

Aplasia of bilateral submandibular glands with compensatory hypertrophy of bilateral sublingual glands: A case report

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Abstract

Congenital aplasia of bilateral submandibular glands is a rare clinical condition with only a few cases reported in literature. Patients can either be asymptomatic or present with dryness of mouth, dental caries, gingival problems or difficulty in chewing/swallowing. Aplasia of submandibular glands may be associated with compensatory hypertrophy of rest of the salivary glands, which may present as palpable masses. We report the case of a 12-year-old female with bilateral submandibular gland aplasia and hypertrophy of bilateral sublingual glands, presenting as a palpable mass demonstrated by magnetic resonance imaging (MRI) and

ultrasonography (US). It is important to be aware of this rare entity to avoid unnecessary biopsy, provide reassurance and conservative treatment, if necessary to the patient.

Keywords: Aplasia, compensatory hypertrophy, case report, sublingual gland, submandibular gland.

Introduction

The major salivary glands of human beings begin developing between the sixth and seventh weeks of gestation. The parotid glands are the first to develop which arise from the ectodermal lining of the stomodeum. The submandibular glands develop a short time later and arise from the endodermal layer of the

floor of the stomodeum.[1] Congenital absence of the submandibular gland is a rare clinical entity. Usually, this condition is bilateral. It may also be found associated with other development anomalies of the head and neck. The first case of bilateral submandibular gland aplasia was presented in 1885 by Gruber.[2]

Patients may be asymptomatic clinically or may present with various symptoms. The common presenting symptoms are found to be dryness of the mouth, difficulty in chewing and swallowing and dental caries. Compensatory hypertrophy of the contralateral or other major salivary glands, or accessory salivary tissue, can result in clinical and or radiological pseudo masses.[3]

In this report, we describe the US and MRI findings of a 12-year-old girl with bilateral submandibular gland aplasia associated with sublingual gland hypertrophy and herniation of left sublingual gland through a defect in the mylohyoid muscle, presenting as a palpable mass.

Case report

A 12-year-old female was referred to our department for evaluation of a painless swelling in the left submandibular region over the last twelve months. She had no associated symptoms such as dry mouth, dysphagia, teeth and gum problems. There was no other relevant medical history and no family history of similar complaints. On physical examination, a firm non tender swelling was palpable in the left submandibular region, measuring 3 x 3 cm. She was referred to our department for further evaluation.

Magnetic resonance imaging (Fig. 1-4) showed absence of bilateral submandibular glands. The right sublingual gland appeared elongated and extending into the ipsilateral submandibular area, posterior to the right mylohyoid muscle. The left sublingual gland was hypertrophied and seen herniating through a defect

(measuring 1.8cm) in the mylohyoid muscle and extending laterally to present as a palpable external lump in the left submandibular region. The hypertrophied sublingual glands were homogeneous with distinct margins and normal post contrast enhancement. There was no radiological sign of inflammation or tumor. Few benign cervical lymph nodes were seen in the left submandibular region.

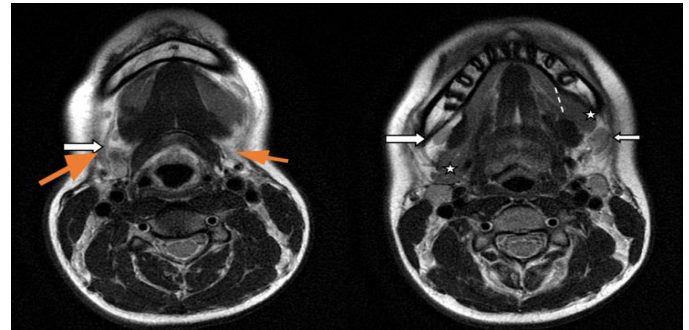


Figure 1: Axial T2W MRI images showing (A) Absence of bilateral submandibular glands. Only fat, facial vessels (orange arrows in (a)) and lymph node (white arrows in (a, b)) present at the expected location for submandibular glands. (B) Hypertrophied right sublingual gland (star) extending into right submandibular area and hypertrophied left sublingual gland (star) herniating through ipsilateral anterior mylohyoid muscular defect (dotted line in (b)).

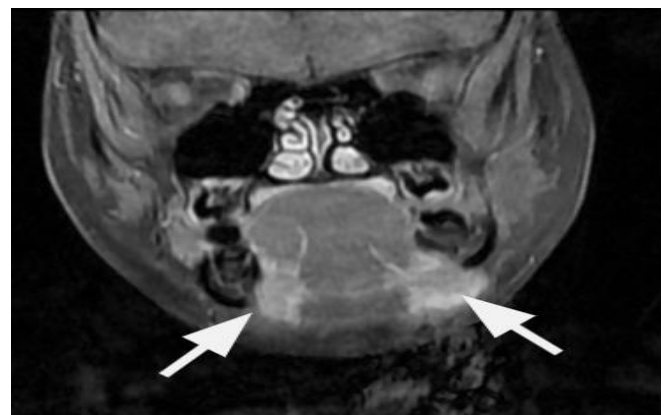


Figure 2: Coronal contrast enhanced T1FS MR image showing bilateral hypertrophied sublingual glands (arrows)

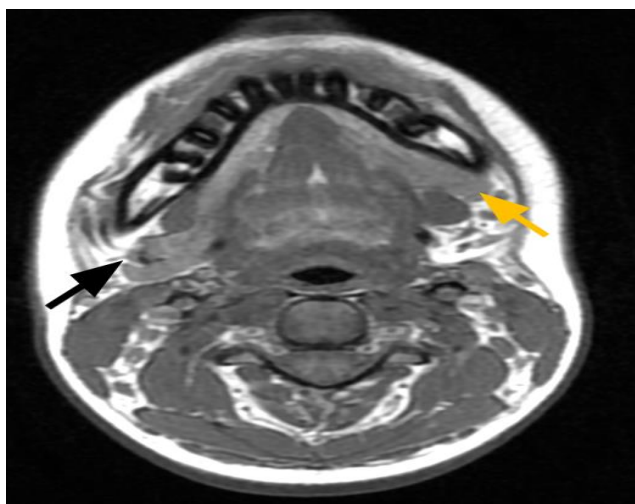


Figure 3: Axial T1W MRI images showing hypertrophied right sublingual gland (black arrow) extending into the right submandibular area and hypertrophied left sublingual gland (yellow arrow) herniating through ipsilateral anterior mylohyoid muscular defect.

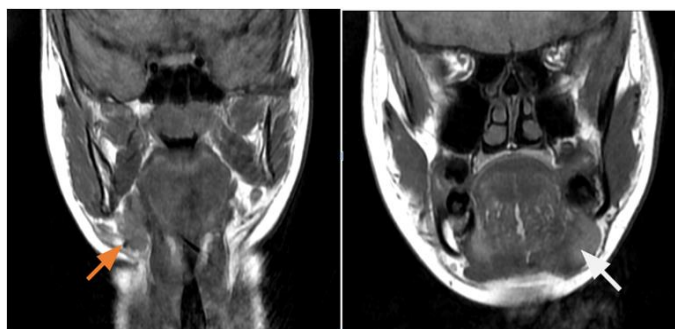


Figure 4: (A, B) Coronal T1W MRI images showing hypertrophied right sublingual gland (orange arrow) extending into the right submandibular area and hypertrophied left sublingual gland (white arrow) herniating through ipsilateral anterior mylohyoid muscular defect.

Correlative ultrasonographic examination of the head and neck area (Fig. 5, 6) confirmed that bilateral submandibular glands were completely absent. The other major salivary glands were present and there was compensatory hypertrophy of bilateral sublingual glands (Left>right). The right sublingual gland was seen extending into the ipsilateral submandibular fossa. The

left sublingual gland was seen to herniate through a defect in the ipsilateral mylohyoid muscle and presenting as a palpable swelling externally. Only a benign cervical lymph node was present in the left submandibular fossa.

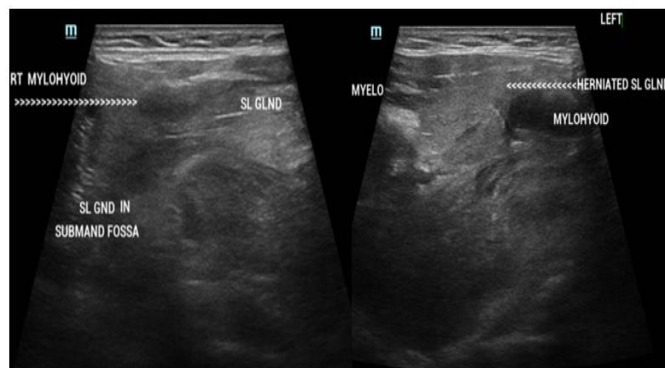


Figure 5: Transverse sonograms showing (A) the enlarged right sublingual gland extending into the submandibular fossa(B) Enlarged left sublingual gland herniating through defect in mylohyoid.

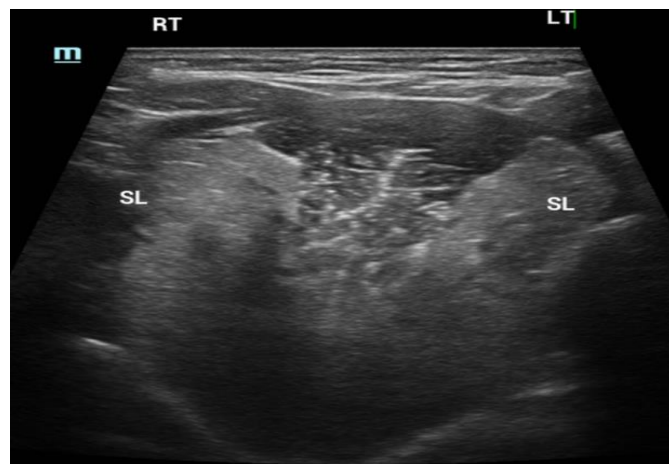


Figure 6: Transverse sonogram showing bilateral hypertrophied sublingual glands.

Discussion

Major salivary gland aplasia is a relatively rare disorder. The term “aplasia” is described as the total or partial agenesis of the gland.⁴ Any of the three major groups of salivary glands may be congenitally absent. The cause of the aplasia is unknown, but likely results from a disturbance during fetal development.[1] There is no sex

predilection.[5]. The age of diagnosis does not correspond with the age of onset since this is a congenital condition.

Aplasia of the major salivary glands can be seen in several genetic syndromes or in association with developmental anomalies, such as Treacher Collins syndrome, lacrimo-auriculo-dento-digital (LADD) syndrome, or aplasia of lacrimal and salivary glands (ALSG).[1]

The diagnosis of salivary gland aplasia can be made with a variety of imaging techniques, the most common of which are CT, MRI, and technetium-99m pertechnetate nuclear medicine studies.[3]

CT and MRI demonstrate the absence of a salivary gland, with the glandular beds replaced by fat.[4] Compensatory hypertrophy of salivary glands can be seen with gland aplasia and should be considered if gland enlargement is found on physical examination.[6] Mylohyoid defect or boutonnière can provide a gap for hypertrophied sublingual gland to prolapse into the submandibular space. Herniation of the hypertrophied sublingual salivary tissue through the mylohyoid muscular defect can present as a submandibular mass. This rare entity was reported in literature by Ahmed et al. [7] They reported a case of non-syndromic bilateral submandibular gland aplasia with hypertrophied sublingual salivary tissue herniating through the mylohyoid boutonnière to present as a palpable mass on the left side with corresponding CT findings.

Differential diagnoses of submandibular swellings include malignant and nonmalignant swellings. Nonmalignant swelling can be caused by mumps, sialadenitis, Sjogren syndrome, cysts, infections and submandibular lymphadenopathy. The neoplastic growths in the submandibular area may include tumors

of the submandibular gland, the tail of the parotid gland, Hodgkin's disease, non-Hodgkin's lymphomas and metastatic disease.[8]

Conclusion

We have reported a case of agenesis of bilateral submandibular gland aplasia with compensatory hypertrophy of sublingual glands (L>R) mimicking a mass, confirmed by a combination of MRI and US. It is important for radiologists to be familiar with this anomaly to avoid unnecessary biopsy, provide reassurance to the patient and supportive treatment, if symptomatic.

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