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CASE REPORT

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Rare condition of unilateral submandibular gland aplasia and its diagnostic significance in oral cavity carcinoma

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Abstract

Background: The unilateral submandibular gland aplasia (agenesis) is a rare, asymptomatic condition usually discovered incidentally on imaging. This is associated commonly with either compensatory hypertrophy of contralateral submandibular gland or sublingual glands.

Case presentation: We report the case of a 34-year-old male with incidentally detected unilateral submandibular gland aplasia associated with hypertrophy of ipsilateral sublingual gland, demonstrated by imaging modalities, where we have highlighted the diagnostic significance of such rare findings in oncology, particularly in oral cavity carcinoma cases with metastatic submandibular lymph nodes (level IB) mimicking as submandibular gland. Hence, lymphadenopathy can be missed preoperatively which is an important part of staging and treatment planning.

Conclusions: Aim of the present report is to create awareness about such rare entity in both clinicians/radiologists and highlight the imaging features for correct identification and to avoid any diagnostic dilemmas.

Keywords: Submandibular gland aplasia, Oral cavity carcinoma, Computed tomography, Magnetic resonance imaging, Case report

Background

Aplasia (agenesis) of major salivary glands is a rare disorder affecting submandibular or parotid glands with an incidence of 1 in 5000 births [1]. Its etiology is unknown and it was thought to be due to the defect that occurs during the fetal development [2–6]. The exact incidence of major salivary gland agenesis is difficult to establish due to the asymptomatic nature. Till Date, approximately 40 aplasia cases were reported in the English literature [4], The unilateral submandibular gland (SMG) aplasia is an extremely rare condition (~ 20 cases) and its association with oral cancers is extremely rare with < 5 reported cases in the literature till date.

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The unilateral SMG aplasia patients are often asymptomatic (other gland compensates the secretion) [1, 4–9], whereas in symptomatic patients, due to inadequate amount of saliva, presenting symptoms were reported to be xerostomia, difficulty in swallowing, and dental problems. These patients can also present with a pseudomass due to compensatory hypertrophy of contralateral SMG or sublingual glands [1, 3–5, 7–12]. The treatment of this condition is primarily supportive and consists of reducing xerostomia and its effects, if present [4].

We present a case of aplasia (agenesis) of the right SMG, which was incidentally diagnosed during presurgical Magnetic Resonance Imaging (MRI) imaging of known case of right buccal mucosa carcinoma with metastatic right submandibular lymph node (level IB). This metastatic submandibular lymph node mimics as submandibular gland which is actually absent (agenesis). Hence, the aim of this case report is to emphasize the imaging characteristics and the clinical and diagnostic

significance of this rare entity to avoid any confusion in preoperative TNM (tumor—T, nodes—N, and metastases—M) staging and management.

Case presentation

A 34-year-old male was presented to our surgical department in the month of January 2022 with a chief complaint of a growth in the right side of the cheek since one month. Other than history of tobacco chewing (~10 to 12 years), no other significant past medical and dental history was reported.

On intraoral examination, right buccal mucosa showed mass with ulcerated surface, non-tender measuring ~ 4.1 × 3.0 cm. The mouth opening was decreased. On palpation the mass had soft to firm consistency. The mass involved upper gingivobuccal sulcus adjacent retromolar trigone, extending till angle of mouth. The floor of mouth was not involved. It was close to the skin; however, overlying skin was not involved. On extraoral examination, no palpable neck lymph nodes were noted. Further, the patient underwent punch incisional biopsy of the right buccal mucosal lesion, where histopathology report revealed a well differentiated squamous cell carcinoma (SCC).

