ejpmr, 2016,3(6), 463-465

SJIF Impact Factor 3.628

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Report

ISSN 2394-3211

EJPMR

INCIDENTALLY DETECTED UNILATERAL SOLITARY NON-SYNDROMIC SUB-MANDIBULAR GLAND APLASIA – A CASE REPORT

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Article Received on 13/04/2016 **Article Revised on 03/05/2016** **Article Accepted on 23/05/2016**

ABSTRACT

Major salivary gland aplasia, or hypoplasia is a rare ailment with few cases reported in literature. It can affect submandibular, parotid and sublingual glands. It can be complete or partial, unilateral or bilateral. The patient's presentation varies from being asymptomatic to dry mouth (xerostomia), taste alteration, oral burning, difficulty in chewing and swallowing to increased incidence of caries. Prompt diagnosis and treatment strategy can prevent or minimize additional dental damage and dysphagia. Patients with salivary gland hypoplasia may or may not be symptomatic and therefore diagnosed incidentally during imaging. In this case we report a 70 year old male patient with complaints of swelling below the mandible and dysphagia who was examined on ultrasonography and computed tomography leading to diagnosis of carcinoma tongue with metastatic cervical lymph nodes and right submandibular aplasia.

KEYWORDS: Submandibular gland (MeSH unique ID: D013363). Aplasia of Lacrimal and Salivary Glands (MeSH unique ID: C562407). Lacrimoauriculodentodigital syndrome (MeSH unique ID: C538132). Agenesis (MeSH unique ID: Q000002).

INTRODUCTION

Congenital absence of submandibular gland is rare. Initially, it was described by GRUBER in 1885 which was bilateral submandibular gland aplasia.[1] However, unilateral affection of submandibular gland is also noted. It has an unknown etiology, conversely, a likely possibility of foetal development defect is to be considered.[2] Clinically, syndromes like lacrimo-auriculo-dento-digital syndrome & mandibulofacial dysostosis are also seen.[3]

Developmental anomalies of major salivary gland include total absence of major salivary gland (aplasia) or salivary hypofunction associated with reduced glandular tissue (hypoplasia). Any of the major glands may be absent unilaterally or bilaterally.[4]

Clinical presentation varies from being asymptomatic to dry mouth, difficulty in chewing & swallowing to increased incidence of caries.[5]

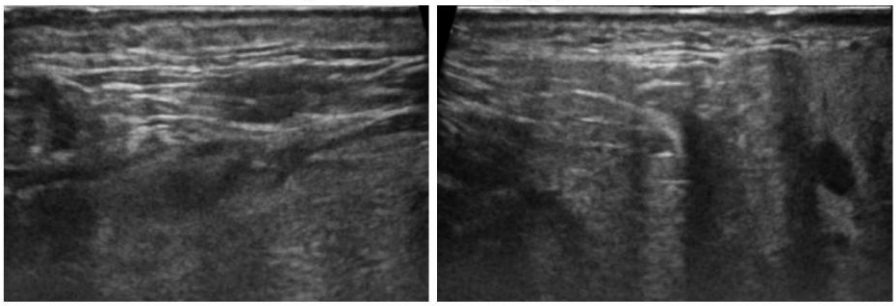
We report a case of an incidentally detected unilateral aplasia of right submandibular gland demonstrated on ultrasonography and computed tomography.

CASE REPORT

A 70 years old male patient was referred for ultrasonography for the complaints of swelling below the mandible & dysphagia. On examination, there was asymmetry in the sub-mandibular region.

USG neck was done with linear high frequency transducer (range 6-10 MHz). USG showed multiple eccentrically enlarged, round, sharply defined, heterogeneously hypoechoic lymph nodes with loss of echogenic hilum & mixed peripheral vascularity along the jugular chain on both sides of midline. No evidence of intranodal calcification was noted. Common carotid artery & internal jugular vein on either side appeared normal. Right submandibular gland was not visualized. Left submandibular gland appeared mildly bulky and otherwise normal (**Figure 1**). Parotid glands appeared normal. No ductal dilatation, calculus was noted. Both masseter muscles appeared normal. Both orbits & lacrimal glands appeared normal.

