Case Report

Segmental Odontomaxillary Dysplasia in a 5-Year-Old Girl: A Rare Developmental Anomaly with Clinical and Radiographic Correlation

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ABSTRACT

Segmental odontomaxillary dysplasia is a rare developmental anomaly of the maxilla, typically diagnosed in early childhood. It is characterized by unilateral buccolingual expansion of the posterior alveolar process, gingival enlargement, and delayed eruption of adjacent teeth, and dental anomalies such as malformed primary molars and absence of premolars in the affected region. Several cases exhibit homolateral cutaneous manifestations, including facial scars or hyperpigmented patches. Radiographically, the affected bone shows thickened and irregular, with a coarse, vertically oriented trabecular pattern, producing a radiopaque granular appearance. A brief systematic review of previously reported cases was conducted to analyze demographic characteristics, clinical presentation, radiographic features, and management strategies. Based on the literature and current findings, a diagnostic algorithm was developed to facilitate differentiation from similar unilateral maxillary conditions. Herein, we report the case of a 5-year-old girl who presented with mild facial asymmetry and a hyperpigmented scar over the mid-cheek region and gingival overgrowth in the left posterior maxilla.

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Introduction

Segmental odontomaxillary dysplasia (SOD) is an uncommon, non-hereditary developmental disorder of the maxilla that typically presents in childhood [1]. The condition is characterized by unilateral maxillary enlargement, gingival hyperplasia, facial asymmetry, ipsilateral dental anomalies, and distinctive radiographic bone changes [2]. In cases where skin hyperpigmentation accompanies the oral and facial findings, Welsch and Stein (2004) [3] proposed the acronym HATS- referring to hemimaxillary enlargement, asymmetry of the face, teeth abnormalities, and skin findings. The etiology and pathogenesis of SOD remain unclear [4].

SOD is most commonly diagnosed in the first decade of life, with reported cases ranging from 5 to 27 years of age. There is a slight male predominance. Radiographically, the affected maxillary bone often exhibits an ill-defined radiopacity with a coarse, vertically oriented trabecular pattern, sometimes described as a ground-glass appearance. Additional findings may include reduction or obliteration of the maxillary sinus. Histopathological examination typically reveals immature woven bone with prominent reversal lines, and an absence of osteoblastic or osteoclastic rimming [5-6].

The associated dental abnormalities may include missing premolars, malformed primary molars, delayed eruption, or the presence of supernumerary teeth. Cutaneous findings, though variable, have been reported in several cases. These may include facial hypertrichosis, hyperpigmented patches, or less commonly, lesions resembling Becker's nevus [7]. Its diagnosis is mainly based on clinical and radiographic presentation [2].

Given the rarity of SOD and the limited number of reported cases, this study presents a new case accompanied by a brief literature review to analyze previously documented clinical and radiographic characteristics and to enhance the academic understanding of this condition. Herein, we present a case report on SOD in a 5-year-old girl.

Case Presentation

A 5-year-old girl was brought to the dental outpatient department by her parents with concerns regarding swelling of the left maxilla. There was no history of trauma, systemic illness, or significant family medical conditions. The child's birth and developmental history were normal. On extraoral examination, mild facial asymmetry was noted with flattening of left middle third of her face. The left angle of her mouth was pulled upwards even at rest. The lips were incompetent and mild drooping of left eye was seen (Figure 1).

A well-defined, elongated hyperpigmented scar measuring approximately 1.8×0.7cm was observed on the mid-region of the left cheek (Figure 2). The overlying skin was intact, and the area was non-tender and not associated with any discharge or inflammation. No lymphadenopathy or neurosensory deficits were detected. Intraoral examination revealed gingival overgrowth in the left posterior maxillary region with buccolingual expansion of the alveolar ridge. The mucosa overlying the region appeared firm and of normal color (Figures 3a-b).