Later an MRI scan of the neck was advised for further evaluation. Plain and contrast MRI neck examination was performed on a 1.5 T MRI machine. Spin echo sequences as T1W, T2W, short tau inversion recovery (STIR), and diffusion weighted imaging (DWI) were obtained. Post-Gadolinium T1W fat saturated sequences were obtained in axial, coronal, and sagittal planes. Two radiologists with an experience of more than five years have assessed and reported the MRI images. Results from MRI have revealed an ill-defined enhancing soft tissue mass size ~ 36 × 26 × 39 mm in AP × TRANS × CC (AP, anteroposterior; TR, transverse; CC, craniocaudal) involving right buccal mucosa, both upper, and lower gingivobuccal sulcus extending into retromolar trigone (Fig. 1A–C). Right SMG in the right submandibular region was absent. Whereas, the left SMG was observed to be normal in its size with few linear T2 weighted high signal intensities within (Fig. 2A–C). A mild hypertrophy was noted in the right sublingual gland (Fig. 3A, B). A well-defined homogeneously enhancing round lymph node measuring approximately 15 × 14 mm was seen at right level IB (submandibular region). It shows restricted diffusion on DWI and apparent diffusion coefficient (ADC) images. This was mimicking as SMG (Fig. 3A–D). No other enlarged cervical lymph nodes were detected. The right parotid gland was atrophic (Fig. 1B, C). Plain computed tomography (CT) neck screening was done to look for any bone erosion and no such thing was noted (Fig. 4). Further workup was done with low dose screening of CT thorax (LDCT), revealing no evidence of lung nodules/metastasis or enlarged mediastinal lymph nodes.

The patient underwent bite composite resection (right marginal mandibulectomy + right maxillary alveolectomy) and right modified neck dissection (MND) preserving spinal accessory nerve, internal jugular vein (IJV), and sternocleidomastoid muscle. Microvascular reconstruction was done with left anterolateral thigh (ALT) free flap and anastomosis of the flap vessels were done with right facial artery (END to END) and right IJV (END to SIDE). No post-operative complications were reported.

Final surgical histopathology report revealed moderately differentiated squamous cell carcinoma of buccal mucosa with regional lymph node involvement. The pathological stage classification was reported to be pT4a pN2a pMx with 20 mm of DOI. One lymph node out of

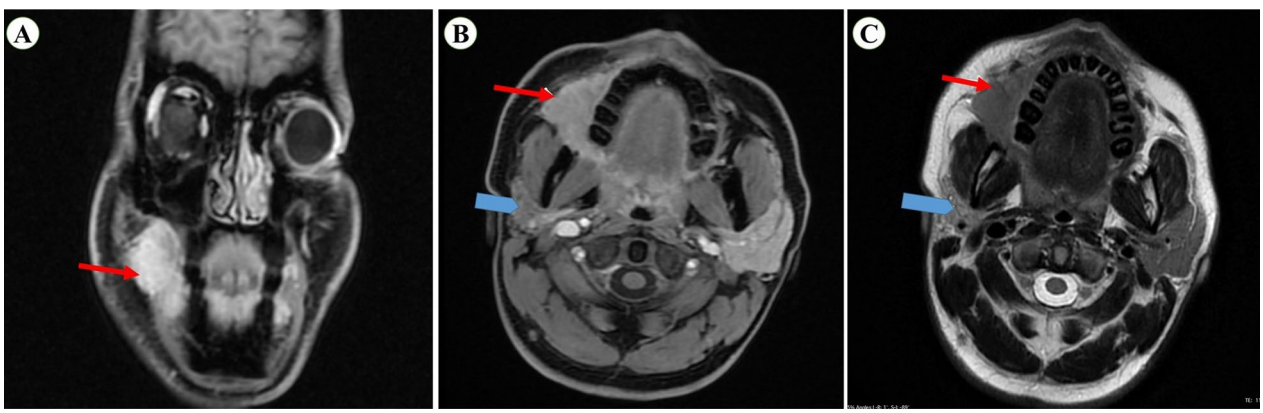

Fig. 1 MRI images—**A** Axial T2W, **B** Axial post-contrast T1W FS, and **C** coronal post-contrast T1WFS revealing an ill-defined enhancing soft tissue mass lesion involving right buccal mucosa, both upper and lower gingivobuccal sulcus, extending into retromolar trigone (red arrow—**A**, **B**, **C**). The right parotid gland was small in size—atrophy (pentagon arrow—**B**, **C**)

