Segmental odontomaxillary dysplasia: report of a series of 5 cases with long-term follow-up

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We report a series of 5 cases of segmental odontomaxillary dysplasia (SOMD) with follow-up periods ranging from 8 to 21 years, bringing the total number of reported cases to 45. SOMD is a sporadic, mesoectodermal dysplasia that presents early in life, possibly as early as in utero and exhibits male gender predominance (1.7:1.0). Its features include enlargement of the soft tissue and/or bone of 1 hemimaxilla that may produce mild facial asymmetry. Subsequent growth of the affected area is proportional to that of the unaffected hemimaxilla. Sclerotic radiographic bone changes and dental developmental abnormalities are also present. The dense bone, which often exhibits a radiographic vertical orientation of the trabecular bone pattern, is typically associated with delayed eruption of the teeth. Congenitally missing premolar teeth (either or both) is a common feature of this condition that is of significant diagnostic value. Although ipsilateral cutaneous findings have been reported in 23%, our cases exhibited none. Computed tomographic imaging demonstrated extensive involvement of the maxillary bone, including the lateral wall and floor of the maxillary sinus and the hard palate. The affected bone presents no impediment to either orthodontic tooth movement or to the successful osteointegration of dental implants. The cause of SOMD is unknown; the clues to the cause of this unusual phenotypic expression are buried in the intricacies of developmental biology within the first branchial arch. (Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2011;112:e29-e47)

First described in 1987,1 segmental odontomaxillary dysplasia (SOMD) is a noninherited, sporadic, developmental, mesoectodermal dysplasia that presents early in life and is characterized by asymptomatic enlargement of 1 hemimaxilla that exhibits growth proportionate to the remainder of the maxillofacial complex. It may be accompanied by mild facial asymmetry secondary to the hemimaxillary enlargement.2-4 A number of variable features may be seen in the affected region, including increased spacing and delayed eruption of teeth, hypoplasia, malformation and abnormal resorption of primary molars, hypodontia involving the permanent premolar teeth, and fibrous hyperplasia of the overlying soft tissues.3 Radiographically, the bone of the affected region exhibits localized, ill-defined increased bone density owing to coarse, irregular trabeculae with a variable vertical orientation.3,5 Encroachment on the maxillary sinus may reduce its volume. The cause and pathogenesis of SOMD are unknown.

The original report of 2 cases was published as hemimaxillofacial dysplasia (HD).1 The authors of the second report of 8 cases suggested segmental odontomaxillary dysplasia as more descriptive.2 There have been 13 additional publications,3,4,6-16 bringing the total number of reported cases to 40. Table I summarizes the general clinical features of the 40 previously reported cases of SOMD and the 5 additional cases presented here. We excluded 2 reports from inclusion in our review. One report17 provided insufficient clinical, historical, and radiographic information to substantiate the diagnosis of SOMD; we were unable to exclude the possibility that this case represents fibrous dysplasia on the basis of the information provided. A second report18 was excluded because it fails to meet the established diagnostic criteria for SOMD.1,2

The prevalence of SOMD is unknown, as only single cases or series of cases have been reported. It is probably more common than the literature indicates, because it is likely that cases have been classified as fibrous dysplasia or atypical fibrous dysplasia.2 Several2,3,7 have cited an example of a case with features consistent with SOMD as representing a portion of the radiographic spectrum of fibrous dysplasia.19,20

The purpose of this article is to review the literature and present 5 additional cases of this rare developmen-
## Table 1. Demographic and general clinical features of previously reported cases of SOMD, including the present 5 cases, in reverse chronologic order of publication

