Bilateral Submandibular Gland Aplasia: A Case Report and Review of the Literature

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The congenital absence of major salivary glands, particularly the submandibular gland, is a rare condition. Although the etiology of aplasia is unknown, it is thought to occur owing to defects that emerge during early fetal development. Agenesis of 1 or more of the major salivary glands may occur alone or in association with other congenital anomalies. Very few cases of bilateral submandibular gland aplasia have been reported in the literature. Patients with this condition can be either symptomatic or asymptomatic. Due to the probability of there being additional anomalies, patients and their families should be carefully evaluated. We present the ultrasound and computed tomography findings for a case of bilateral submandibular gland aplasia that was detected incidentally. A review of the literature on major salivary gland aplasia was also conducted. [*P R Health Sci J 2022;41(3):172-175*]

Key words: Submandibular gland, Salivary gland, Aplasia, Ultrasound, Computed tomography

Submandibular gland aplasia is a very uncommon abnormality. Approximately 40 cases with the congenital absence of the submandibular gland have been reported in the literature. The first case was presented in 1885 and was a bilateral submandibular gland aplasia (1). Patients may be asymptomatic or may present with complaints of dry mouth (xerostomia), difficulty in chewing and swallowing, or both (2).

In this report, we present the ultrasound (US) and computed tomography (CT) findings of congenital isolated bilateral submandibular gland aplasia with no congenital or developmental disorder observed. Furthermore, a pertinent literature review was conducted. In addition, for the first time, we present a 3D, volume-rendered CT image of this anomaly.

Case Report

A 33-year-old female patient was admitted to the otorhinolaryngology outpatient clinic for a thyroid nodule follow-up. Her medical history was unremarkable except for said thyroid nodule. Her physical examination and laboratory findings were within normal limits. The patient was referred to the radiology department for a US examination, which showed bilateral absence of the submandibular glands (Figure 1). We confirmed the absence of the bilateral submandibular glands by checking a previous CT image of the patient, which scan had been performed for a different reason (Figures 2, 3). Radiological imaging showed adipose tissue, the facial vein and artery, and lymph nodes in the bilateral submandibular gland space instead of the glands. The patient was diagnosed with congenital isolated bilateral submandibular gland aplasia.

Discussion

Aplasia has been defined as the defective development or congenital absence of tissue or an organ. To be relevant to our case, aplasia would be more specifically described as the total or partial agenesis of 1 or more of the salivary glands (3). Embryologically, the major salivary glands (MSGs) develop from the ectoderm (2). The congenital absence of 1 or more of the MSGs (parotid, submandibular and sublingual) is a rare disorder. The parotid gland is more affected than the submandibular gland is. Agenesis of the MSGs can be an isolated finding or may be part of a syndrome, and it can occur unilaterally or bilaterally. Aplasia can affect more than 1 of the MSGs (4, 5). The absence of MSGs likely results from disturbances of the first and second branchial arches that take place during fetal development (3).

The submandibular gland is the second largest MSG and is located in the submandibular triangle (space). Anatomically, the submandibular gland has a close relationship with the facial vein and artery, in that it partly envelops the mylohyoid muscle, which itself serves to divide the submandibular gland into a superficial lobe and a deep lobe. The duct of the submandibular gland, the Wharton's duct, opens into the floor of mouth on the sublingual papilla (6, 7). Although the etiology of submandibular gland aplasia is unclear, it is claimed to be a defect of MSGs and oral

