Segmental Odontomaxillary Dysplasia: A Case Report with Review of Diagnostic and Clinical Features

Cota Jochima E.[1], Spadigam Anita E.[1], Dhupar Anita[1], Syed Shaheen[1], Rebello Nairica[1]
[1]*Dept of Oral and Maxillofacial Pathology, Goa Dental College and Hospital, Bambolim, Goa*

Corresponding Author
Jochima Eudora Cota
Email ID: jochimacota@gmail.com

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Introduction

Segmental odontomaxillary dysplasia (SOD) is a rare, non-progressive developmental disorder that affects the maxillary region, leading to unilateral maxillary enlargement and dental anomalies.[(1)] Initially described in 1987 as hemifacial dysplasia,[(2)] SOD continues to be underrecognized, with an evolving understanding of its pathogenesis and clinical presentation. The 5th edition of the World Health Organization (WHO) Classification of Head and Neck Tumours (2022) has added SOD in the group of fibro-osseous lesions as a localized, benign developmental anomaly that presents with distinct clinical and radiographic features involving both soft and hard tissues.[(3)] Herein we present a unique case of SOD diagnosed in a 40-year-old female patient.

Case Report

Forty-year-old female patient, unmarried, working as a househelp, reported to the institute for a denture. However, on examination a firm, non-fluctuant swelling (figure 1B) was noted on right maxillary palate which was fixed to the underlying bone. Multiple missing teeth were noted, the patient has no recollection of extraction or exfoliation in the affected quadrant. A history of exfoliation of mandibular teeth is noted. There was no relevant medical history. No history of trauma, pain or bleeding from the affected side. Extraorally, facial asymmetry and facial depression on the affected side is noted (figure 1A). Radiographic findings revealed ill-defined, coarse trabeculation of the right posterior maxilla, horizontally impacted 13 and several missing teeth (figure 1c). Serum analysis revealed no abnormality. Histopathology (figure 2) revealed a hyperplastic fibrocellular stroma with thickened blood vessels. Areas of hyalinisation of collagen fibres and dystrophic calcifications were evident. Hard tissue revealed mature vital bone with resting as well as reversal lines and no osteoblastic rimming. The surrounding stroma was hypocellular, with no inflammation. Based on the above findings a diagnosis of SOD was arrived upon.

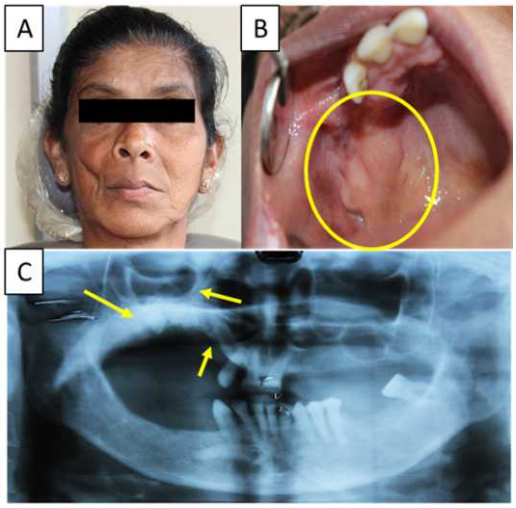

Figure 1: Clinical images (A & B); Orthopantomograph showing course trabeculation, small sinus and impacted 13 (C).

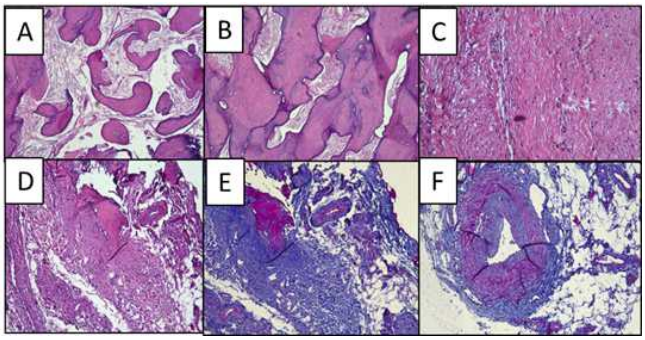

Figure 2: A & B: trabeculae of vital bone with resting and reversal lines (H &E, 10x, 40x); C and D: Connective tissue stroma showing hyalinised collagen fibres and thickened blood vessels (H & E stained, 10x); E and F: Masson trichrome stain showing collagen fibres and thickened blood vessels (10x)

Discussion

SOD appears in childhood, often with the eruption of permanent teeth, though diagnosis may be delayed due to subtle progression. Clinically, it is more common in males, shows unilateral maxillary hard and soft tissue enlargement and dental abnormalities like hypoplastic or missing teeth,

delayed eruption, and malocclusion. Facial asymmetry and variable cutaneous abnormalities are part of the clinical presentation. In 2004 the acronym HATS (Hemimaxillary enlargement, Asymmetry of the face, Teeth abnormalities, Skin findings) was introduced by Welsch and Stein. Packota et al put forth radiographic diagnostic criteria which included sclerosis of bone with thickened trabeculae, missing premolars with delayed eruption of permanent teeth, vertical orientation of bony trabeculae, spacing between deciduous molars, and a small maxillary sinus on the affected side.[4,5] The pathogenesis of SOD remains incompletely understood. It is thought to arise from a localized defect in the ectomesenchymal tissue of the maxilla during early embryogenesis and Phosphatidylinositol-4,5-Bisphosphate 3-Kinase Catalytic Subunit Alpha (*PIK3CA*) or actin beta (*ACTB*) gene mutations are suspected.[6,7]

One of the key diagnostic challenges in SOD is distinguishing it from similar conditions. The WHO update has reinforced the importance of differentiating SOD based on its characteristic clinical and radiographic findings, such as unilateral maxillary involvement, non-ossifying fibrous tissue proliferation, and absence of the more aggressive features seen in conditions like fibrous dysplasia.[3,8] Radiologically, the presence of hypoplastic or absent teeth in the affected region, along with trabecular bone thickening, can aid in the diagnosis.[5]

Management of SOD is primarily conservative, focusing on addressing dental and orthodontic complications arising from hypodontia, malocclusion, and delayed tooth eruption. Surgical intervention may be required in cases with significant facial asymmetry, but treatment is generally individualized based on the severity of functional and aesthetic concerns.[7] The prognosis for patients with SOD is favourable, with most individuals achieving satisfactory outcomes with minimal long-term complications.[9]

In conclusion, SOD is a rare but clinically significant developmental anomaly that requires careful diagnosis to avoid confusion with other craniofacial disorders. The recent updates from the WHO classification have clarified key aspects of its pathogenesis and diagnostic criteria, aiding in the accurate identification of this condition. Further research into the genetic and embryological basis of SOD is warranted to fully understand its etiology and improve diagnostic precision.

Source of Support: Nil

Conflict of Interest: Nil

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