<table>
<thead>
<tr>
<th>Author</th>
<th>Age in years at time of diagnosis</th>
<th>Gender</th>
<th>Affected side</th>
<th>Hypodontia</th>
<th>Ipsilateral cutaneous findings</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whitt et al. Case # 1</td>
<td>4</td>
<td>F</td>
<td>R</td>
<td>Both PM missing</td>
<td>None present</td>
<td>21 years of follow-up, orthodontic treatment, 2 dental implants osteointegrated with 8 years of follow-up orthodontic treatment</td>
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<td>Whitt et al. Case # 2</td>
<td>13</td>
<td>M</td>
<td>R</td>
<td>Both PM present</td>
<td>None present</td>
<td>11 years of follow-up, orthodontic treatment</td>
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<td>Whitt et al. Case # 3</td>
<td>11</td>
<td>F</td>
<td>R</td>
<td>1st PM missing</td>
<td>None present</td>
<td>8 years of follow-up, orthodontic treatment</td>
</tr>
<tr>
<td>Whitt et al. Case # 4</td>
<td>4</td>
<td>M</td>
<td>R</td>
<td>Both PM present</td>
<td>None present</td>
<td>15 years of follow-up, orthodontic treatment</td>
</tr>
<tr>
<td>Whitt et al. Case # 5</td>
<td>9</td>
<td>M</td>
<td>R</td>
<td>2nd PM missing</td>
<td>None present</td>
<td>10 years of follow-up, orthodontic treatment</td>
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<tr>
<td>Bhatia et al. (2008)8</td>
<td>3</td>
<td>M</td>
<td>L</td>
<td>Both PM missing</td>
<td>Ectopic eyelashes</td>
<td>4 years of follow-up, orthodontic treatment</td>
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<tr>
<td>Koenig et al. (2008)13</td>
<td>26</td>
<td>F</td>
<td>R</td>
<td>Both PM missing</td>
<td>Hypertrichosis, commissural lip cleft, hyperlinear palms</td>
<td>Vertical alveolar defect at distal of canine TE primary canine</td>
</tr>
<tr>
<td>Armstrong et al. (2004)6</td>
<td>3.5</td>
<td>M</td>
<td>L</td>
<td>1st PM missing</td>
<td>Not stated</td>
<td>7.5 years of follow-up, TE primary canine and primary 1st molar</td>
</tr>
<tr>
<td>Armstrong et al. (2004)6</td>
<td>3.5</td>
<td>M</td>
<td>L</td>
<td>2nd PM missing</td>
<td>Not stated</td>
<td></td>
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<td>Gavalda (2004)11</td>
<td>8</td>
<td>M</td>
<td>R</td>
<td>2nd PM missing</td>
<td>Not reported. Image exhibits no lesions</td>
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<tr>
<td>Welsch and Stein (2004)16</td>
<td>5</td>
<td>M</td>
<td>L</td>
<td>One unspecified PM missing</td>
<td>Becker’s nevus</td>
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<tr>
<td>Drake (2003)10</td>
<td>7</td>
<td>M</td>
<td>L</td>
<td>Both PM missing</td>
<td>None present</td>
<td>Dental implant, osteointegrated with short-term follow-up</td>
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<tr>
<td>Becktor et al. (2002)7 Case # 1</td>
<td>3</td>
<td>F</td>
<td>L</td>
<td>1st PM missing</td>
<td>Erythema</td>
<td>6 years of follow-up, TE primary molars</td>
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<tr>
<td>Becktor et al. (2002)7 Case # 2</td>
<td>8</td>
<td>M</td>
<td>L</td>
<td>2nd PM missing</td>
<td>None present</td>
<td>10 years of follow-up, TE permanent first molar</td>
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<tr>
<td>Becktor et al. (2002)7 Case # 3</td>
<td>2</td>
<td>M</td>
<td>L</td>
<td>Both PM missing</td>
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<tr>
<td>Becktor et al. (2002)7 Case # 4</td>
<td>3</td>
<td>M</td>
<td>L</td>
<td>1st PM missing</td>
<td>None present</td>
<td>3 years of follow-up</td>
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<tr>
<td>Velez et al. (2002)15 Case # 1</td>
<td>3.5</td>
<td>M</td>
<td>R</td>
<td>NR</td>
<td>Hyperpigmentation and hypertrichosis</td>
<td></td>
</tr>
<tr>
<td>Velez et al. (2002)15 Case # 2</td>
<td>14</td>
<td>F</td>
<td>L</td>
<td>Both PM present</td>
<td>None present</td>
<td></td>
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<tr>
<td>Prusack et al. (2000)4</td>
<td>9</td>
<td>F</td>
<td>R</td>
<td>Both PM missing, 3 M missing, 3 M missing,</td>
<td>None present</td>
<td>Vertical alveolar defect at distal of canine Special education, speech delays, mental illness, orthodontic treatment</td>
</tr>
<tr>
<td>Jones and Ford (1999)12</td>
<td>7</td>
<td>M</td>
<td>L</td>
<td>Both PM missing, 3 M missing, 3 M missing,</td>
<td>Becker’s nevus</td>
<td></td>
</tr>
<tr>
<td>Paticoff et al. (1997)14 Case # 1</td>
<td>23</td>
<td>M</td>
<td>R</td>
<td>2nd PM missing</td>
<td>Hairy nevus</td>
<td></td>
</tr>
<tr>
<td>Paticoff et al. (1997)14 Case # 2</td>
<td>7</td>
<td>M</td>
<td>R</td>
<td>1st PM missing</td>
<td>Hypertrichosis</td>
<td></td>
</tr>
<tr>
<td>DeSalvo et al. (1996)9 Case # 6</td>
<td>4</td>
<td>F</td>
<td>L</td>
<td>NR</td>
<td>Upper lip hypopigmentation</td>
<td></td>
</tr>
<tr>
<td>Packota et al. (1996)3 Case # 6</td>
<td>14</td>
<td>M</td>
<td>L</td>
<td>Both PM missing</td>
<td>Discontinuity of vermilion border with depression of cheek commissural cleft and bilateral commissural pits</td>
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</tbody>
</table>
tal disorder. Extended follow-up periods ranging from 8 to 21 years confirm the nonprogressive clinical behavior. Most reports of SOMD have appeared in the oral and maxillofacial surgery, pathology, and radiology literature, with only 3 reports in the pediatric dentistry literature\(^8,9,13\) and 1 in the orthodontic litera-

<table>
<thead>
<tr>
<th>Author</th>
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<th>Affected side</th>
<th>Hypodontia</th>
<th>Ipsilateral cutaneous findings</th>
<th>Comments</th>
</tr>
</thead>
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<tr>
<td>Packota et al. (1996)(^3) (11 remaining cases)</td>
<td>5, 6, 9, 11, 16, 27 (NR for 5 patients)</td>
<td>6 M, 5 F</td>
<td>L7, R4</td>
<td>9 cases missing both PMs, 2 cases missing 1 PM</td>
<td>Not reported</td>
<td></td>
</tr>
<tr>
<td>Danforth et al. (1990)(^2) Case # 1</td>
<td>6</td>
<td>M</td>
<td>R</td>
<td>2nd PM missing</td>
<td>None present</td>
<td></td>
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<tr>
<td>Danforth et al. (1990)(^2) Case # 2</td>
<td>4</td>
<td>F</td>
<td>R</td>
<td>Both PM missing</td>
<td>None present</td>
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<tr>
<td>Danforth et al. (1990)(^2) Case # 3</td>
<td>8</td>
<td>M</td>
<td>L</td>
<td>Both PM missing, primary canine missing</td>
<td>None present</td>
<td></td>
</tr>
<tr>
<td>Danforth et al. (1990)(^2) Case # 4</td>
<td>6</td>
<td>F</td>
<td>R</td>
<td>Both PM missing</td>
<td>Not stated</td>
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<td>Danforth et al. (1990)(^2) Case # 5</td>
<td>12</td>
<td>M</td>
<td>R</td>
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<td>Not stated</td>
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<td>Danforth et al. (1990)(^2) Case # 6</td>
<td>8</td>
<td>M</td>
<td>R</td>
<td>2nd PM missing</td>
<td>Not stated</td>
<td></td>
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<tr>
<td>Danforth et al. (1990)(^2) Case # 7</td>
<td>6</td>
<td>F</td>
<td>L</td>
<td>1st PM missing</td>
<td>Not stated</td>
<td></td>
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<tr>
<td>Danforth et al. (1990)(^2) Case # 8</td>
<td>28</td>
<td>F</td>
<td>R</td>
<td>Both PM missing</td>
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<tr>
<td>Miles et al. (1987)(^1) Case # 1</td>
<td>15</td>
<td>M</td>
<td>R</td>
<td>Both PM missing</td>
<td>Hypertrichosis</td>
<td></td>
</tr>
<tr>
<td>Miles et al. (1987)(^1) Case # 2</td>
<td>3.5</td>
<td>F</td>
<td>L</td>
<td>Both PM present</td>
<td>Not stated</td>
<td></td>
</tr>
</tbody>
</table>

F, female; L, left side; M, male; PM, premolar; R, right side; TE, tooth extraction; NR, not reported.

Fig. 1. Case 1. Clinical images depicting the patient’s appearance at age 4 years. The lesion involved the right maxilla and produced no significant facial asymmetry beyond a mild fullness of the right upper lip. The increased buccolingual dimension of the maxillary alveolus extended from the mesial of the primary canine to the tuberosity. The occlusal surfaces of the primary molar teeth were hypoplastic and there was spacing between the primary teeth (the intraoral image was exposed in a mirror reflection).
ture.10 Wider recognition and reporting of this condition will aid in elucidating its prevalence and manifestations.

CASE REPORT
Case 1
This 4-year-old girl was seen in 1986, 1 year before the initial recognition of this condition as a specific entity.1 She exhibited unilateral enlargement of the right maxilla that produced mild asymmetry of the right upper lip, characterized by a taut appearance of the overlying vermillion, with more fullness compared with the contralateral side. Intraorally, the right maxilla exhibited enlargement extending from the mesial of the primary canine to the tuberosity, accompanied by increased spacing between the primary teeth (Fig. 1). The occlusal surfaces of both primary molars exhibited hypoplasia.

The lesion was investigated with multiple imaging methods, including periapical radiographs, panoramic radiographs, and computed tomographic (CT) scanning. Although these

Fig. 2. Case 1. Clinical images at age 14. The lesion exhibited a moderate increase in buccolingual dimension that was proportionate to overall growth and extended further anteriorly to involve the incisor region. It caused no significant facial asymmetry. Both permanent premolar teeth were missing and the permanent canine tooth was positioned distally.

Fig. 3. Case 1. The patient’s appearance at age 15 years. The enlarged area extended from the tuberosity to the canine-incisor region. The permanent premolars were missing and there is retention of the second primary molar.

Fig. 4. Case 1. Clinical image depicting the appearance of patient’s dentition at age 15 years. The buccal aspect of the right maxilla is enlarged in comparison with the uninvolved side. There is a sessile, white papule, accompanied by an erythematous, blanchable area of increased vascularity on gingival mucosa of the affected side.
materials could no longer be located for inclusion into this article, the involved area was described as exhibiting an ill-defined density of the right maxilla. Bone scintigraphy was negative. The patient’s medical history was noncontributory and there were no associated cutaneous findings. The parents, as well as 2 siblings, were unaffected, although her father exhibited congenitally missing mandibular second premolars. Based on the clinical appearance, behavior (asymptomatic lesion with slow enlargement proportional to overall growth), and imaging findings, it was concluded that this lesion represented a developmental process, possibly similar to fibrous dysplasia. It was decided to follow the patient closely. A biopsy of the lesion was not performed as it was believed that it was a non-neoplastic lesion with low potential for significant morbidity.

Over the next 10 years, the lesion exhibited moderate enlargement that was proportionate to overall growth. By the early teenage years, her maxillary enlargement was slightly more prominent and the expanded buccolingual dimension of the maxilla had advanced anteriorly beyond the canine to involve the incisor region (Figs. 2–4). The mild fullness of the right upper lip remained and there was no remarkable facial asymmetry. Both permanent premolar teeth were missing, the secondary primary molar was retained, and the permanent canine tooth was incompletely erupted. A panoramic radiograph, exposed at the age of 15 years exhibited an ill-defined density involving the area of the missing premolar teeth on the affected side (Fig. 5). The permanent second molar was unerupted and was extracted after unsuccessful attempts to facilitate its eruption; the permanent third molar was subsequently moved into its position.

Over the next 10 years, the lesion exhibited little change. Orthodontic treatment was initiated at age 15. At the age of 17 years, 2 dental implants were placed in the area of dense bone to replace the missing premolar teeth. A panoramic radiograph exposed at age 25 years exhibited a circumscribed area of bone density involving the maxillary sinus floor that reduced its size relative to the uninvolved side (Fig. 6). On periapical radiographs, the bony trabeculae of the involved right maxilla appeared coarse and vertically oriented, compared with the normal trabecular bone pattern of the uninvolved left maxilla (Fig. 7).

The patient described a number of small, white, sessile papules involving the facial gingival mucosa of the affected region that had been incised in the past. One such lesion involving the buccal gingiva, clinically resembling a dental lamina inclusion cyst of the newborn, was observed in a clinical image exposed at the age of 14 years (Fig. 4) and was no longer present at clinical follow-up at age 25 years (see Discussion section). There was a circumscribed, erythematous, blanchable zone of increased vascularity associated with this lesion that persisted through the most recent follow-up at age 25 years.

During the 21-year follow-up period, from ages 4 years through 25 years, enlargement of the lesion was proportionate to the growth of the uninvolved side. The reduced buccolingual dimension of the affected area (Fig. 8) is the result of surgical reduction performed to establish physiological tissue contours. This was accomplished about the time of orthodontic therapy.

Case 2
This 13-year-old boy presented with delayed eruption of the teeth of the right maxilla, accompanied by enlargement that extended from the tuberosity to the incisor area, increased spacing between the teeth, and hypoplasia of the second primary molar (Fig. 9). His chief complaint was fullness of the right upper lip secondary to the maxillary enlargement (Fig. 10). The patient’s medical history was noncontributory. A sibling exhibited normal oral findings. There were no associated cutaneous findings and biopsy of the lesion was not performed.

A panoramic radiograph at age 14 years exhibited a radiodensity of the right posterior maxilla that involved the floor of the maxillary sinus, reducing its size in relation to the uninvolved left side (Fig. 11). Both premolars in the affected hemimaxilla were present. The second premolar erupted without assistance, but the first premolar required surgical exposure to assist eruption and, when erupted, exhibited a gray discoloration and was determined to be nonvital (Fig. 12).
possibly as a result of the assisted eruption procedure performed between the ages of 14 and 18 years.

In a panoramic radiograph exposed at the age of 18 years (Fig. 13), the vertically oriented appearance of the involved trabecular bone could be seen clearly and the lesion exhibited a sharply demarcated anterior margin that was also apparent in periapical (Fig. 14) and oblique radiographs (Fig. 15) exposed at the age of 24 years.

Orthodontic treatment was complicated by the soft tissue enlargement that interfered with bracket placement and required gingivoplasty to provide access for orthodontic appliances. Closure of the spacing between the teeth of the involved hemimaxilla was not completed because treatment was discontinued when the patient entered military service.

A photograph taken at age 18 years exhibited a number of small, white, sessile papules, clinically resembling dental lamina inclusion cysts of the newborn, involving the facial gingival mucosa of the affected region. These lesions regressed and were not observed at clinical follow-up at age 24 years (see Discussion section).

The photograph of the maxillary arch taken at age 24 years (Fig. 12), gives the impression that the enlarged contour of the maxillary anterior alveolus has regressed; however, this appearance is the result of surgical reduction of the area that was performed between the ages of 14 and 18 years to address the patient’s chief complaint of fullness of the upper lip. The permanent second premolar partially erupted without assistance, although chronologically late.

Over the 11-year period of follow-up, between the ages of 13 years and 24 years, the lesion did not progress clinically beyond enlargement that was proportional to the growth of the uninvolved side and it exhibited no detectable change in radiographic appearance.

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Fig. 6. Case 1. Panoramic radiograph at 25 years. On the patient’s right side in the area of the dental implants, there was circumscribed area of bone density involving the maxillary sinus floor that reduced its size relative to the uninvolved side. The unerupted second permanent molar was extracted and the third molar was moved into its position.

Fig. 7. Case 1. Periapical radiographs at age 25 years. The premolar implants were placed at age 17 and underwent uncomplicated osteointegration. The bony trabeculae of the involved right maxilla appeared coarse and vertically oriented, compared with the normal trabecular bone pattern of the uninvolved left maxilla. The size of the right maxillary sinus was reduced in comparison with the uninvolved side. The anterior margin of the lesion appeared sharply defined.
Case 3

This 11-year-old girl presented with unilateral expansion of the right hemimaxilla that produced mild asymmetry of the right upper lip and cheek. The enlarged area extended from the tuberosity to the incisor area (Fig. 16) and was accompanied by delayed eruption of teeth on the affected side without an increase in space between the teeth. The first premolar on the affected side was missing and the first primary molar exhibited hypoplasia. Radiographically, the affected area exhibited an ill-defined radiodensity with a coarse, granular trabecular pattern (Fig. 17).

At the age of 19 years, a panoramic radiograph exhibited a radiodensity with a coarse, granular pattern and a well-defined anterior margin in the canine-premolar area that had reduced the size of the maxillary sinus in comparison with the uninvolved side (Fig. 18). Periapical radiographs exhibited coarse, granular bone trabeculae with a vertical orientation (Fig. 19). The remaining first premolars in the uninvolved quadrants were extracted to gain space to facilitate orthodontic treatment, during which it was noted that the teeth on the affected side moved more slowly because of the dense bone.

During the 8-year period of clinical follow-up between the ages of 11 years and 19 years, the lesion was stable and did not exhibit enlargement that was disproportionate to the growth of the uninvolved side. At the age of 19 years, the enlargement of the maxillary alveolus appeared to have regressed somewhat (Fig. 20); however, this appearance is the result of surgical reduction of the area that was performed at the age of 15 years to address the patient’s concerns of fullness of the midface on the affected side. Microscopic examination of the excised mucosa revealed fibrous and fibromyxoid hyperplasia, containing thick collagen bundles commingled with zones of hematoxylinophilic myxoid ground substance (Fig. 21). The patient’s medical history was noncontributory and there were no associated cutaneous findings. A bone biopsy was not performed.

Case 4

This 4-year-old boy presented with increased buccolingual dimension of the right posterior maxilla extending from the tuberosity to the mesial of the primary canine. This was accompanied by delayed eruption of the primary first molar and increased spacing between the teeth of the affected side (Fig. 22). Panoramic radiographs exposed at the ages of 4 and 5 years (not shown) exhibited an ill-defined radiodensity with a coarse trabecular pattern involving the premolar area of the right maxilla. This was accompanied by marked hypoplasia of the unerupted primary first molar. CT imaging of the patient’s maxilla performed at the age of 6 years exhibited a moderately increased buccolingual dimension of the alveolar pro-
cess of the right posterior maxilla. There was a coarse, granular pattern of the trabecular bone in the expanded area and prominent thickening of the anterior and lateral walls of the maxillary sinus that reduced its size in comparison with the unaffected side (Fig. 23).

An incisional biopsy of alveolar bone at age 4 exhibited coarse, thickened trabeculae composed of both woven and mature lamellar bone with a pagetoid appearance (Fig. 24). There was no evidence of appositional osteoblastic activity or of active osteoclastic resorption. The marrow spaces contained predominately fatty marrow. At the periosteal surface, the cortex was irregular and ill defined and transitioned abruptly to cellular fibrous connective tissue (Fig. 25). The patient’s medical history was noncontributory; there were no associated cutaneous findings and no clinically significant facial asymmetry.

A panoramic radiograph taken at age 17 exhibited an ill-defined density of the right maxilla that reduced the size of the right maxillary sinus in comparison with the uninvolved side (Fig. 26). Management of unerupted teeth was a particular challenge in this case. The first permanent molar had not fully erupted by the age of 8 years and required 2 years of active intervention, including surgical exposure and elastic traction to bring it into occlusion. The permanent canine tooth on the affected side was impacted and also required surgical exposure and traction to bring it into occlusion. Both premolars in the affected hemimaxilla were present and erupted without active intervention, although both were delayed chronologically. The first premolar crown was markedly hypoplastic. We were unable to reconstruct the sequence of events to determine whether this represented developmental hypoplasia or environmental hypoplasia, secondary to pulpal...
disease involving the overlying hypoplastic primary molar (Turner tooth). Both maxillary first premolars were extracted as part of the orthodontic treatment plan. The permanent second molar erupted late and did not require intervention to bring it into occlusion.

During the 15-year interval of follow-up, between the ages of 4 years and 19 years, the lesion exhibited clinical enlargement that remained in proportion to the growth of the uninvolved hemimaxilla.

Case 5

This 9-year-old boy presented with delayed eruption of teeth and increased buccolingual dimension of the right posterior maxilla (Fig. 27). The right hemimaxilla exhibited an increased contour of the facial surface that was more prominent compared with the uninvolved side (Fig. 28). A panoramic radiograph exhibited an ill-defined radiodensity involving the right maxilla, associated with a missing second premolar in the affected region (Fig. 28). A periapical radiograph of the enlarged posterior maxilla exhibited diffuse density of the alveolar bone accompanied by a coarse, vertically oriented trabecular pattern and irregular root resorption of the second primary molar (Fig. 29). There was no increased spacing between the teeth of the involved hemimaxilla. A CT scan performed at the age of 13 years exhibited extensive involvement of the maxillary bone with thickening of the lateral wall and floor of the maxillary sinus,
reducing its volume compared with the uninvolved side. The lesion also involved the hard palate (Fig. 30).

Surgical reduction of soft tissue in the involved area was required to place orthodontic appliances. The retained second primary molar was extracted and the space was adjusted for the placement of a dental implant to replace the missing permanent second premolar.

During the 10-year follow-up period, from ages 9 years to 19 years, the lesion exhibited mild enlargement that was proportional to the growth of the uninvolved hemimaxilla. A biopsy was not performed. There was no clinically significant facial asymmetry, no associated cutaneous findings, and the patient’s medical history was noncontributory.

REVIEW OF THE LITERATURE

The general clinical features of the 40 previously reported cases of SOMD and the additional 5 cases described here are summarized in Table I with comments on treatment performed. The mean age at diagnosis was 9 years with a range of 2 years to 28 years. A number of reports disclosed that maxillary enlargement had been recognized earlier than the age at which the diagnosis was established and was observed at birth in 4 instances.1,2,14 There was a male gender predilection with a male-to-female ratio of 1.7 to 1.0. Race was stated for only 70% of the reports and recorded as Caucasian1-4,8-10,13 for 63% (25 cases), including all of our cases, and as African American15,16 for 8% (3 cases).

There was no predominance for location in one hemimaxilla over the other, with essentially equal distribution between the left (23 cases) and the right (22 cases). Facial asymmetry was reported in 42%.1,2,6,8,9,11,13-16 and was frequently described as appearing as a prominence of the
upper lip. Absence of facial asymmetry was reported in 12%,\textsuperscript{10,12,15} including 2 of our cases and was not recorded for the remainder of the cases. Ipsilateral cutaneous findings over the affected area were associated with 28%\textsuperscript{3,7-9,12-16} and included hypertrichosis, hyperpigmentation, hypopigmentation, ectopic eyelashes, commissural clefting, hairy nevus and Becker nevus. The absence of cutaneous findings was reported in 33%, including all of our cases. The remainder of the authors did not comment on this feature. Both hyperlinear palms and commissural clefting were associated with 1 case.\textsuperscript{13} Two reports\textsuperscript{12,13} described a marked lucency distal to the canine that was suspicious for a bony cleft radiographically.

The radiographic appearance of the margin of the affected bone, when commented on, was described as poorly defined. Adjectives used to describe the radiographic appearance of the altered pattern of the cancellous bone of the affected region included coarse, sclerotic, thickened, granular, irregular, mottled, and vertically oriented trabeculae. Vertically oriented trabeculae were described in 37%\textsuperscript{3,4,12,15} including 3 of our cases, where this pattern was observed in both periapical and CT images for

Fig. 17. Case 3. Panoramic radiograph at age 11 years that exhibited delayed tooth eruption in the right posterior maxilla, accompanied by an ill-defined radiodensity with a coarse appearance of the trabecular bone pattern. The first premolar tooth was missing, the first primary molar exhibited root resorption, and there was no increased spacing of teeth. The opaque, white pyramidal area obliterating a portion of the anterior mandible is an artifact produced by a lead-lined thyroid shield.

Fig. 18. Case 3. Panoramic radiograph at age 19 years. The remaining 3 first premolars in the unaffected quadrants were extracted as part of the orthodontic treatment plan. The coarse, granular appearance of the trabecular bone in the affected quadrant exhibits a well-defined anterior margin and the right maxillary sinus is reduced in size in comparison with the left.
1 case. We observed a well-defined, distinct, anterior radiographic margin of the bony lesion at the canine-premolar region in 3 of our cases, as others have observed. A radiographic reduction in the size of the maxillary sinus was reported in 49%, including all of our cases. An absence of radiographic encroachment on the maxillary sinus was specifically reported by 5%, and the remainder of the authors did not comment on this issue.

Approximately one-half of the reported cases included a biopsy as part of the diagnostic workup. The enlarged gingival mucosa exhibited nonspecific changes, consisting of collagenous fibrous connective tissue (Fig. 21), with variable areas of myxoid change, reminiscent of the appearance of perifollicular connective tissue as others have observed. The histopathology of the affected bone was also nonspecific and generally consisted of thickened, irregularly shaped trabeculae of immature woven bone with numerous resting and reversal lines. There was a lack of osteoblastic rimming and, in most instances, no evidence of ongoing osteoclastic resorption, with the exception of focal areas of active resorption and osteoblastic rimming in a 2-year-old patient. The marrow spaces were variably described as containing loose, paucicellular, fibrous tissue, containing only a few fat cells. One report described the marrow as hypervascular.
Some combination of missing premolars in the affected area was a common finding reported in 91% (Table I), and involved the absence of both premolars in 53% and of 1 premolar in 40%. When 1 premolar was absent, the missing premolar was distributed essentially equally between the first premolar (6 cases) and the second premolar (8 cases). The identity of the missing premolar was unspecified in 3 cases. In only...
9% were both premolars present, including 2 of our cases; the status of the complement of premolars was not reported in 2 cases.

Delayed eruption of teeth in the involved segment was observed in 70%,2,3,6-12,14,15 including all of our cases. The remainder did not comment on this issue. Root resorption of primary molars was reported in 42%,2,3,7,12,13 including 2 of our cases and was specifically reported as not being present in 21%,2,3,7 including 1 of our cases. The remainder of the reports made no comment on this finding. Dysplasia of primary molar crowns was reported as present in 51%,1-3,6-11,13-15 including 4 of our cases, and not present in 12%,2 including 1 of our cases. The remainder of the reports did not comment on this feature. A wide range of additional morphologic anomalies of the primary teeth in the affected area were described by Packota et al.3 in a radiographic series of 12 cases. An additional report examined the histopathology of the deciduous teeth from the affected area and found that most of the dentin was histologically normal with the exception of localized areas of dysplastic dentin.6

Fig. 24. Case 4. Photomicrograph of an incisional biopsy of alveolar bone from the involved area obtained at the age of 4 years that exhibited thickened trabeculae containing both woven (W) and mature lamellar bone (L) with numerous reversal lines. There was no evidence of appositional osteoblastic activity or of active osteoclastic resorption. The marrow spaces contained predominately fatty marrow (hematoxylin and eosin stain, original magnification ×200).

Fig. 25. Case 4. Photomicrograph of an incisional biopsy of alveolar bone at the periosteal surface of the affected area (inferior portion of image), obtained at the age of 4 years, that exhibited an irregular and ill-defined cortical zone that transitioned abruptly to cellular fibrous connective tissue (hematoxylin and eosin stain, original magnification ×200).

9% were both premolars present, including 2 of our cases; the status of the complement of premolars was not reported in 2 cases.

Delayed eruption of teeth in the involved segment was observed in 70%,2,3,6-12,14,15 including all of our cases. The remainder did not comment. Increased spacing between teeth in the involved segment was a common finding reported in 76%,1,3,6-8,10-12,15,16 including 3 of our cases. No increased spacing was specifically reported in 12%,2 including 2 of our cases. The remainder did not comment on the issue. Root resorption of primary molars was reported in 42%,2,3,7,12,13 including 2 of our cases and was specifically reported as not being present in 21%,2,3,7 including 1 of our cases. The remainder of the reports made no comment on this finding. Dysplasia of primary molar crowns was reported as present in 51%,1-3,6-11,13-15 including 4 of our cases, and not present in 12%,2 including 1 of our cases. The remainder of the reports did not comment on this feature. A wide range of additional morphologic anomalies of the primary teeth in the affected area were described by Packota et al.3 in a radiographic series of 12 cases. An additional report examined the histopathology of the deciduous teeth from the affected area and found that most of the dentin was histologically normal with the exception of localized areas of dysplastic dentin.6

Long-term clinical follow-up, ranging from 2 to 21 years was recorded for 10 cases,2,6-8 including all of our cases. For cases with follow-up, progressive enlargement that was proportionate to overall growth was reported in 27%,2,4,8,9,12 including all of our cases and was described as slight by those who commented on the degree. In 30%,1,2,7,14,15 the authors specifically stated that they observed no enlargement and the remainder made no comment. During the follow-up period, the altered trabecular bone pattern remained stable, as we were able to observe in 2 of our cases and as others have reported.2

DISCUSSION

The cause of SOMD is unknown. The preponderance of evidence suggests that it is a noninherited developmental disorder, as cases appear sporadically. Reports describing jaw enlargement at birth1,2,4,14 imply that the process may begin as early as in utero. Consistent involvement of a unilateral maxillary segment2,4 and hypodontia involving premolar teeth3,4,14 suggest that the process involves a field defect involving the anlagen of the first branchial arch.14 Somatic mutation occurring in a specific progenitor cell influencing the development of the involved hemimaxilla3,6 may account for the mesoectodermal changes4 in a manner similar to that described for fibrous dysplasia.21,22 Why such an effect would be limited to only 1 side of the maxilla is a mystery.

Long-term follow-up indicates that SOMD is a nonprogressive condition that persists and although it may exhibit progressive enlargement, the increase in size is proportionate to the overall growth of the individual; the affected segment does not grow out of proportion to unaffected areas.2,16 This behavior is in distinct contrast with fibrous dysplasia in which the enlargement of the affected bone exhibits disproportionate growth. The
negative findings reported on bone scintigraphy\textsuperscript{7} and in one of our cases support the concept of a lesion with low metabolic activity and low growth potential. Although SOMD exhibits definite male gender predominance (1.7:1.0), there is insufficient evidence to draw conclusions regarding possible racial predilection.

The differential diagnosis has been extensively discussed by others\textsuperscript{2,4,6,9,14,23} and is limited to entities that may cause unilateral enlargement of 1 hemimaxilla, including gingival fibromatosis, segmental hemifacial hyperplasia, regional odontodysplasia, craniofacial fibrous dysplasia, and neurofibromatosis. Clinical, radiographic and when required, histopathologic findings help to narrow the differential diagnosis. We believe that, in most cases, the clinical and radiographic appearance of SOMD is sufficiently distinctive to establish the diagnosis on those grounds alone. Biopsy of the involved bone is not essential for diagnosis. In unusual

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**Fig. 26.** Case 4. Panoramic radiograph at age 17 years. There was delayed eruption of teeth on the affected side (second molar, in this image) and an ill-defined density of the right maxilla that reduced the size of the involved maxillary sinus. The first permanent molar required active intervention over 2 years to achieve eruption, whereas the second molar eventually erupted without intervention, although chronologically late. The permanent canine tooth on the affected side was impacted and required surgical exposure and traction to bring it into occlusion. Both premolars were present and both erupted chronologically late without assistance. The first premolar exhibited marked enamel hypoplasia. Both maxillary first premolars were extracted as part of the orthodontic treatment plan.

**Fig. 27.** Case 5. The patient’s facial appearance and maxillary arch (exposed in a mirror reflection) at age 9 years. The lesion involved the right hemimaxilla, produced very mild facial asymmetry, and was accompanied by delayed eruption of the teeth on the affected side. The right hemimaxilla exhibited a moderate increase in buccolingual dimension extending from the tuberosity to the canine region that is partly obscured by the cheek in this image and is better appreciated in the following figure.
presentations, bone biopsy may be helpful, particularly to exclude other lesions, such as fibrous dysplasia. In questionable situations, soft tissue biopsy may also be helpful in narrowing the differential diagnosis, for example, by eliminating neurofibromatosis from consideration.

In discussing the cause of the clinical enlargement, Danforth et al. observed that in some cases the maxillary enlargement could be caused by excessive gingiva rather than excessive bone, an observation that was confirmed by Packota et al., who concluded that there was no radiographic evidence of bone enlargement in 6 of their 12 cases. In reports in which the authors attempted to determine the source the maxillary enlargement, they concluded that in 35% it was caused by a combination of both bone and soft tissue. The data required to assess the proportion of cases in which the enlargement is caused by bone and/or soft tissue are incomplete, as the remainder of the reports contain only partial information. For our cases, we were able to demonstrate both hard and soft tissue enlargement in Case 5, hard tissue enlargement in Case 4, and soft tissue enlargement in Cases 1, 2, and 3. We were unable to draw further conclusions because of the incomplete and fragmentary nature of the records available. Regardless of the source of the enlargement, the radiographic alterations appear to be present in all patients. The vertical orientation of coarse trabecular bone in the affected region is a distinctive feature reported in close to 40% of cases. We fancifully likened it to the appearance of falling water in a waterfall.

In contrast to other reports that described the involved bony trabeculae as exhibiting immature woven bone, the histopathology of our Case 4, a biopsy from a 4-year-old, exhibited a combination of both lamellar and woven trabecular bone (Figs. 24 and 25). Apparently, bone biopsies of other cases have exhibited some degree of mature lamellar bone formation, as others have alluded to it by stating that mature lamellar bone was usually not present. The marrow spaces of the biopsy from our Case 4 exhibited unremarkable fatty marrow, in contrast to fibrous marrow or a fibrous connective tissue stroma surrounding the trabeculae of affected bone as reported by others. Histologically, we, and others, observed a lack of a well-defined bony cortex and an irregular transition from pagetoid bony trabeculae to the cellular fibrous connective tissue that composed the soft tissue portion of the enlargement (Fig. 25). This correlates well with operative reports that described the lack of typical buccal cortical plate and described that the fibrous tissue appeared to grow into the bone and blend with it. This finding that has also been correlated with the ill-defined nature of the cortex observed on CT imaging.

As we noted in our review of the literature section, the lesional bone was reported to have encroached upon the maxillary sinus in close to half of the reported...
We suspect that maxillary sinus involvement has been underreported, as this determination is highly dependent on the type and quality of the imaging studies performed. CT imaging of a limited number of cases\(^4,11\) and 2 of our cases exhibit extension of the affected bone beyond the alveolar process to involve the lateral wall and floor of the maxillary sinus. The lesional bone may extend superiorly to the zygoma and orbital floor to surround the infraorbital canal.\(^4\) Although others have reported that the horizontal palatal shelf does not appear to be involved,\(^2,4,11\) our Case 5 does exhibit involvement of the horizontal palatal shelf. Interestingly, 3 of our cases exhibited a distinct, well-defined anterior radiographic margin on plain films. We suspect that the ability to observe this feature may be dependent on the type and quality of the imaging studies performed.

Hypodontia involving the premolars is a variable feature in SOMD. The approximately equal distribution between the first and second premolars when 1 premolar is missing is quite unusual because, in the general population, the second premolar is much more commonly missing than the first premolar. Indeed, the finding of congenitally missing first premolars is so unusual that finding alone should prompt closer scrutiny of the patient. Excessive tooth spacing in the involved segment is a frequent, but variable, finding and is likely related not only to enlargement of the alveolar process\(^3\) but also to more mundane causes, such as tooth and arch size discrepancies. Indeed, some patients exhibited no increased spacing,\(^2\) and even crowding, as seen in one of our cases.

Two of our cases exhibited a variable number of small, white, sessile papules, resembling dental lamina cysts of the newborn, involving the facial gingival mucosa at ages 14 and 18 years (Fig. 31) that regressed by ages 25 and 24 years, respectively. One patient reported that one of these lesions had been “incised” and that no tissue had been submitted for histopathologic examination.

The acronym HATS (Hemimaxillofacial enlargement, Asymmetry of the face, Tooth abnormalities, and Skin findings) has been proposed for this condition.\(^16\) Although this is a handy mnemonic device that emphasizes the occurrence of cutaneous findings (23%), it may overemphasize both the degree of the asymmetry that is clinically observed in affected individuals and possibly the extent of bone involvement.

Patients with SOMD present a wide variety of management considerations, including concerns for possible progressive facial deformity and hereditary transmission, management of delayed eruption, enlargement of soft tissue, excessive spacing, missing teeth, and supraeruption of opposing teeth.\(^10\) Reports of treatment and outcomes for patients with SOMD are limited. Soft tissue and/or bone removal to facilitate tooth eruption has been required,\(^9,10\) as well as in 2 of our cases. In addition to the successful osteointegration of dental implants in the affected bone of one of our cases (Figs. 3 and 4), there is one additional report of successful dental implant placement in the affected bone.\(^7\) Surgical recontouring of the enlarged area was performed in all of our cases for a variety of reasons, including providing access for placement of orthodontic appliances, to improve access for personal oral hygiene, and to address patients’ cosmetic concerns with respect to lip and cheek enlargement. In one of our cases, orthodontic tooth movement through the affected bone was described being slower that in the unaffected quadrants.
With respect to the pathogenesis of SOMD, at some point in development, there is a convergence of the developmental pathways of gnathogenesis and odonto-genesis that result in the phenotype of SOMD. The constellation of findings suggest that mosaicism may account for this expression. The clues to the cause of the unusual phenotypic expression are buried in the intricacies of the developmental biology of the first branchial arch.

The authors would like to express their appreciation to Harry W. Mueller for his professional bibliographic support services, without which our progress would have been greatly delayed. We also wish to thank Drs Kent Willett, Scott Robinson, John Gardner, and Francis Otradovec of Columbia, MO; Jeff Thompson, Mark Jensen, and Trisha Canzonere of Overland Park, KS; Frank Christ and Shara Dunlap of Lee’s Summit, MO; Woody Soonattrakul of Kennett, MO; Ricardo Gapski and Kelly Casement of Denver, CO; Don Weber, Joseph Harvey, David Jones, and Mike McBride of Lawrence, KS; Kurt Hoffman of Leawood, KS; Randolph Oliver and Sid McKnight of Prairie Village, KS; and Jane Cummings of Kansas City, MO for their cooperation and generous contribution of case material. We would also like to express our appreciation to Drs Brenda Bohatv and Jerry Katz of the University of Missouri Kansas City School of Dentistry for their consultative services, and to Jim Thomas and Alicia Wright of the University of Missouri Kansas City School of Dentistry Biomedical Communications Center for their photographic and graphics expertise.

REFERENCES


Fig. 31. Clinical images of case 1 (left) and case 2 (right), at ages 14 and 18 years, respectively. The gingivae of the affected areas exhibited variable numbers of small, white, sessile papules that later regressed. They were not observed at clinical follow-up at ages 25 and 24 years, respectively.


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