Original Article

Congenital agenesis of unilateral parotid gland with ipsilateral type I first branchial cleft anomaly: A rare presentation

Tripti Maithani a,*, Apoorva Pandey a, Seema Acharya b

a Department of E.N.T, Shri Guru Ram Rai Institute of Medical & Health Sciences, Patel Nagar, Dehradun, India
b Department of Pathology, Shri Guru Ram Rai Institute of Medical & Health Sciences, Patel Nagar, Dehradun, India

Received 8 August 2013; accepted 1 November 2013
Available online 23 November 2013

Abstract  Aim: To report a rare case of unilateral parotid agenesis with ipsilateral type I first branchial cleft anomaly.

Material and methods: A case study with special emphasis on the embryology, outlining the complex developmental process of parotid and branchial arches and highlighting the probable reason for development of such anomalies.

Results: The literature states that unilateral parotid agenesis is a rare entity with few reported cases occurring solely or in conjunction with other head and neck anomalies. In our attempts to find its association with first branchial cleft anomalies we found a single reported case of parotid agenesis with type II first branchial cleft anomaly; thus to the best of our knowledge this is first case of unilateral parotid agenesis with ipsilateral type I first branchial cleft anomaly.

Conclusions: Unilateral parotid agenesis is usually asymptomatic and is generally diagnosed when it occurs along with other head and neck conditions.

ª 2013 Production and hosting by Elsevier B.V. on behalf of Egyptian Society of Ear, Nose, Throat and Allied Sciences.

1. Introduction

Congenital agenesis of parotid gland is uncommon. This condition is usually asymptomatic and its diagnosis is generally made in light of accompanying developmental anomalies.3 As parotid agenesis is asymptomatic and goes unnoticed, thus the true incidence of this condition is unknown. Till date about 32 cases of salivary gland aplasia have been reported in the English literature2 and of these there was just one case of parotid agenesis associated with type II first branchial cleft anomaly.3 To the best of our knowledge...
we report the first case of unilateral parotid agenesis associated with type I first branchial cleft anomaly.

2. Case report

A young male 21 years of age presented in our outpatient department complaining of recurrent swelling over left parotid region since 3 years. Swelling was painful and associated with discharge from a pit present on the left side of neck. Patient had undergone incision and drainage for the same 1 year ago. Apart from this there was history of recurrent gingivitis. However there were no complaints of ear discharge, dryness of mouth or eyes. On examination there was a scar mark present in the left parotid region and a sinus present on left infra-auricular region just behind mandibular ramus (Fig. 1a) about 0.5 x 0.5 cm in diameter. On palpation over the parotid region an ill-defined swelling was found, milking of which resulted into purulent discharge from the sinus in the neck. Ear and nose examination was normal. Oral cavity examination revealed multiple tobacco stained teeth with chronic gingivitis. The papillae of right Stensen’s duct and bilateral Wharton’s duct were normal; however papillae of left Stensen’s duct could not be located. Oral cavity mucosa was adequately moist. FNAC from swelling in the parotid region was suggestive of infected cyst with possibility of first branchial cleft anomaly. Sinogram showed pooling of contrast in an irregular cavity. In MRI T1 weighted image revealed complete absence of the parotid gland on the left side with tethering and inward pulling of overlying skin likely due to fibrosis; T2 weighted sequence showed irregular fluid filled cystic area in the parotid region (Fig. 2). Thus a diagnosis of left parotid agenesis with first branchial cleft anomaly was established. Surgical resection of the sinus tract was planned. A standard face lift incision was given including a cuff of the skin around the sinus opening (Fig. 1b). The facial nerve was identified the moment the SMAS flap was elevated. The sinus tract was found to run in between the branches of the facial nerve (Fig. 1c) terminating as cul-de-sac near the cartilaginous part of the external auditory canal, without communicating with the same. The tract was excised Toto (Fig. 1d) and the specimen was sent for HPE. Postoperative period was uneventful. Histopathology report revealed Work’s type I first branchial cleft anomaly. Our patient is asymptomatic 8 months following surgery and is maintaining a regular follow-up.