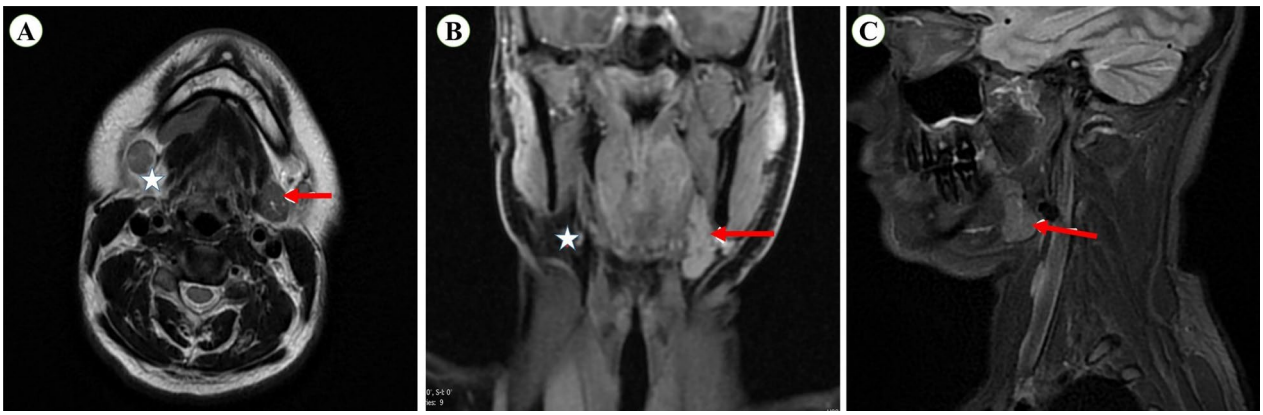

Fig. 2 MRI—**A** Axial T2W, **B** Coronal post-contrast T1W FS, and **C** Sagittal STIR images revealing the left submandibular gland of normal in size with normal signal intensity (red arrow—**A**, **B**, **C**). The right submandibular gland was absent in right submandibular region (star—**A**, **B**)

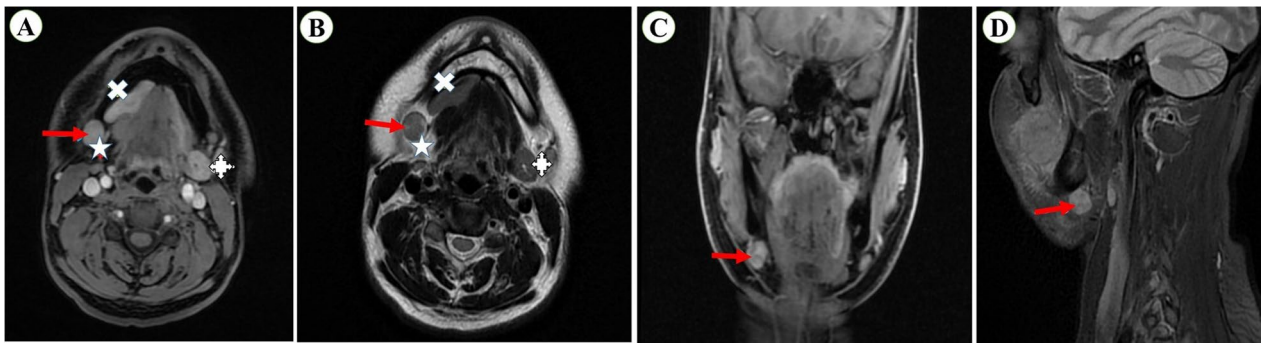

Fig. 3 MRI—**A** A single well-defined rounded homogeneously enhancing lymph node noted in the right submandibular region (right level IB) (red arrow—**A**, **B**, **C**, **D**). This mimics as right submandibular gland. **B** T2W axial image, linear high signal seen in left submandibular gland due to ducts. The right submandibular gland was absent in right submandibular region (star—**A**, **B**). The left submandibular gland was normal (quad arrow callout—**A**, **B**). The right sublingual gland was showing hypertrophy (cross—**A**, **B**)

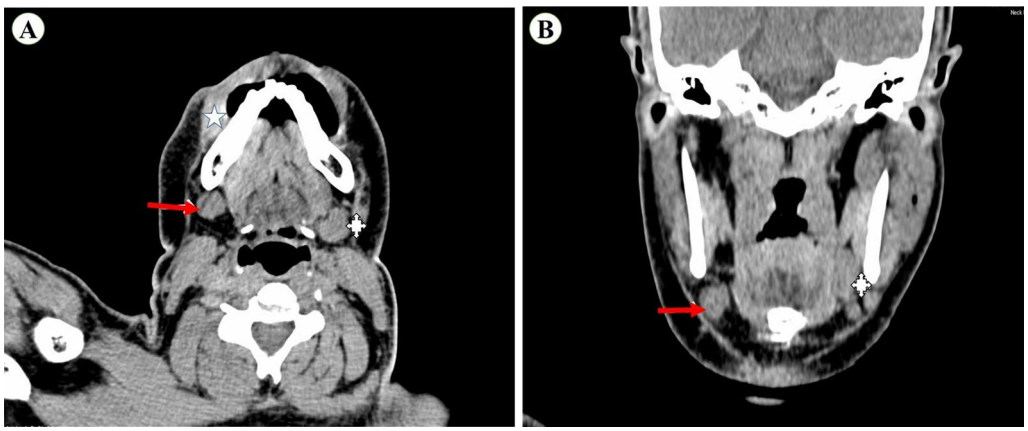

Fig. 4 CT scan neck plain—**A** Axial **B** coronal CT images revealing normal left submandibular gland (quad arrow callout—**A**, **B**) and enlarged lymph node in the right submandibular fossa (arrow—**A**, **B**). Right buccal mucosa mass also observed (star—**A**, **B**). No bone erosion was seen. Right sublingual gland hypertrophy not well seen on CT images

28 lymph nodes dissected has showed a tumor deposit with extracapsular extension (ECE) of > 2 mm and this node measuring 18 × 10 mm. Which was correlated with enlarged round lymph node seen on MRI images in SMG region mimicking as normal submandibular gland. In MND specimen, right SMG was not found confirming SMG agenesis.

Only on careful evaluation of MRI images and related supportive findings as ipsilateral sublingual gland hypertrophy have helped us in identifying the right SMG aplasia and differentiating it from enlarged submandibular lymph node. The patient was further planned for adjuvant radiation therapy with chemotherapy (RT + CT) and treatment was underway.

Discussion

Normally, the major salivary glands develop from oral ectoderm proliferation and outpouching during the fourth to eighth weeks of fetal development. The aplasia (agenesis) of SMG occurs due to defects during fetal development [3]. This condition is related to the first and second branchial arch abnormalities. It is also seen in genetic syndromes such as Treacher-Collins syndrome, hemifacial microsomia, ectodermal dysplasia, and lacrimoauriculo-dento-digital syndrome (LADD) [4]. However commonly, unilateral absence of SMG is an isolated condition without any abnormalities [8].

The SMG agenesis is commonly diagnosed incidentally during imaging. The bimanual clinical examination of the neck is insufficient for definitive diagnosis. The various imaging modalities can be useful such as sialography, ultrasound, CT, MRI, and nuclear medicine (technetium T99m pertechnetate scintigraphy) imaging. Ultrasound is an initial imaging investigation but it is difficult to interpret. Next, the scintigraphy gives the information only about the presence of functioning salivary tissue. For a better analysis, the cross-sectional imaging as CT or MRI is preferred and it gives the complete anatomy of salivary glands. However, among all the imaging modalities, the modality of choice for the diagnosis of salivary gland agenesis can be MRI [1, 3, 4, 7, 8, 10–13].

In our patient, neither family history nor facial or other abnormalities were reported. In general, absence of major salivary gland may lead to decrease in saliva causing symptoms such as dryness of mouth, dental caries, and difficulty in swallowing [9]. However, our patient has not shown any such symptoms on presentation explaining the adequate flow of saliva by the other gland present.

In most of these SMG aplasia patients, a contralateral SMG hypertrophy will be present as pseudomass [3–5, 9, 14]. In our patient, the contralateral SMG is normal size with normal signal intensity. However, the enlargement of ipsilateral sublingual gland was noted and such hypertrophy was well identified on MRI over CT. Presence of such ipsilateral sublingual gland hypertrophy itself helped us in ascertaining the absence of SMG. In addition, the right parotid gland was also observed to be atrophic. Overall on MRI, a well-defined round homogeneously enhancing lymph node in submandibular region (level IB) was seen occupying the right submandibular region fat without an SMG.

The lymph node differentiation from normal gland was truly difficult on palpation without an MRI. Even in our patient, these preliminary MRI images might have been overlooked. But the finding of ipsilateral asymmetric sublingual gland hypertrophy has grabbed our attention for careful evaluation. Which lead to identification of right SMG aplasia, avoiding confusion with single enlarged right submandibular metastatic lymph node (right level IB). The final surgical histopathological report has also confirmed and reaffirmed the absence of right submandibular gland. In 28 dissected lymph nodes, only 1 lymph node was reported to be metastatic as seen on MRI imaging.

In general, on imaging modalities like MRI, we observe linear T2 weighted high signal intensity showing the ductal system in SMG differentiating from the lymph node. Also the metastatic lymph node will show a heterogeneous post-contrast enhancement due to necrosis, where an SMG will be showing a homogenous enhancement. Even on DWI, metastatic lymph nodes will show a restricted diffusion which would be absent in normal SMG.

From this case report, we strongly suggest that MRI can be highly beneficial in identifying and solving such conditions. However, a good understanding of submandibular space anatomy and awareness about this rare entity is necessary for this purpose. The familiarity with these imaging findings can help in optimum preoperative staging and treatment planning.

Conclusions

We strongly suggest and recommend all the clinicians and radiologists to get familiarized with this rare condition of SMG aplasia and its imaging features as absent SMG and associated findings as hypertrophy of contralateral SMG or hypertrophy of sublingual glands. The imaging modalities such as MRI or CT are highly beneficial for diagnosis of this condition. In general treatment of this condition is supportive and reassuring. In oncology, especially in oral cavity carcinoma, the level IB (submandibular lymph node) is a common site of lymph node metastases and this may masquerade as normal SMG. Hence, it is very important to avoid this diagnostic pitfall to avoid impact on preoperative staging and treatment planning.

Abbreviations
LADD: Lacrimoauriculo-dento-digital; SMG: Submandibular gland; MRI: Magnetic resonance imaging; CT: Computed tomography; LDCT: Low dose screening of computed tomography; MND: Modified neck dissection; IJV: Internal jugular vein; ALT: Anterolateral thigh; ECE: Extracapsular extension; RT: Radiation therapy; CT: Chemotherapy; AP: Anteroposterior; TR: Transverse; CC: Craniocaudal; TNM: Tumor, nodes, and metastases; STIR: Short tau inversion recovery; DWI: Diffusion weighted imaging; ADC: Apparent diffusion coefficient.

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Author contributions
MT was involved in the concept. MT and RN were involved in supervision and material collections. Data collection and/or processing, design, literature search and critical review were performed by MT and VRY. MT, VRY and RN were involved in analysis and/or interpretation. VRY was involved in writing the manuscript. All authors have read and approved the manuscript.

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Availability of data and materials
Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.

Declarations

Ethics approval and consent to participate
The manuscript has got an ethical review exemption from the Ethical review committee of our hospital as case reports are exempted from review according to our ERC's policy; written informed consent was obtained from the patient for publication of anonymized images.

Consent for publication
A written consent was obtained from the patient for publication of data and anonymized images.

Competing interests
The authors declare that they have no competing interests.

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