Panoramic radiography demonstrated a unilateral alteration involving the left posterior maxilla. The affected region extended from the primary canine area posteriorly to the maxillary tuberosity. The involved segment exhibited an irregular mixed radiolucent–radiopaque appearance with coarse and disorganized trabecular architecture. The alveolar process appeared enlarged compared with the contralateral side. Multiple developing and erupting teeth within the affected segment showed delayed eruption and altered orientation; however, no evident root resorption was observed.

The axial cone beam computed tomography (CBCT) sections provided more detailed characterization. The lesion demonstrated expansion of the alveolar process with buccal cortical plate thinning but without evidence of cortical perforation. The internal bone pattern was coarse and irregular, lacking the normal uniform trabecular organization seen on the contralateral side. The palatal cortex showed outward expansion. Tooth follicles within the involved segment appeared enlarged and closely approximated, contributing to crowding and displacement of the developing teeth. Comparison with the unaffected right maxilla revealed normal trabecular architecture, preserved cortical boundaries, and symmetrical sinus pneumatization. No similar osseous or dental alterations were identified on the contralateral side, highlighting the distinctly unilateral nature of the lesion. Overall, the imaging findings demonstrate a loc-

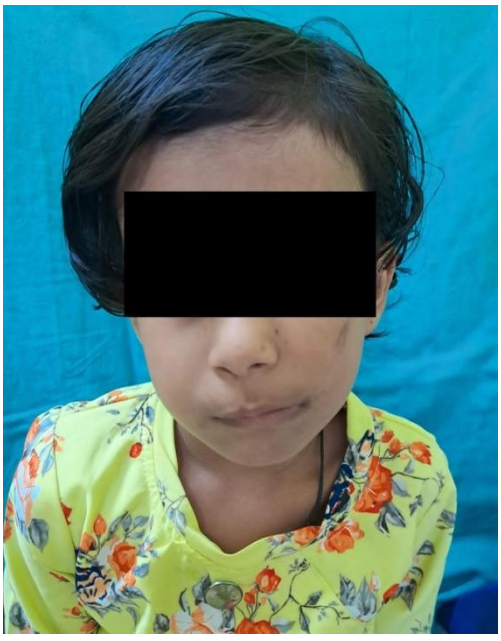

Figure 1: Clinical photograph showing mild facial asymmetry with flattening of the left cheek, pulling up of the left corner of the mouth

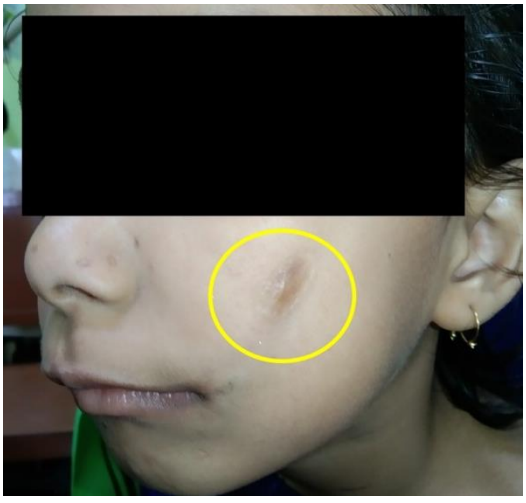

Figure 2: Clinical photograph showing well-defined hyperpigmented scar on the left mid-cheek region

