facial skin erythema or lip hyper- or hypopigmentation in the affected side [1, 3, 10, 12, 13]. In the current patients, there were no alterations of pigmentation, but cases 1 and 3 showed association of a mild hypertrichosis and lip clefting. Yassin and Rihani [3] reported a case in a 12-year-old boy associated to hyperkeratotic diffuse skin erythema, hypopigmentation of the lip and loss of the vermillion border of the affected side. The same authors proposed an interesting clinical classification of SOD based on the presence and type of facial cutaneous manifestations. They recognized two major types: type I or classical with gingivo-dento-alveolar manifestations without extraoral facial involvement and type II or cutaneous with dermato-gingivo-dento-alveolar involvement. Type II was further subclassified into four subtypes: type IIa hypertrichotic, type IIb pigmentary, type IIc erythematous, and type IId commissural defect. Combinations between type II subtypes are possible; for example, type IIab with hypertrichotic-pigmentary combination or type IIabd with hypertrichotic-pigmentary-commissural defect combinations have been observed [3]. However, we think that the subclassification of type II is unnecessary and that a simple classification system can be useful to plan the treatment, establish a prognosis, and carry out epidemiological studies. Based on this system, our cases 1 and 3 are classified as type II, and case 2 as type I.

Prior to our cases, there have been 50 reported cases of SOD [1–21] (Table 1). These disorders are included in the same spectrum as SOD and HMD. Danforth et al. [6] proposed the name SOD because HMD is a term that implied a more extensive process and ignored the dental abnormalities. Welsch and Stein [7] proposed the acronym

HATS to represent the spectrum of these conditions when there are skin alterations. Our review indicated that SOD would be the better term, as this condition has well-defined clinical, radiologic and histopathologic features and the terms HMD and HATS could cause confusion. Porwal et al. [22] reported a case with the diagnosis of HMD, and discussed the spectrum of SOD/HMD/HATS. The clinical history and features and radiologic findings showed by these authors were consistent with the diagnosis of hemifacial microsomia or craniofacial microsomia, but not with SOD/HMD because one of the classical features of this condition, the unilateral maxillary enlargement, was not present.

All reported cases showed unilateral diffuse maxillary sclerosis with thickened trabeculae, as was seen in the current cases [4, 8, 10]. In the present cases, we observed on imaging, an altered tooth eruption of the affected side and, especially in case 1, a progressive diminution of the right maxillary sinus. The natural history of this disorder is not entirely clear, but some authors suggest a non-progressive nature for this condition [3, 5, 10]. Becktor et al. [10], analyzing panoramic radiographs of three patients with different ages, reported that the bony involvement in SOD seems not to be progressive. However, chronologic analysis of the X-rays of case 1 presented progressive changes in the bone, because the maxillary sinus was smaller and the bone was denser in the last radiographs than in the first one, suggesting that SOD has a progressive natural history. Involvement of the maxillary sinus of the affected side is noted in the majority of the reported cases. Packota et al. [4] and Prusack et al. [1] reported that SOD exhibits parallel, vertically arranged and coarse trabeculae in periapical radiographs, a feature recognized as a common finding of SOD.

Delayed eruption of permanent teeth is commonly reported, particularly of the first molar [1]. The abnormal shape of the roots was previously reported in primary teeth, but the absence or partial root formation seen in case 2 is not usually reported [1, 10]. Studies with CT analysis and conventional radiography showed similar changes in the affected bone [11], but CT obviously provides a better image of the adjacent craniofacial structures, such as the maxillary sinus and nasal cavity.

The histopathologic characteristics reported in these cases were similar to those described by other authors and included a nonspecific gingival fibrosis and absence of inflammation [1, 3, 6]. The involved bone is composed of large and irregular trabeculae of woven bone. The surrounding fibrous stroma is hypocellular and focally vascular [1, 3, 6]. It is probable that the alteration in the development of the bone and fibrous soft tissue in the area contributes to different development of the teeth, such as absence of the premolar teeth, abnormal morphology of the

Table 1 Summary of published cases

Author, year	No of cases	Gender	Age (years)	Affected side	Bone biopsy
Miles et al., 1987 [5]	2	1 M/1F	3–15	1R/1L	No
Danforth et al., 1990 [6]	8	4 M/4F	4–6–6–8–8–12–28	6R/2L	No
Packota et al., 1996 [4]	12	7 M/5F	5–6–9–11–14–16–27 (age not given for 5 patients)	4R/8L	No
DeSalvo et al., 1996 [14]	1	F	7	L	Yes
Paticoff et al., 1997 [13]	2	2 M	7–23	2R	No
Jones and Ford, 1999 [12]	1	M	7	L	No
Prusack et al., 2000 [1]	1	F	9	R	Yes
Velez et al., 2002 [15]	2	1 M/1F	3–14	1R/1L	No
Becktor et al., 2002 [10]	4	3 M/1F	2–3–3–8	4L	Yes
Drake, 2003 [16]	1	M	7	L	No
Armstrong et al., 2004 [9]	2	2 M	3–3	L	Yes
Welsch and Stein, 2004 [7]	1	M	5	L	No
Gavalda, 2004 [17]	1	M	8	R	Yes
Bhatia et al., 2008 [18]	1	M	7	L	No
Koenig et al., 2008 [2]	1	F	26	R	No
Yassin and Rihani, 2008 [3]	1	M	12	L	Yes
Özpınar et al., 2009 [19]	1	F	47	L	No
Kuklani and Nair, 2010 [11]	1	M	4	L	Yes
Whitt et al., 2011 [8]	5	3 M/2F	4–4–9–11–13	5R	Yes (1)/No (4)
Minett and Daley, 2011 [21]	1	M	5	R	1
Friedlander-Barenboim et al., 2011 [20]	1	M	12	R	Yes
Present cases	3	2 M/1F	5–17–22	3R	Yes (2)/No (1)

M male, *F* female, *R* right, *L* left

roots of the molars and abnormal eruption of the adjacent molars [1, 3, 10].

