

Case Report

Congenital Leiomyomatous Hamartoma of the Anterior Maxillary Gingiva with 4-Year Follow-up: A Rare Case Report and Literature Review

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KEY WORDS

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ABSTRACT

This report presents a rare case of congenital oral leiomyomatous hamartoma (OLH) involving the anterior maxillary gingiva and includes a review of previously reported cases and the present case. A 5.5-year-old girl presented with a solitary, painless, and exophytic lesion on the labial gingiva that had been present since birth and had gradually increased in size. Based on the clinical presentation, the lesion was provisionally diagnosed as congenital epulis. However, histopathological examination revealed features consistent with OLH. The patient underwent excisional biopsy and was followed for 4 years, during which no recurrence was observed. The presence of a polypoid lesion in the midline region of the anterior maxillary gingiva should prompt consideration of OLH in the differential diagnosis. Comprehensive histopathological evaluation, supplemented by immunohistochemical staining when indicated, is essential for establishing an accurate diagnosis and guiding appropriate management. This case contributes to the limited literature on OLH by providing additional insights into its clinical presentation, diagnosis, treatment, and favorable prognosis, while emphasizing the importance of meticulous histopathological evaluation.

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Introduction

The term hamartoma is derived from the Greek word hamartia, meaning defect or error [1]. Willis [2] defined a hamartoma as a "tumor-like, but primarily non-neoplastic malformation or inborn error of tissue development, characterized by an abnormal mixture and overgrowth of relatively normal tissues indigenous to the affected site". Hamartomas are classified according to the predominant tissue components involved in the lesion [3]. Leiomyomatous hamartoma is a benign developmental malformation characterized by an abnormal proliferation of smooth muscle tissue. These lesions may occur as either solitary or multiple lesions. The presence of multiple hamartomas in a single patient is commonly referred to as hamartomatosis or pleiotropic hamartoma [1, 4].

Most reported cases of oral leiomyomatous hamartoma (OLH) present as solitary lesions. Although a few cases with multiple lesions have been described [5-6], these were not associated with any recognized syndrome and occurred in otherwise healthy individuals [7].

Several hallmarks of hamartomas have been described in the literature. They are typically asymptomatic [8], exhibit self-limited growth, enlarge proportionally with the surrounding tissues, are usually unencapsulated with ill-defined margins, and may be associated with chromosomal abnormalities or congenital syndromes. Developmental malformations may be present at birth but manifest later and may regress spontaneously [1]. The majority of reported cases occur exclusively in the tongue including the tip, dorsum, and base, or in the anterior maxillary gingiva and alveolar ridge, partic-



Figure 1: Clinical and radiographic findings, **a:** Initial appointment showing a well-circumscribed, smooth-surfaced, non-ulcerated lesion on the anterior maxillary labial gingiva, **b:** Second appointment demonstrating surface ulceration attributed to traumatic occlusion, **c:** Periapical radiograph showing no evidence of underlying osseous involvement

ularly the incisive papilla region [7-8].

Case Presentation

A 5.5-year-old girl with no significant medical history presented to a private dental clinic in April 2022 for a routine oral examination. Her mother reported an uncomplicated full-term pregnancy, and there was no family history of similar lesions or congenital anomalies. Clinical examination revealed a solitary, painless, exophytic lesion arising from the labial gingiva in the anterior maxillary midline, located between the primary maxillary central incisors and adjacent to the labial frenum (Figure 1a). According to the patient's mother, the lesion had been present since birth and had gradually increased in size over time without any noticeable changes in its surface characteristics or consistency.

The patient was initially referred for further evaluation but returned five months later with complaints of pain and feeding difficulties caused by repeated traumatic occlusion. She was subsequently referred to the Department of Pediatric Dentistry, School of Dentistry, Islamic Azad University of Medical Sciences, Tehran, Iran. Intraoral examination revealed a mobile, pedunculated, firm nodule measuring approximately 13×4×2 mm, covered by ulcerated mucosa (Figure 1b). No regional lymphadenopathy was detected. Based on the

clinical findings, a provisional diagnosis of congenital epulis was made. Radiographic examination revealed no evidence of intraosseous involvement (Figure 1c).