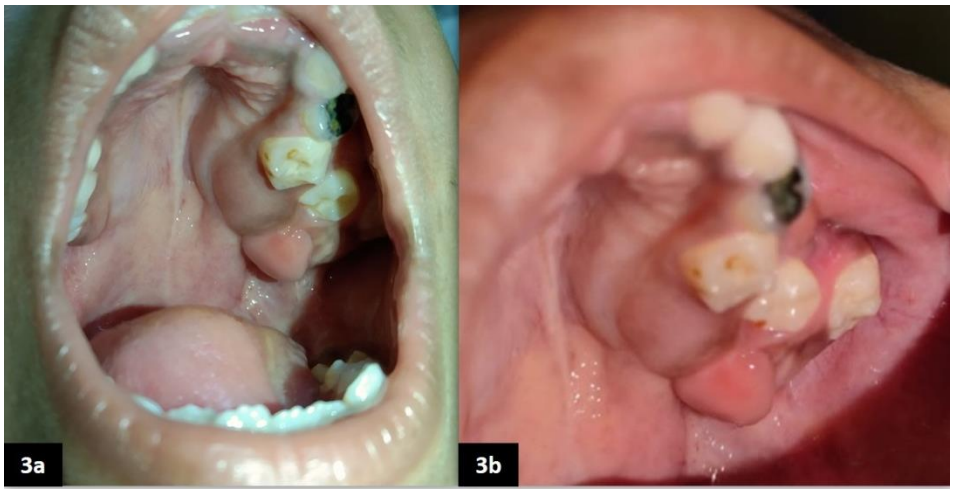

Figure 3: a: Intraoral photographs showing gingival overgrowth and **b:** buccolingual expansion in the left maxillary posterior region

alized unilateral developmental anomaly characterized by alveolar expansion, altered trabecular pattern and delayed tooth eruption (Figures 4a-c).

Based on the clinical and radiographic findings, a diagnosis of SOD was established. The patient was asymptomatic, and there were no functional limitations or signs of infection. The family was counseled about the benign and non-progressive nature of the condition and reassurance was provided. No active intervention was indicated at the time of diagnosis. The patient was placed under regular clinical and radiographic follow-up to monitor maxillofacial growth, dental development, and potential need for future multidisciplinary management, including surgical, orthodontic, or prosthetic interventions as required.

Discussion

Facial asymmetry in paediatric patients may arise from

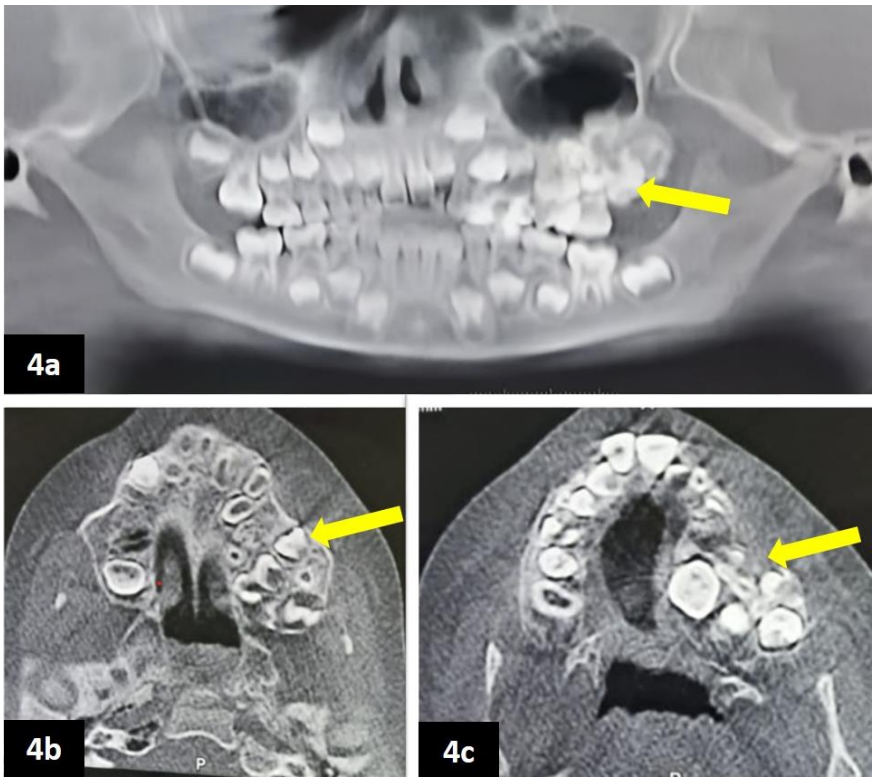

Figure 4: CBCT (Cone Beam Computed Tomography) images showing segmental odontomaxillary dysplasia involving the left maxilla. **a:** Panoramic radiograph reveals malformed and delayed-erupting teeth in the left posterior maxilla (yellow arrow), **b-c:** Axial CBCT slices demonstrate buccolingual expansion, increased bone density with coarse, irregular trabeculae in the affected maxillary segment (yellow arrows)