There are few reports about treatment in patients with SOD. Due to the mild facial asymmetry in SOD, recontouring osteotomy is not advised. In addition, there is no long-term follow-up of patients with this condition to confirm if SOD is a self-limited disorder. Treatment is only limited to orthodontic correction and/or prosthetic rehabilitation of edentulous areas [16]. In the last few years, success of the rehabilitation of these patients with implants showing normal healing and osseointegration has been reported [8, 10]. When an early diagnosis is made, as in case 3, the first objective is to maintain the deciduous teeth, facilitating the eruption of permanent teeth and occlusion, and definitive treatment is usually delayed until the pubertal growth spurt [11]. Cases 1 and 2 were diagnosed in more advanced ages, complicating the treatment and rehabilitation because of early loss of the deciduous teeth. A report on the long-term treatment and follow-up of patients with SOD is necessary to define a standard protocol of treatment and provide better management.

In summary, we reported three additional cases of SOD, in which the patients previously consulted other professionals without receiving the right diagnosis. Therefore, it is

important to recognize this disorder as a differential diagnosis of unilateral maxillary/teeth/skin alterations, in order to better manage these patients.

References

1. Prusack N, et al. Segmental odontomaxillary dysplasia: a case report and review of the literature. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod.* 2000;90(4):483–8.
2. Koenig LJ, Lynch DP, Yancey KB. Segmental odontomaxillary dysplasia presenting with facial hypertrichosis, commissural lip clefting, and hyperlinear palms. *Pediatr Dermatol.* 2008;25(4):491–2.
3. Yassin OM, Rihani FB. Combined cutaneous findings with segmental odontomaxillary dysplasia: review of the literature and proposal of a new clinical classification. *Int Med Case Rep J.* 2008;1:7–11.
4. Packota GV, Pharoah MJ, Petrikowski CG. Radiographic features of segmental odontomaxillary dysplasia: a study of 12 cases. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod.* 1996;82(5):577–84.
5. Miles DA, Lovas JL, Cohen MM Jr. Hemimaxillofacial dysplasia: a newly recognized disorder of facial asymmetry, hypertrichosis of the facial skin, unilateral enlargement of the maxilla, and hypoplastic teeth in two patients. *Oral Surg Oral Med Oral Pathol.* 1987;64(4):445–8.
6. Danforth RA, et al. Segmental odontomaxillary dysplasia. Report of eight cases and comparison with hemimaxillofacial dysplasia. *Oral Surg Oral Med Oral Pathol.* 1990;70(1):81–5.

7. Welsch MJ, Stein SL. A syndrome of hemimaxillary enlargement, asymmetry of the face, tooth abnormalities, and skin findings (HATS). *Pediatr Dermatol*. 2004;21(4):448–51.
8. Whitt JC, et al. Segmental odontomaxillary dysplasia: report of a series of 5 cases with long-term follow-up. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod*. 2011;112(2):e29–47.
9. Armstrong C, et al. Histopathology of the teeth in segmental odontomaxillary dysplasia: new findings. *J Oral Pathol Med*. 2004;33(4):246–8.
10. Becktor KB, et al. Segmental odontomaxillary dysplasia: clinical, radiological and histological aspects of four cases. *Oral Dis*. 2002;8(2):106–10.
11. Kuklani RM, Nair MK. Segmental odontomaxillary dysplasia: review of the literature and case report. *Int J Dent*. 2010; 837283.
12. Jones AC, Ford MJ. Simultaneous occurrence of segmental odontomaxillary dysplasia and Becker's nevus. *J Oral Maxillofac Surg*. 1999;57(10):1251–4.
13. Patricoff K, et al. Hemimaxillofacial dysplasia: a report of two new cases and further delineation of the disorder. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod*. 1997;83(4):484–8.
14. DeSalvo MS, et al. Segmental odontomaxillary dysplasia (hemimaxillofacial dysplasia): case report. *Pediatr Dent*. 1996; 18(2):154–6.
15. Velez I, et al. Segmental odontomaxillary dysplasia. Report of two cases and review of the literature. *Today's FDA*. 2002; 14(12):20–1.
16. Drake DL. Segmental odontomaxillary dysplasia: an unusual orthodontic challenge. *Am J Orthod Dentofac Orthop*. 2003; 123(1):84–6.
17. Gavalda C. Segmental odontomaxillary dysplasia. *Med Oral*. 2004;9(2):181.
18. Bhatia SK, et al. Case report: segmental odontomaxillary dysplasia—a rare disorder. *Eur Arch Paediatr Dent*. 2008;9(4):245–8.
19. Ozpinar B, et al. Prosthetic rehabilitation of segmental odontomaxillary dysplasia: seven-year follow-up. *Cleft Palate Craniofac J*. 2009;46(1):103–7.
20. Friedlander-Barenboim S, Kamburoglu K, Kaffe I. Clinical and radiological presentation of hemimaxillofacial dysplasia/segmental odontomaxillary dysplasia: Critical analysis and report of a case. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod*. 2011.
21. Minett, CP, Daley TD. Hemimaxillofacial Dysplasia (Segmental Odontomaxillary Dysplasia): Case Study With 11 Years of Follow-Up From Primary to Adult Dentition. *J Oral Maxillofac Surg*. 2011.
22. Porwal R, et al. Hemimaxillofacial dysplasia: a variable presentation. *J Craniofac Surg*. 2008;19(6):1554–7.