An excisional biopsy was performed in September 2022 under local anesthesia using 2% lidocaine with 1:100,000 epinephrine. The lesion was gently grasped with a hemostatic forceps to temporarily control vascular supply (Figure 2a) and completely excised using a No. 15 surgical blade. A conventional frenectomy was performed concurrently (Figure 2b). Hemostasis was achieved through direct pressure with sterile gauze, and the surgical site was closed using seven single interrupted sutures (Figure 2c). The excised specimen was fixed in 10% buffered formalin and submitted for histopathological evaluation. Postoperative healing was uneventful. No recurrence was observed at the 24-month and 48-month follow-up examinations (Figures 3-4).

Gross examination revealed an ovoid, creamy-brown, firm soft-tissue specimen measuring 13×4×2mm (Figure 5). Histopathological analysis demonstrated keratinized stratified squamous epithelium overlying fibrous connective tissue containing well-circumscribed fascicles of spindle-shaped cells with cigar-shaped nuclei interspersed with thin-walled blood vessels and occasional nerve bundles. On hematoxylin and eosin staining, the spindle cells exhibited morphological feat-



Figure 2: Surgical procedure, **a:** The lesion was grasped with a hemostatic forceps to facilitate excision and minimize bleeding, **b:** Complete surgical excision of the lesion using a conventional scalpel technique, with simultaneous frenectomy, **c:** Primary wound closure achieved with seven simple interrupted sutures



Figure 3: 2-year follow-up revealing successful healing with no evidence of recurrence, **a:** Occlusal view and **b:** Frontal view

ures consistent with mature smooth muscle fibers. Mild chronic mononuclear inflammatory cell infiltration was present, with no evidence of cytologic atypia or malignancy (Figure 6).

To further characterize the lesion, Masson's trichrome staining was performed, demonstrating smooth muscle fibers stained red in clear contrast to the surrounding collagen fibers, which stained blue (Figure 7). Based on the clinical presentation, histopathological findings, and special staining results, a diagnosis of OLH was established.

Immunohistochemical staining with smooth muscle actin and desmin was performed. However, interpretation was limited because only a very small amount of residual tissue remained in the paraffin block after routine diagnostic processing. Internal positive controls were identified in vascular smooth muscle, whereas immunoreactivity within the lesional tissue was inconclusive.

Discussion

Hamartomas are histopathologically classified according to their predominant tissue components [9-10]. OLHs, in particular, are benign developmental malformations composed predominantly or exclusively of smooth muscle tissue [1, 5, 11-12]. A comprehensive review of the literature suggests that congenital OLHs

are likely underreported. This underreporting may be attributed to their asymptomatic nature and slow, self-limited growth, with lesions rarely exceeding 3 cm in diameter. Consequently, these lesions often remain undiagnosed or are overlooked for several years. Increased reporting of such cases may contribute to a more accurate understanding of their epidemiology and clinico-pathological characteristics [12].

A literature search was conducted in PubMed, Scopus, and Google Scholar using combinations of the terms "oral leiomyomatous hamartoma," "leiomyomatous hamartoma," "congenital leiomyomatous epulis," and "smooth muscle hamartoma." Publications identified from the reference lists of relevant articles were also screened. Publications published from 1945 onward were considered eligible for inclusion. One report was excluded because it lacked sufficient clinical and histopathological information for case characterization. Overall, 70 cases of OLH, including the present case, were identified in our literature review. Of these, 41 involved the tongue, 27 occurred in the anterior maxillary region, and 2 were located on the palate. Table 1 summarizes the previously reported cases of OLH involving the anterior maxillary region, together with the present case.

Earlier reports have documented OLH in both females and males [13-14]. Kuperan *et al.* [15] reported

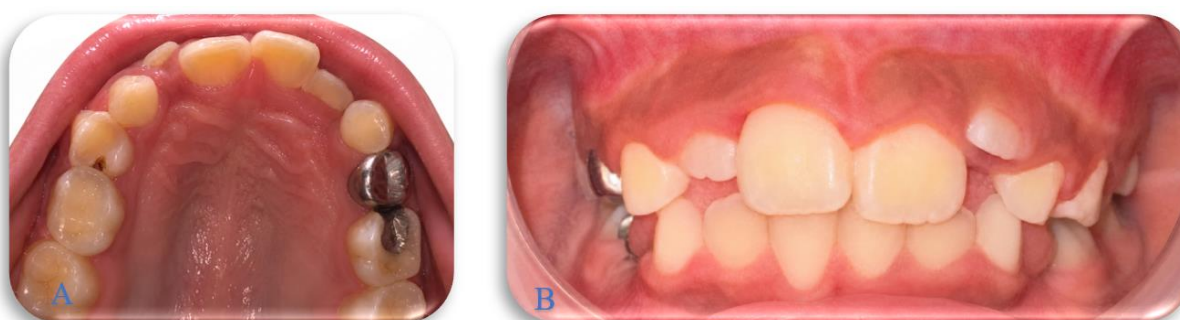


Figure 4: 4-year follow-up revealing successful healing with no evidence of recurrence, **a:** Occlusal view and **b:** Frontal view