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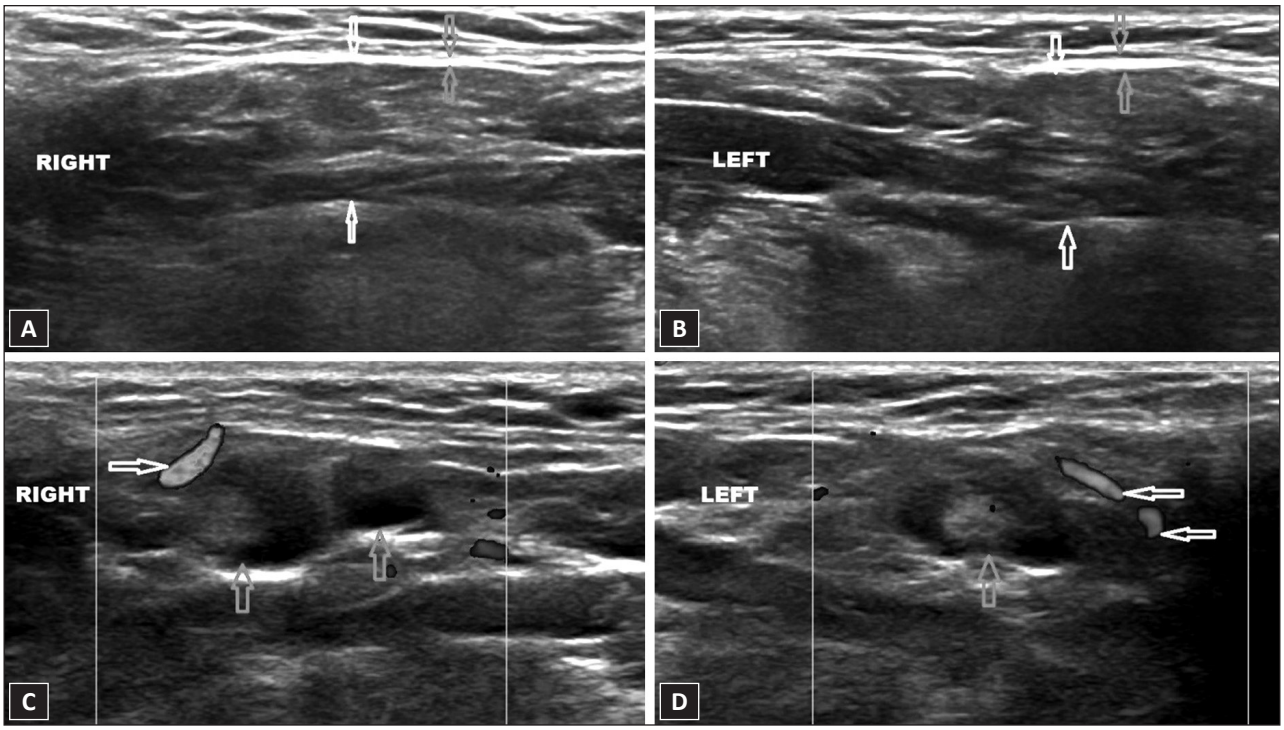

Figure 1. A and B. Gray scale US images reveal that the bilateral submandibular glands are absent under the superficial fascia (between the grey arrows), and fatty tissue (between the white arrows) has replaced the bilateral submandibular glands. C and D. Color doppler US images show facial vessels (white arrows) and lymph nodes (grey arrows) in the bilateral submandibular space.

ectoderm proliferation and migration that occurs from the 4th to the 8th weeks of fetal development (8). The exact incidence of MSG aplasia is not known due to the asymptomatic nature of most of the cases (2). MSG aplasia may be hereditary or syndromic and has an incidence of 1/5000 births. The absence of MSG may be seen as part of a clinical syndrome, such as oculo-auriculo-vertebral spectrum (hemifacial microsomia), mandibulofacial dysostosis (Treacher Collins syndrome), lacrimo-auriculo-dento-digital syndrome (LADD syndrome, Levy-Hollister syndrome), and ectodermal dysplasia associated with agenesis or dysplasia of lacrimal or thyroid glands. Rarely, MSG aplasia develops without any familial history and may exist with no associated anomalies (1, 3, 9–11).

Patients with this condition can be either symptomatic or asymptomatic. The absence of symptoms is probably due to the secretions of other salivary glands that compensate for this

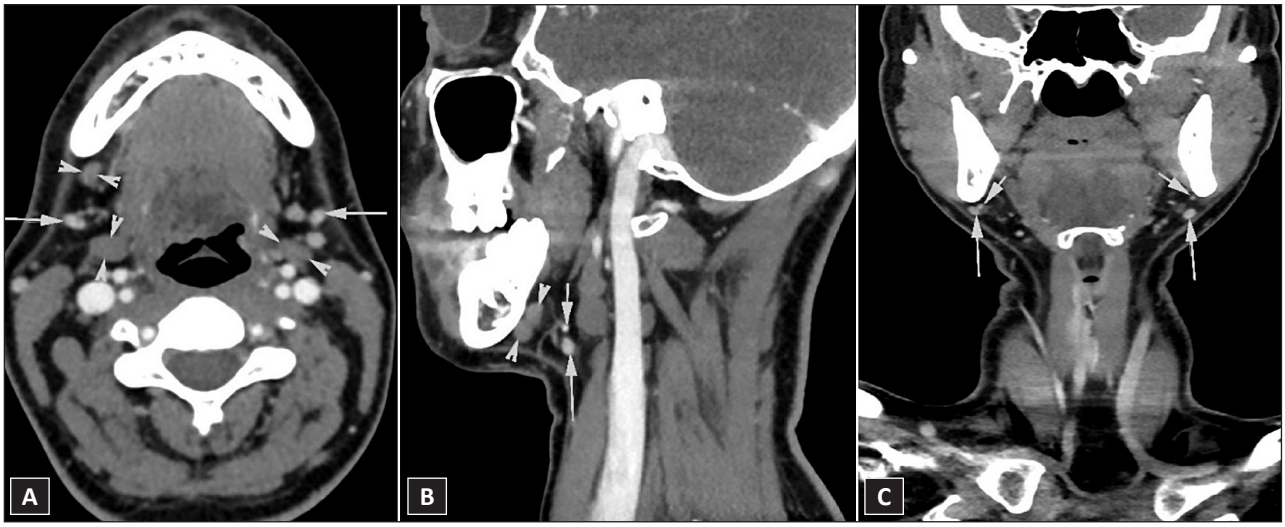

Figure 2. A, B, and C. Axial, sagittal, and coronal intravenous contrast-enhanced CT images show absence of the bilateral submandibular glands. Lymph nodes (arrowheads), facial veins (long arrows), facial arteries (short arrows), and fatty tissue are seen in the bilateral submandibular space.

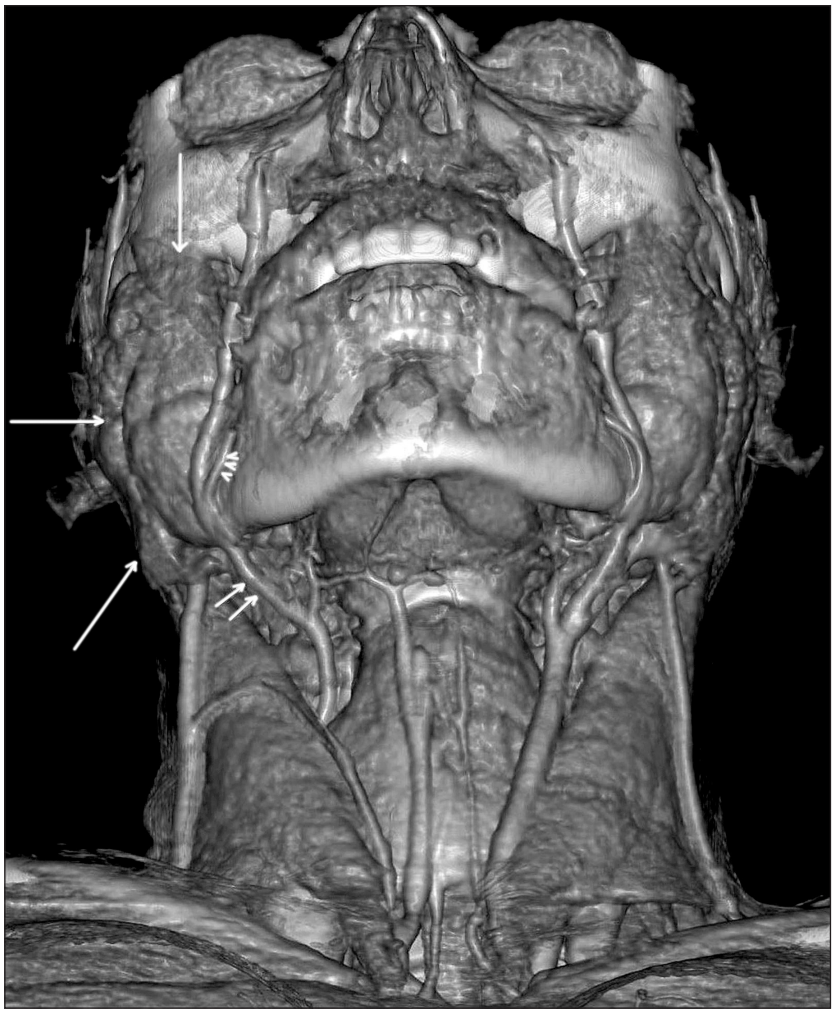

Figure 3. Intravenous contrast-enhanced 3D coronal volume-rendered CT image depicts absence of the bilateral submandibular glands. Parotid glands (long arrows), facial veins (short arrows), and facial arteries (arrowheads) are seen as normal.

deficiency. The symptoms of patients with MSG aplasia include dry mouth, difficulty chewing, occasional bouts of dysphagia, and an increased incidence of dental caries, all of which can accompany the decreased production of saliva; these patients may also complain of a neck mass due to the compensatory hypertrophy of the other MSGs. Common consequences of decreased saliva production are substandard oral self-care habits, increased incidence of opportunistic infection, angular cheilitis, impaired taste, and gingival problems. Given these symptoms, agenesis of the MSGs can be suspected on clinical examination. That being the case, the clinician must evaluate the Wharton duct and its opening, though it is possible that neither will be observable if there is total aplasia. The diagnosis of MSG aplasia is usually made radiologically. Ultrasound, CT scanning, magnetic resonance imaging (MRI), sialography, and Tc-99m pertechnetate scintigraphy can be used for said diagnosis. Of these methods, US is highly sensitive, inexpensive, does not expose the patient to radiation, is noninvasive, and is widely available. Magnetic resonance imaging can be used to confirm the diagnosis because of its advantage of not exposing the patient to ionizing radiation and its ability to characterize soft tissue. The characteristic finding on imaging is the nonvisualization of the submandibular gland, which is often replaced by fatty tissue and lymph nodes. Nevertheless, many radiologists may prefer MRI and CT because of the ability of these tests to precisely document this abnormality. It is also very important to have an experienced radiologist, as some confusion may occasionally occur in cases of unilateral agenesis or compensatory hyperplasia of other glands. Acquired fatty atrophy of the submandibular glands, which can be seen in some conditions such as chronic obstruction and autoimmune diseases, can cause diagnostic difficulties. The treatment is symptomatic and typically includes the use of salivary substitutes. Dental health care should be maintained with preventive care (1–4, 6, 10).

Our case was detected incidentally, as the patient had already undergone a CT scan for another reason, allowing the confirmation of the diagnosis of bilateral submandibular gland aplasia by using a combination of US and CT imaging. There were no bilateral submandibular glands on US or CT. As in previously reported cases, the submandibular space was replaced by adipose tissue, facial vessels and lymph nodes. Compensatory hypertrophy mimicking a mass lesion was not detected in any of the other MSGs. She did not describe any additional problems in subsequent questioning.

In conclusion, clinicians and radiologists should be aware of this rare condition. Submandibular gland aplasia should be kept in mind in patients with symptoms such as xerostomia, dental or gingival pathologies, chewing difficulty, dysphagia or a neck mass (or any combination of the previous). Due to the likelihood of additional anomalies, the signs and symptoms must be carefully evaluated, and an assessment should be made for other possible cases in the family.

Resumen

La ausencia congénita de glándulas salivales mayores, en particular la glándula submandibular, es una condición poco común. Aunque se desconoce la etiología de la aplasia, se cree que se debe a los defectos que surgen durante el desarrollo fetal temprano. La agenesia de una o más de las principales glándulas salivales puede ocurrir sola o en asociación con otras anomalías

congénitas. En la literatura se han descrito muy pocos casos de aplasia bilateral de glándulas submandibulares. Los pacientes con esta condición pueden ser sintomáticos o asintomáticos. Debido a la probabilidad de que existan anomalías adicionales, los pacientes y sus familias deben ser evaluados cuidadosamente. Presentamos los hallazgos ecográficos y de tomografía computarizada de un caso de aplasia bilateral de glándulas submandibulares que se detectó de manera incidental. También se realizó una revisión de la literatura sobre aplasia de glándulas salivales mayores.

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