Figure 1 - Ultrasonography

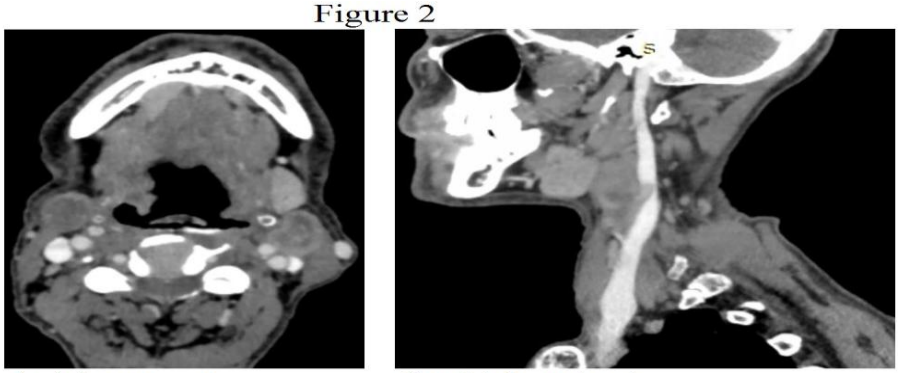


Absent right submandibular gland Present left submandibular gland

Figure 1: USG submandibular region showing absent right submandibular gland and normal left submandibular gland.

CT scan of neck revealed absence of right submandibular gland. Left submandibular gland appeared normal. Heterogeneously enhancing lymph nodes with few of them showing central hypodense areas of necrosis noted at level II-a on both sides and level III on left side (**Figure 2**). At higher sections through oropharynx, an ulcerated solid heterogeneously enhancing lesion involving posterior third of tongue on both sides of midline, lingual tonsil and left palatine tonsil was noted with extension to the vallecula on the left side of midline (**Figure 3**). This was suggestive of neoplastic growth, which was later confirmed on cytology to be squamous cell carcinoma.3D CT scan of maxilla-facial region revealed normal maxilla and mandible (**Figure 4**).

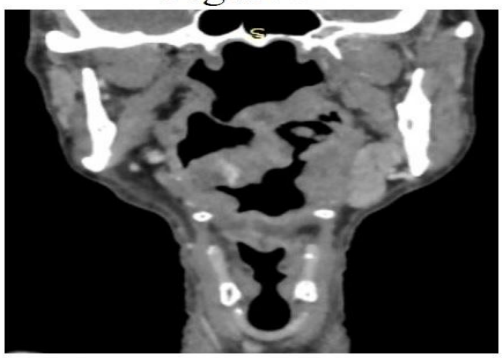
Figure 2



Axial contrast enhanced CT Sagittal contrast enhanced CT

Figure 2: Axial and sagittal contrast enhanced CT scan showing absent right submandibular gland along with normal left submandibular gland. Heterogeneously enhancing necrotic lymph nodes at level II-a on both sides.

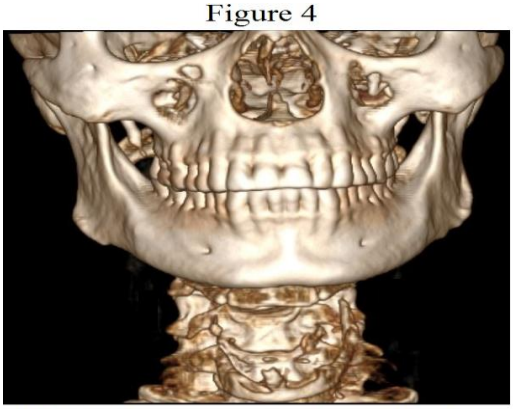
Figure 3



Coronal contrast enhanced CT

Figure 3: Coronal contrast enhanced CT showing ulcerated solid heterogeneously enhancing lesion involving lingual tonsil and left palatine tonsil with extension to the vallecula on the left side of midline.

Figure 4



3D CT showing normal maxilla & mandible.

Figure 4: 3D CT scan of maxilla-facial region revealed normal maxilla and mandible.

Our case is unique as other salivary glands were unaffected and maxillary-mandibular bones were normal and there was no indication of a similar condition in the family. Dental specialists should be mindful of unusual patterns of dental breakdown that could be a consequence of salivary gland agenesis.

DISCUSSION

Congenital absence of submandibular was initially described by GRUBER in 1885 which was bilateral submandibular gland aplasia.[1] Congenital absence of salivary glands is rare and commonly affects multiple major salivary glands.[6] Unilateral aplasia of the salivary gland is rare with only few cases reported.[6] Etiology is not known, the foetal developmental defect is to be considered.[2]

Aplasia of major salivary gland is infrequent with an incidence of 1 in 5000 births. It may be hereditary or syndromic, familial with or without associated syndromes. Unilateral aplasia of submandibular gland is an extremely rare disorder, only 15 cases have been reported. In 75 % of cases submandibular aplasia occurred on the right side.[7]

Salivary gland development takes place during 4^{th} to 12^{th} week of embryonic development.[8] Salivary glands arise at specific locations from the epithelial lining of the oral cavity as primordial buds which extend into underlying mesenchymal tissue as a solid core of cells. Furthermore, branching occurs, leading to formation of ducts with lumina. Glands are divided by surrounding adjacent mesenchyme ultimately resulting in lobulated structures with a capsule. Parotid gland appears at 4^{th} week of gestation. Submandibular gland appears at 6^{th} week of gestation and sublingual glands appear at 9^{th} week of gestation.[5]

Aplasia occurs due to arrest in organogenesis and may be pleomorphic autosomal dominant disorder. Functional innervation of salivary gland is essential for proper growth and maintenance of the salivary gland structure. Developmental basis with sympathetic denervation is suspected as etiology of agenesis.[8]

It may be associated with anomalies like first branchial arch defects in the Treacher-Collins syndrome (mandibulofacial dysostosis), orbital abnormalities (lacrimal hypoplasia, canalicular atresia and absence of lacrimal puncta).[9] It can also be a part of lacrimo-auriculo-dento-digital syndrome characterized by hypoplasia, aplasia or atresia of lacrimal system, deafness and ear abnormalities, dental & digital anomalies.[6] 1^{st} & 2^{nd} branchial arch syndrome or occulo-auriculo-vertebral spectrum has been noted as an associated condition.[10]

Less frequently, aplasia occurs as an isolated phenomenon.[11] Our patient presented with symptoms of swelling below mandible, dysphagia without any other symptoms related to gland aplasia. His relative lack of salivary gland related symptoms was presumably due to the unilateral nature of aplasia with other major salivary glands being present.

Intra-oral examination should be done thoroughly which includes mucosal and dental status as well as salivary flow. Early diagnosis is essential to minimize both decay, periodontal disease, swallowing difficulties, taste alterations and oral burning.[7]

Salivary gland aplasia can be diagnosed using various techniques like USG, CT, MRI and nuclear medicine.[6] Associated abnormalities of orbit, maxilla and mandible can also be detected.[7] Information regarding presence/ hypoplasia/ agenesis of the contralateral salivary gland can also be obtained.[7]

CONCLUSION

Unilateral submandibular gland aplasia is a rare entity. However, accurate incidence of the condition is often difficult to determine in patients who are asymptomatic. Timely diagnosis is of utmost importance to minimalize complications of periodontal disease, tooth decay and swallowing difficulties. Examination of these patients should be carried out using ultrasonography to assess other major salivary glands and computed tomography/ magnetic resonance imaging to rule out associated maxilla, mandibular and orbital abnormalities.

ACKNOWLEDGEMENT

We would like to thank the patient described for his cooperation, understanding and humble attitude towards the process of investigations and department of pathology in Dr. DY Patil Medical College Pune for advice and performing aspiration cytology.

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