3. Embryology

Formation of head and neck is a complex process that begins very early in human development and most congenital malformations of head and neck originate as the branchial arches transform into their adult derivatives. The major salivary glands develop embryologically during the 6th to 8th weeks of gestation. The parotid gland is the first to develop followed by submandibular and sublingual salivary glands. Its development begins roughly around 6 weeks in utero, in the 7th week it moves in dorsal and lateral directions to reside in the pre-auricular region and by the 10th week the facial nerve divides the parotid gland into superficial and deep compartments.

The development of glandular tissue in mammals involves interaction of epithelium with underlying mesenchyme to form the functional part of the gland. This epithelial–mesenchymal interaction regulates the initiation, growth and eventual cytodifferentiation within the salivary glands. An unknown disturbance in this process can result in a rare presentation
of agenesis of the salivary glands. This agenesis may be a feature of first and second branchial arch anomalies and occasionally may be seen in mandibulofacial dysostosis as well as hemifacial microsomia. Branchial apparatus begins to form in the second week of fetal life and is completed by the 6th or 7th week. The pathogenesis of first branchial cleft anomalies remains controversial. The proposed theories are incomplete obliteration of fetal branchial arches, pouches or both specifically, the lack of degeneration of cervical sinus of His. Work divided first branchial cleft anomalies into two types based on embryologic criteria. The Work type I cysts are derived from ectoderm and represents a duplication anomaly of membranous external auditory canal, whereas type II cysts are derived from both ectoderm and mesoderm thus involving both the external auditory canal and cartilaginous pinna.

4. Discussion

Parotid agenesis is a rare entity. The first case of parotid aplasia with resultant absence of saliva was described by Bradbury in 1879. Agenesis may be partial or total, unilateral or bilateral and may be associated with other salivary gland agenesis. Unilateral absence of the parotid gland has been reported less commonly than bilateral absences in the literature. This condition may exist alone or in conjunction with ectodermal defects of the first and second branchial arches. The exact etiology and pathogenesis of agenesis has not been established however it is likely due to unknown arrest in organogenesis and represents problems in morphogenesis of the first and second branchial arches. This was probably the reason for the development of a rare case of parotid agenesis with type I first branchial cleft anomaly in our patient.

The majority of patients with unilateral parotid agenesis are asymptomatic due to sufficient production of saliva by other glands. However symptoms may range from facial asymmetry, dry mouth, increased thirst, dental decay, gingival infections and difficulty in wearing dentures. Signs and symptoms of first branchial anomalies include recurrent pre- and post-auricular swelling, a fistula within the external auditory canal presenting as drainage, or a fistula below the angle of mandible. A common occurrence is incision followed by drainage. Our patient presented typically with recurrent swelling over the parotid region with a discharging sinus in the neck and a history of incision and drainage for the same. Apart from this he also had complaints of recurrent gingivitis.

CT or MR imaging is useful in attaining diagnosis of parotid agenesis as well as for evaluation of branchial anomalies. In cases of agenesis MRI typically demonstrates anatomically absent glands with glandular fossae being replaced by fat. Meanwhile branchial anomaly typically demonstrates a low T1 weighted image and a high T2 weighted signal. FNAC is an important adjunct to clinical diagnosis in cases of branchial cleft anomalies. In our case clinical evaluation, FNAC and MR imaging helped in establishing the diagnosis.

Management of agenesis is supportive, including saliva substitutes, mouth washes and maintenance of oral hygiene; while branchial anomalies are best treated by complete surgical excision. Surgical approach to first branchial cleft anomalies must include the possibility of exposure and protection of the facial nerve. In our case complete excision of the tract with preservation of the facial nerve was done and our patient remains symptom free 8 months following the surgery.

5. Conclusion

Unilateral parotid agenesis is rare and its association with type I first branchial cleft anomalies is even rarer. Majority of times unilateral agenesis may go unnoticed as this condition is generally asymptomatic. Salivary agenesis is managed conservatively whereas first branchial cleft anomalies require complete surgical excision of the tract.

References