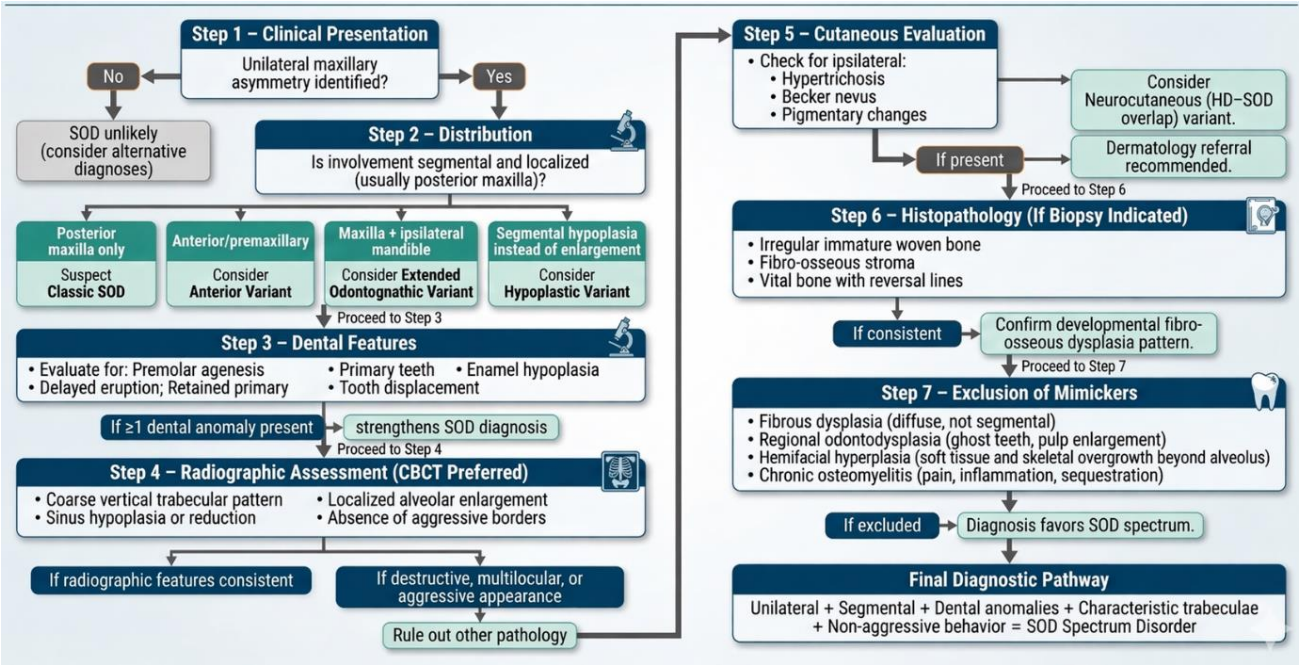

Figure 5: Diagnostic algorithm for segmental odontomaxillary dysplasia (SOD) and its phenotypic spectrum (SOD: Segmental odontomaxillary dysplasia; CBCT: Cone Beam Computed Tomography; HD-SOD: Hemimaxillofacial dysplasia - Segmental odontomaxillary dysplasia)

a broad spectrum of developmental, inflammatory, neoplastic, and congenital conditions. A comprehensive differential diagnosis includes fibrous dysplasia, hemifacial hypertrophy, congenital hemifacial hyperplasia, and vascular or lymphatic malformations [8]. Among these entities, SOD represents a rare but significant developmental anomaly, particularly when unilateral maxillary enlargement is associated with gingival overgrowth and dental abnormalities [2].

SOD was first described by Miles *et al.* [9] in 1987 under the term of hemimaxillofacial dysplasia. In 1990, Danforth *et al.* [10] formally introduced the designation segmental odontomaxillary dysplasia, establishing it as a distinct clinicopathological entity. Subsequently, Packota *et al.* [11] reviewed 12 cases and further characterized its radiographic profile, contributing significantly to the recognition of its imaging features.

Clinically, SOD typically presents as a non-progressive unilateral facial asymmetry involving the gingiva, dentition, and alveolar bone. The affected region most commonly extends from the canine eminence to the maxillary tuberosity. Cutaneous manifestations may or may not be present and have been reported in approximately 48% of cases, with a male predominance approaching 80%. Reported dermatologic findings include facial hypertrichosis, erythema, upper lip hypopigmentation, and lesions resembling Becker's nevus. Less frequent features include hairy naevi, vermilion border discontinuity, focal cheek depressions, and patchy hyperpigmentation [12].

Othman *et al.* [13] proposed a clinical classification system distinguishing two primary forms:

- **Type I (Classical):** isolated gingivo-dento-alveolar involvement
- **Type II (Cutaneous):** combined dermato-gingivo-dento-alveolar involvement

Type II is further subclassified into hypertrichotic (IIa), pigmentary (IIb), erythematous (IIc), and commissural defect (IId) variants, with combinatorial forms also described. Our case corresponds to the Type IIb (pigmentary) subtype.

Radiographically, SOD is characterized by irregular, vertically oriented trabeculae producing a coarse, granular, or ground-glass appearance. Buccolingual expansion of the alveolar process is common, often accompanied by hypoplasia or partial obliteration of the ipsilateral maxillary sinus. Impacted, delayed, or displaced teeth are frequently observed within the dysplastic segment. In some cases, nasal septal deviation may occur, secondary to unilateral maxillary enlargement. Although not pathognomonic, these imaging features strongly support the diagnosis when correlated with clinical findings [8, 14].

Histopathological findings are generally non-specific and serve primarily as supportive evidence. Gingival specimens typically demonstrate connective tissue hyperplasia without inflammatory changes. Osseous samples reveal thick trabeculae of immature woven bone with prominent reversal lines within a fibrous stroma lacking significant osteoblastic or osteoclastic activity [13].

The etiopathogenesis of SOD remains uncertain. Proposed mechanisms include a localized developmental disturbance in utero, a unilateral developmental field defect involving the first and second branchial arches, or infection affecting the maxillary division of the trigeminal nerve during early development. A post-zygotic mutation has also been suggested to explain the segmental and sporadic presentation [14]. Supporting this hypothesis, Gibson *et al.* [15] identified a somatic *PIK3CA* mutation in a case with mandibular involvement, raising the possibility that SOD may fall within the PIK3CA-related overgrowth spectrum.

A comparative analysis of the previously reported cases reveals that unilateral posterior maxillary enlargement, delayed or abnormal tooth eruption, and maxillary sinus hypoplasia represent the most consistent radiographic features of SOD. Cutaneous manifestations, although frequently described, are not universally present and therefore should not be considered mandatory for diagnosis. The present case aligns closely with the classical radiographic pattern reported in the literature, particularly regarding unilateral alveolar expansion and ipsilateral sinus hypoplasia. The absence of aggressive radiographic characteristics further supports its developmental rather than neoplastic nature.

In most cases, diagnosis can be established based on characteristic clinical and radiographic findings, with histopathology and genetic testing reserved for selected cases. Based on the clinical, radiographic and literature findings summarized in this study (Table 1), a diagnostic algorithm is proposed to assist clinicians in differentiating SOD from other unilateral maxillary pathologies (Figure 5).

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Conclusion

SOD is a rare, non-hereditary developmental anomaly that primarily affects the maxilla, presenting with characteristic clinical, dental, and radiographic features. Although its etiology remains uncertain, emerging genetic insights such as somatic PIK3CA mutations may offer a broader understanding of its pathogenesis. Diagnosis is largely based on clinical and radiographic findings, often sufficient without the need for invasive investigations. Awareness of this condition is essential to avoid misdiagnosis and unnecessary treatment. Long-term follow-up and multidisciplinary management may be required, and further case reporting is encouraged to enhance knowledge and develop standardized treatment protocols.

Conflict of Interest

The authors declare that they have no competing interests.

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