Isolated unilateral Non -Syndromic non-familial hypoplastic Parotid gland: Case Report

Sanjay M. Khaladkar, Mridul Ayush, Kuldip Chaudhary, Dhaval K. Thakkar.

Dept. of Radio-Diagnosis, Dr. D Y Patil Medical College, Pimpri, Pune 411018

Corresponding Author: Dr. Dhaval K. Thakkar ; Email: drdhaval2308@gmail.com

ABSTRACT:

Major salivary gland agenesis or hypoplasia is a rare and unusual condition with few cases documented in literature. The anomaly can involve parotid, submandibular and sublingual glands. It can be total or partial, unilateral or bilateral. Patients with salivary gland aplasia may suffer from xerostomia which leads to extensive dental demineralization and / or dental caries, several periodontal disease, taste alteration, dysphagia, oral burning. Early diagnosis and a therapeutic strategy can prevent or minimize further dental damage/ decay, periodontal disease and swallowing difficulties. With partial agenesis or hypoplasia of major salivary glands, symptoms may not be significant. Hence patients may be asymptomatic and diagnosed incidentally during imaging studies.

Key Words : Agenesis , Parotid gland, major salivary glands  , LADD Syndrome

Case Report

83 years old female patient was referred for USG neck for pulsatile lesion in right supraclavicular region. USG neck was done with linear high frequency transducer (Range 6 – 10 MHz). USG showed multiple small well defined mixed echoic solid nodules of variable sizes (0.2 to 1 cm) in both thyroid lobes s/o early multinodular goitre. Innominate artery & proximal portion of right subclavian artery appeared ectatic (which was felt clinically as pulsatile lesion in right supraclavicular region). No cervical lymphadenopathy was noted on either side. Common carotid artery & internal jugular vein on either side appeared normal. Right parotid, both submandibular salivary glands appeared normal (fig 1,2,3,4) Left parotid appeared small in size with normal echo texture. No ductal dilatation, calculus or solid/cystic mass noted. Prominent fat was noted in left parotid region (fig 3). Both orbits and lacrimal glands appeared normal. Mandible and maxilla on either side appear normal (fig 7). Same findings were seen on MRI (axial, coronal and sagittal T1WI & T2WI) (fig 8-11). X ray skull AP view (Fig 12) showed normal maxilla, mandible and orbits. The case report is unique in that no other anatomical structures were involved and there was no indication of similar condition in any family member. Dental practitioner should be aware of unusual pattern of dental breakdown that could be a result of salivary gland agenesis.

Discussion

During 4th and 12th week of embryonic development, each of salivary gland is formed at specific locations in the oral cavity through the growth of a bud from the oral epithelium into underlying ectomesenchyme. This epithelial bud grows and is branches forming ducts. The parotid glands appear during 4th week of
gestation between maxillary process & mandibular arch. Submandibular gland appears in 6th week & Sublingual arises during 9th week between tongue & mandibular arch. The aplasia occurs due to arrest in organogenesis, but the exact etiology is unknown/Cryptogenic. It may be pleotrophic Autosomal Dominant disorder. Functional innervations of salivary glands is essential for proper growth and maintainance of salivary gland structure. Parasympathetic denervation results in loss of glandular weight, while sympathetic denervation results in either atrophy of major salivary glands or hypertrophy of others. Developmental basis with sympathetic denervation is suspected as etiology of agenesis.\cite{1}

During normal embryonic development amongst all salivary glands, the parotid glands is the last to become encapsulated. Several structures become embedded within the substance of the salivary gland. Inspite of close approximation of these tissues during embryonic development, the normally embedded structures are not affected by aplasia of salivary gland. Hence, facial nerve with its branches, retromandibular vein, external carotid artery are normal. Accesory parotid tissue is found usually unilaterally in approx. 20% of normal population. Its anatomy is described in detail by Currarino and Votteler.\cite{2}

Aplasia of major salivary gland is uncommon with incidence of 1 in 5000 births. It may be hereditary or syndromic. It may develop in absence of familial history and may exist with no associated anomalies. Aplasia of major salivary gland was first described in 1885 by Gteuber; who reported a case of bilateral absence of submandibular gland. Since then 41 cases of salivary gland aplasia have been documented.\cite{3}

Any of the glands may be involved, however absence of all 4 major salivary glands is the most frequently encountered pattern. Unilateral aplasia of submandibular gland is an extremely rare disorder, only 15 cases have been reported in medical literature to date. The 4 major salivary glands are most often affected. The parotid gland is more often commonly affected than submandibular gland. Unilateral submandibular salivary gland aplasia was first reported by YILMAZ in 1894. In 75% of reported cases the aplasia occurred on right side. Less frequently it appears as an isolated phenomenon as an isolated defect, not associated with other congenital disorders.\cite{1}

In 1879, a first case of parotid aplasia with resultant absence of saliva was described by Bradbury. In 1949, Kabakov described congenital absence of both parotid glands & under development of remaining salivary glands. Unusual cases of unilateral/bilateral parotid aplasia with accessory parotid tissue has also been described.\cite{4}

In 75% of reported cases the aplasia occurred on right side. Less frequently it appears as an isolated phenomenon as an isolated defect, not associated with other congenital disorders. As discussed by YILMAZ et al, Shrinivasan et al, Roh, Mathieson and Hudgins, Ahmed et al, there are few reports of symptoms caused by aplasia of one gland as secretions of other normal glands or compensatory hypertrophy of sublingual glands compensate for it.\cite{1}