Table 1: Summary of clinical characteristics of oral leiomyomatous hamartoma (OLH)

Case	First authors, Reference No.	Year	Ethnic Group	Age	Age of presentation	Gender	Location	Clinical appearance	Size (cm)
1	Takahashi [27]	1962	—	2.5mon	20 days	F	Median maxilla	—	0.4×0.5×0.7
2	Mushimoto [28]	1982	Japan	11mon	At birth	F	Median maxilla	—	0.5×0.5×0.7
3	Kajiyama [29]	1983	—	4yrs and 5mon	5 mon	F	Median maxilla	—	1.5×0.6×0.7
4	Kanekawa [6]	1990	—	3 yrs	At birth	M	Median maxilla	—	0.5×0.5×0.5
5	Seki [30]	1991	—	2yrs and 3mon	At birth	F	Median maxilla	—	0.8×0.5×0.4
6	Ng [31]	1992	Malaysia	3mon	At birth	F	Incisive papilla	Sessile mass	1×0.5
7	Semba [11]	1993	Japan	26mon	At birth	M	Anterior maxillary alveolar ridge	Polypoid mass, producing slight displacement of teeth	0.5×0.5×0.4
8	Takeda [14]	2000	Japan	10mon	At birth	M	Anterior maxillary alveolar ridge	Polypoid mass	0.6×0.6×0.6
9	Correa [3]	2001	Brazil	6 yrs	At birth	F	Incisive papilla	Pedunculated nodule	0.5
10	Kujan [32]	2007	UK	11mon	At birth	F	Anterior maxillary alveolar ridge	Lobular, sessile, soft tissue polyp	1
11	Zaitoun and Triantfyllou [17]	2007	Caucasian	8 yrs	At birth	M	Anterior maxillary alveolar ridge	Pedunculated, smooth, polypoid lesion	0.6
12	Lida [5]	2007	Japan	2 yrs and 7mon	2.5 yrs	M	Incisive papilla	Polypoid mass	0.5×0.3×0.4
13	Scarpelli [13]	2007	Latin America	6mon	—	F	—	—	0.5×0.2×0.3
14	Nava-Villalba [12]	2008	Mexico	19 yrs	Several years	F	Anterior maxillary alveolar ridge	Pedunculated, polypoid, exophytic lesion	0.5×0.8×0.7
15	Zhang [33]	2011	Japan	2 yrs	5 mon	F	Anterior maxillary alveolar ridge	Polyp like lesion	0.5×0.5×0.7
16	AlQahtani and Qannam [34]	2013	Saudi Arabia	18mon	18 mon	M	Anterior maxillary alveolar ridge	Pedunculated, smooth, light pink, and soft polypoid mass.	3×2×3
17	Falci [7]	2013	Brazil	3mon	At birth	M	Anterior maxillary alveolar ridge	Pedunculated nodule	2.0
18	Damm [35]	2014	—	2 yrs	—	F	Incisive papilla	—	0.7×0.5×0.5
19	Raghunath [36]	2016	India	15 yrs	14.5 yrs	F	Anterior maxillary alveolar ridge	Reddish, smooth mass, displacing central incisors	1.5×2
20	Da Silva [37]	2016	USA	4 yrs	2 yrs	F	Anterior maxillary alveolar ridge	Smooth-surfaced pink nodule	0.5×0.4×0.4
21	Loomba [21]	2017	India	2 yrs	At birth	F	Incisive papilla	small nodule, firm, painless, pedunculated	2–2.5×0.5×0.5
22	Sanchez-Romero [8]	2019	Brazilian	3yrs	--	M	Incisive papilla	Painless pedunculated nodule	0.5
23	Yancoskie [38]	2024	USA	1mo	At birth	F	Incisive papilla	Cylindrical nodule	1.2×0.4×0.4
24	Yancoskie [38]	2024	USA	15 yrs	5 yrs	F	Incisive papilla	Smooth, mucosa-colored sessile mass	1.5×0.8
25	Yancoskie [38]	2024	USA	4 yrs	—	F	Incisive papilla	mucosa-colored, smooth-surfaced bi-lobed nodule	1×0.5
26	Sujeet S Pal [39]	2024	India	6 yrs	At birth	F	Anterior maxillary alveolar ridge	gingiva-colored, firm consistency noninflamed.	0.8×0.4×0.4
27	Jadidi (present case)	2026	Iran	5.5 yrs	At birth	F	Anterior maxillary alveolar ridge	mobile, pedunculated nodule, with firm consistency, covered by ulcerative mucosa	1.3×0.4×0.2

yrs, years; mon, months; F, female; M, male; cm, centimeter



Figure 5: Excised specimen immediately post-surgery

39 histologically confirmed cases of leiomyomatous hamartoma, with an equal distribution between males and females. Consistent with these findings, a recent systematic review of 66 cases found a nearly equal sex distribution, with males accounting for 50.7% and females for 49.3% of cases [16]. The lesion varies significantly in size, ranging from just a few millimeters in diameter to several centimeters. It is found on the maxillary alveolar ridge two to three times more frequently than on the mandibular ridge [14]. OLH generally presents as a solitary lesion. Although there have been a few reports of multiple occurrences, such cases are rare, indicating that the presentation of multiple lesions is uncommon [5]. While these lesions are congenital, the reported age at diagnosis ranged from birth to 61 years, with a mean age of 71.23 ± 123.01 months (approximately 6 years) [16].

The frequent occurrence of OLH in the anterior maxillary midline has led to the hypothesis that these lesions arise from developmental errors during fusion of the primary palate with the lateral palatine processes. Alternatively, they may originate from smooth muscle cells associated with blood vessels emerging from the nasopalatine foramen [7, 17].

The differential diagnosis of OLH includes several benign developmental, reactive, neural, myofibroblastic, and myogenic lesions that may present as a congenital or early-life oral mass. Fibrous epulis and fibroepithelial polyp should be considered because both may present

clinically as firm, sessile, or pedunculated mucosal nodules; however, they are composed predominantly of fibrous connective tissue rather than disorganized bundles of mature smooth muscle [18-19]. Leiomyoma represents an important histopathological differential diagnosis because it is a true benign smooth muscle neoplasm, whereas OLH is a developmental malformation composed of mature smooth muscle arranged in a hamartomatous pattern [20]. Other developmental lesions, including palatal and gingival cysts of the newborn and incisive canal (nasopalatine duct) cysts, may also be considered, particularly for congenital lesions arising in the anterior maxillary midline [21]. Rhabdomyoma and skeletal muscle hamartoma should be excluded because they demonstrate skeletal muscle differentiation rather than smooth muscle predominance [22]. Neurofibroma may also enter the differential diagnosis as a benign spindle-cell lesion of the oral mucosa but is distinguished by neural differentiation and immunoreactivity for S-100 protein [23]. Congenital epulis (congenital granular cell tumor) is a particularly important clinical mimic because it typically arises on the anterior maxillary alveolar ridge at birth and may interfere with feeding [24]. Myofibroma should likewise be considered in pediatric patients because it may present as a solitary spindle-cell lesion of the oral cavity or jaws with smooth muscle actin positivity; however, its characteristic biphasic myofibroblastic architecture distinguishes it from OLH [25-26].

The clinical findings of the present case are consistent with previously reported cases described by Semba *et al.* [11], Takeda *et al.* [14], and Correa *et al.* [3]. Histopathological examination demonstrated an admixture of mature smooth muscle bundles, peripheral nerve fibers, and scattered congested, dilated blood vessels, supporting the diagnosis of OLH arising in the

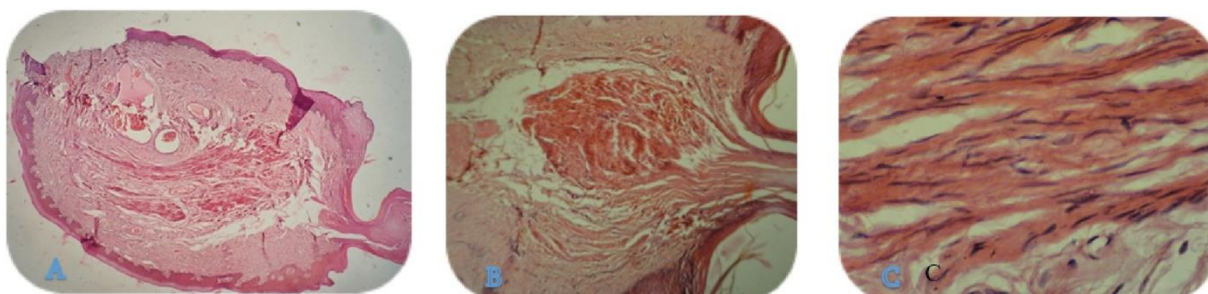


Figure 6: Hematoxylin and eosin staining, reveals keratinized stratified squamous epithelium overlying spindle cells, dilated thin-walled blood vessels, and a few nerve bundles in the fibrous connective tissue. The spindle cell fascicles resemble mature smooth muscle with mild mononuclear inflammatory cell infiltration and no atypical changes. (A) 40× (B) 100× (C) 400×

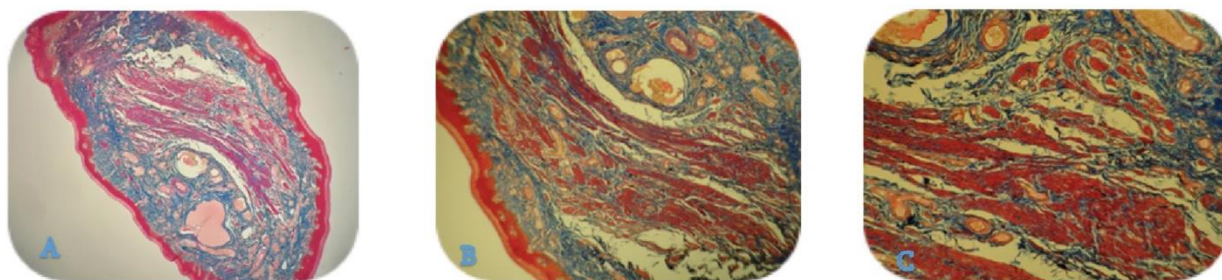


Figure 7: Masson's trichrome staining highlights smooth muscle cells in red, contrasting with blue-stained collagen fibers, confirming the presence of smooth muscle fascicles. These findings support the diagnosis of OLH, **a:** Magnification 40×, **b:** 100×, **c:** 400×

incisive papilla. The diagnosis was established using hematoxylin and eosin staining together with Masson's trichrome stain, which clearly differentiated smooth muscle fibers from the surrounding collagenous stroma. Although immunohistochemistry may facilitate confirmation of smooth muscle differentiation, it is not considered mandatory in all cases of OLH. When characteristic clinicopathological features are present, diagnosis can be established on conventional histopathological examination, with immunohistochemistry serving as an adjunctive tool. In the present case, immunohistochemical evaluation was limited by the very small amount of residual tissue available after routine diagnostic processing.

Complete surgical excision is the most commonly reported treatment for OLH and serves both diagnostic and therapeutic purposes. Following complete surgical excision, the prognosis appears favorable, and recurrence appears to be uncommon. Nevertheless, periodic clinical follow-up may be considered to confirm satisfactory healing and monitor for the possibility of recurrence [16].

Ethical Principles

Prior to data collection, informed consent was obtained from the patient's parent for the use of their medical information in this study. All data were handled confidentially and in accordance with ethical principles.

Conclusion

In conclusion, OLH should be considered in the differential diagnosis of congenital or early-life polypoid lesions involving the anterior maxillary gingiva, particularly those arising in the midline region. Accurate diagnosis relies primarily on careful histopathological evaluation. In the present case, hematoxylin and eosin staining, supported by Masson's trichrome staining, demon-

strated morphological features consistent with OLH. Immunohistochemical staining for desmin and smooth muscle actin may provide additional diagnostic information, particularly in cases with equivocal histopathological findings. However, immunohistochemical interpretation was limited in the present case because of insufficient residual tissue. This case further highlights the importance of comprehensive histopathological assessment in establishing an accurate diagnosis and guiding appropriate management of this rare developmental lesion.

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Conflict of Interest

There is no conflict of interest to declare.

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