Salivary gland may be isolated or in association with other anomalies particularly defect in lacrimal apparatus. It is observed in Lacrimo-Auriculo-Dento-Digital Syndrome, Hemifacial microsomia, mandibulo-facial dysostosis (Treacher-Collin’s Syndrome), multiple facial anomalies, ectodermal dysplasia. It can be a feature in first and second
Branchial arch anomalies. Single case of Bilateral Parotid Agenesis associated with Down’s syndrome, Bilateral agenesis of Parotid glands and unilateral agenesis of Submandibular gland in case of Cleft lip and palate in patients have been described. [3]

Salivary gland hypoplasia and aplasia is not so uncommon in person with oligodontia and ectodermal dysplasia. In cases where both teeth and salivary glands are affected the term oral ectodermal dysplasia may be used. [5] Congenital absence or hypoplasia of major salivary glands may be associated with multiple developmental anomalies especially in face. [6]

Clinically patients may be asymptomatic or may present with dryness of mouth, difficulty in chewing, swallowing and dental caries due to reduction in protective effect of saliva in oral cavity, unusual pattern of dental caries, erosion of teeth, presence of plaques, periodontal disease, chelitis. It may present as incidentally detected unilateral submandibular gland aplasia with ipsilateral sublingual gland hypertrophy mimicking a mass demonstration on CT & MR imaging.

LADD syndrome- lacrimal-aureiculo-dento-digital syndrome also called as Levy Hollister syndrome was first described by Hollister et al in 1973. This rare autosomal dominant syndrome is characterized by malformation of lacrimal apparatus, cup shaped ears, hearing loss, dental anomalies, various kidney anomalies and limb malformation. [7] Aplasia of lacrimal pineta and nasolacrimal duct are also features of LADD syndrome. Hypoplasia, aplasia or atresia of lacrimal system, anomalies of ear, hearing loss, hypoplasia, aplasia or atresia of salivary system, dental anomalies (absent or small peg shaped lateral maxillary incisors and mild enamel dysplasia) and digital malformation including absent hypoplastic, triphalangeal thumb. Anomalies of 2nd, 3rd, 4th or 5th digits and hypothenar areas. [7]

Approximately 20% of normal individual have accessory parotid tissue which lies superficial to masseter muscle and several millimeters to anterior parotid glands. Bilateral parotid aplasia associated with accessory parotid tissue superficial to left masseter muscle. [8]

Goldenhar syndrome-Franceschetti-Goldenhar syndrome also known as fascio-acuriculo-vertebral anomalies (FAV), First and second branchial arch syndrome or occulo-Auriculo-Vertebral (OAV) spectrum. [9]

The oral examination should be done thoroughly which includes close scrutiny of salivary flow, mucosal state and dental status. Early diagnosis of this anomaly is essential to minimize both decay, periodontal disease and swallowing difficulties. Patients with salivary gland aplasia may suffer from xerostomia, dysphagia, taste alteration, oral burning, extensive demineralization and/or dental caries, severe periodontal disease.

With partial agenesis or hypoplasia of major salivary glands, symptoms may not be significant. Hence patients may be asymptomatic and diagnosed incidentally during imaging studies [1].

The true incidence of aplasia or hypoplasia of parotid gland is difficult to ascertain because the condition is often asymptomatic because saliva will also be produced by submandibular, sublingual and minor salivary glands. Hence mucosal dryness will not develop. Clinical suspicious should arise if the papillae of salivary gland ducts are absent. The presence of residual and/or ectopic glandular tissue can be easily excluded on CT / MRI and salivary gland scintigraphy. [10]
Clinical suspicion should arise if the papillae of salivary gland ducts are absent. The presence of residual and/or ectopic glandular tissue can be easily excluded on CT / MRI and salivary gland scintigraphy. The oral examination must be thorough and include close scrutiny of salivary flow, mucosal state and dental status. Hypoplasia of gland is reduced glandular tissue associated with hypofunction. CT and MRI shows absence of glands and replacement with fatty tissues. A salivary flow rate lower than 50% of its normal value is diagnostic. USG, CT and MRI will give information about presence/hypoplasia/agenesis of contralateral salivary gland, bilateral other salivary glands. Associated abnormalities of orbit, lacrimal gland, maxilla and mandible can also be detected.\[11\]

Approximately 20% of normal individual have accessory parotid tissue which lies superficial to masseter muscle and several millimeters to anterior parotid glands. Bilateral parotid aplasia associated with accessory parotid tissue superficial to left masseter muscle.\[8\]

**Figure 1**: USG parotid showing normal sized right parotid gland, hypoplastic left parotid gland with prominent adjoining fat.

**Figure 2**: USG submandibular region showing normal sized bilateral submandibular salivary glands.

**Figure 3**: Axial CT showing hypoplastic left parotid gland with prominent fat. Right parotid gland appears normal.

**Figure 4**: Axial Plain CT showing normal bilateral submandibular salivary glands.

**Figure 5**: USG left parotid region showing hypoplastic left parotid gland with prominent adjoining fat.
**Figure 6**: USG both orbits (A and B) showing bilateral normal sized lacrimal glands.

**Figure 7**: 3D-CT showing normal bilateral orbits, maxilla and mandible.

**Figure 8**: Axial T1WI image showing hypoplastic left parotid gland with prominent fat. Right parotid gland appears normal.

**Figure 9**: Coronal T2WI image showing hypoplastic left parotid gland with prominent fat. Right parotid gland appears normal.

**Figure 10**: Axial T2WI image showing normal bilateral submandibular salivary glands.

**Figure 11**: Axial T2WI image showing hypoplastic left parotid gland with prominent fat. Right parotid gland appears normal.

**Figure 12**: X-Ray Skull AP view showing normal mandible, maxilla and orbits.
References: