

## CASE REPORT

# Congenital agenesis of all major salivary glands and absence of unilateral lacrimal puncta: A case report and review of the literature

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### Abstract

Congenital agenesis of the salivary glands is an extremely rare congenital condition, which may cause severe xerostomia, progressive dental caries, and oropharyngeal candidiasis in children. To date, there have been few documented cases of aplasia of the major salivary glands. Congenital agenesis of the salivary glands accompanied by absence of the lacrimal puncta is even more rare. We report a case of a 5-year-old boy with xerostomia and extensive dental caries. Salivary gland imaging with sodium pertechnetate  $^{99m}\text{TcO}_4$  showed bilateral aplasia of the parotid, submandibular, and sublingual glands. The patient was found to lack the left lacrimal puncta through physical examination. Here we describe the clinical presentation, diagnostic essentials, and medical and dental management of the patient.

**Keywords:** *Xerostomia, salivary gland agenesis, dental caries, oropharyngeal candidiasis*

### Introduction

Agenesis of the major salivary glands is an unusual disorder, which is even rarer when accompanied by the absence of the lacrimal puncta. A review of the literature up to 2011 found 35 cases of agenesis of salivary glands worldwide, and only 14 cases reported a comorbid absence of the lacrimal puncta. The patient reported here appears to be the first case with these two conditions in the Chinese population.

### Case report

In July 2011, a 5-year-old boy was referred to the Oral Medicine Department with the chief complaint of xerostomia and tooth decay. He was initially found to have dry mouth at 4 months of age. At that time, he had difficulty swallowing when his mother added solid food to his diet. Subsequently, he developed rampant caries in his newly erupted primary teeth. Most of these were eventually lost.

His medical history was otherwise unremarkable. Her mother had a normal pregnancy and vaginal

delivery. A review of the dental and medical history of his family members, including his older brother, revealed no remarkable findings.

A physical examination revealed a slim boy, appearing younger than his actual age. Oral examination found dry and erythematous oral mucosa, exfoliative lips, and papillary atrophy on the dorsum of the tongue (Figure 1a, b, c). Unstimulated salivary flow rate was 0 ml per 10 min. Most of his primary teeth were missing due to decay, and caries were present in almost all the remaining primary teeth and the early erupted first molars (Figure 2). The patient did not claim dry eyes. The Shirmer's test, break-up time of tear film, and fluorescein eye staining were normal. No signs of keratitis sicca were found. Further ophthalmic examination was recommended and revealed a congenital absence of the right lacrimal puncta.

His complete blood count and erythrocyte sedimentation rate were within normal limits. Immunologic tests (electrophoresis of serum protein, HIV antibody, antinuclear antibody, and anti-SSA and anti-SSB antibodies) were negative. Salivary gland scintigraphy (Figure 3) was performed with

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(Received 7 October 2011; accepted 20 November 2011)

ISSN 0001-6489 print/ISSN 1651-2251 online © 2012 Informa Healthcare  
DOI: 10.3109/00016489.2011.648273



Figure 1. Clinical manifestations on examination of oral mucosa. (a) Dry and exfoliative lips. (b) Dryness and papillary atrophy on the dorsum of the tongue. (c) Reduced saliva secretion on mouth floor. (d) Recovery of papillae on the dorsum of the tongue after antifungal treatment.

intravenous injection of Tc-99m, and imaging was obtained at 5, 10, 20, 30, and 40 min after the injection. Ascorbic acid was administered topically in the mouth as a stimulant of salivary secretion 30 min after injection. There was no uptake bilaterally of the major salivary gland spaces, measured at multiple time points by scintigraphy. This established the diagnosis of congenital agenesis of all major salivary glands.

In addition to multiple caries, this patient was also diagnosed with oropharyngeal candidiasis, based on a smear test and swab culture. Mixed *Candida* species, *Candida albicans* and *Candida tropicalis*, were found, and a nystatin lozenge was prescribed to treat the candida infection. For xerostomia, traditional Chinese medicine (TCM) patent granules were prescribed, which are used in this hospital to treat dry mouth disorders like Sjögren's syndrome, and have been found effective. Meanwhile, oral hygiene instructions were provided to the patient and his parents, and a cetylpyridinium chloride mouth rinse and fluoride toothpaste were given. At 4 weeks later, his dry mouth symptoms had eased significantly. Upon physical examination, his oral hygiene and

the condition of the oral mucosa were much improved and his tongue papillae had recovered (Figure 1d). He was then referred to a pediatric dentist for treatment of dental caries, fluoride supplement, and subsequent oral rehabilitation. The patient was advised in close follow-up appointments every 3 months.

## Discussion

Congenital agenesis of the major salivary glands is a rare disorder, with only 36 reported cases in the literature worldwide up to 2010 [1]. Agenesis of the salivary gland may be partial or total, and the severely affected patients suffer from a dry mouth, extensive dental decay, and difficulty in swallowing and wearing dentures. Therefore, dentists are commonly the first medical professionals they seek for consultation. The condition should be differentiated from Sjögren's syndrome, drug-induced xerostomia, and other causes of dry mouth. The diagnosis can be confirmed by scintigraphy showing the uptake of  $^{99m}\text{TcO}_4$ .

An even more rare condition is congenital agenesis of the salivary glands accompanied by absence of the

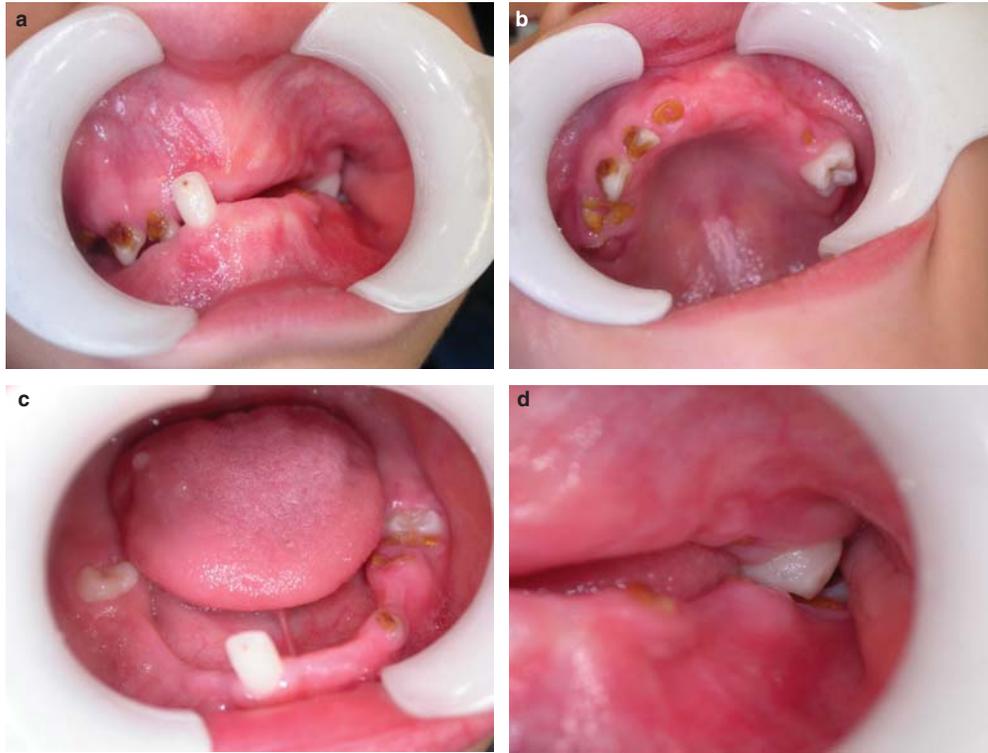


Figure 2. Clinical manifestations on dental examination. (a) Anterior teeth on close bite. (b) Upper teeth missing, residual roots and caries. (c) Lower teeth missing, residual roots, and caries. (d) Relation of posterior teeth on close bite.

lacrimal puncta. Only 14 reported cases of congenital major salivary gland agenesis with lacrimal abnormalities could be found in the literature (Table I) [1–11]. In 1925, Blackmar [2] reported the first case of an 11-year-old girl with congenital absence of the lacrimal puncta associated with an absence of salivary glands. Smith and Smith [3] reviewed 16 cases (11 in living patients) of congenital absence of all four salivary glands in 1977, and found that 3 of the 11 were associated with defects of the lacrimal apparatus.

Organogenesis of major salivary glands and the lacrimal puncta is closely related, which explains the association of such abnormalities. Major salivary glands originate from the proliferation and ingrowth of the oral epithelium between the 4th and 12th week of embryonic life. Agenesis of the major salivary glands may be associated with ectodermal defects of the first and second branchial arches, lacrimal punctum aplasia or hypoplasia and agenesis of the lacrimal gland [6,12]. Although most of the reported cases are isolated forms without family history, two familial forms were described with major salivary gland agenesis and lacrimal abnormalities by Ferreira et al. [8] in 2000 and by Kwon et al. [10] in 2006.

The essential role of saliva in the regulation of the oral microflora, and in defense against infection, is well known. Xerostomia caused by agenesis of major

salivary glands, commonly results in rampant caries and candida infection. Dental treatment for such patients includes prevention and management of dental caries, antifungal drugs for the commonly developed candidiasis, and frequent oral rinses to lubricate the oral cavity and thereby relieve symptoms. Such patients should be instructed in prevention of dental caries, such as dietary counseling, oral hygiene instruction, and the use of a fluoride regimen.

In this case, the patient had early mixed dentition, and all his primary teeth were either lost or reduced to residual roots. Our strategy for his dental management was to rehabilitate salivary function to prevent caries development in his permanent teeth. He was given traditional Chinese medicine granules to relieve his dry mouth. The TCM is a patent medication in our hospital and aimed at promoting circulation and generating body fluid, thereby increasing saliva generation. Additionally, the patient also had oropharyngeal candida infection, diagnosed by his clinical manifestation and laboratory culture, which might worsen his xerostomia symptoms. After topical nystatin for 4 weeks, the erythema of oral mucosa subsided. His oral condition improved. At present, the patient is under close follow-up by a dentist and by an oral medicine specialist.

Agenesis of major salivary glands is rare and can be accompanied by a defect of lacrimal puncta. The case

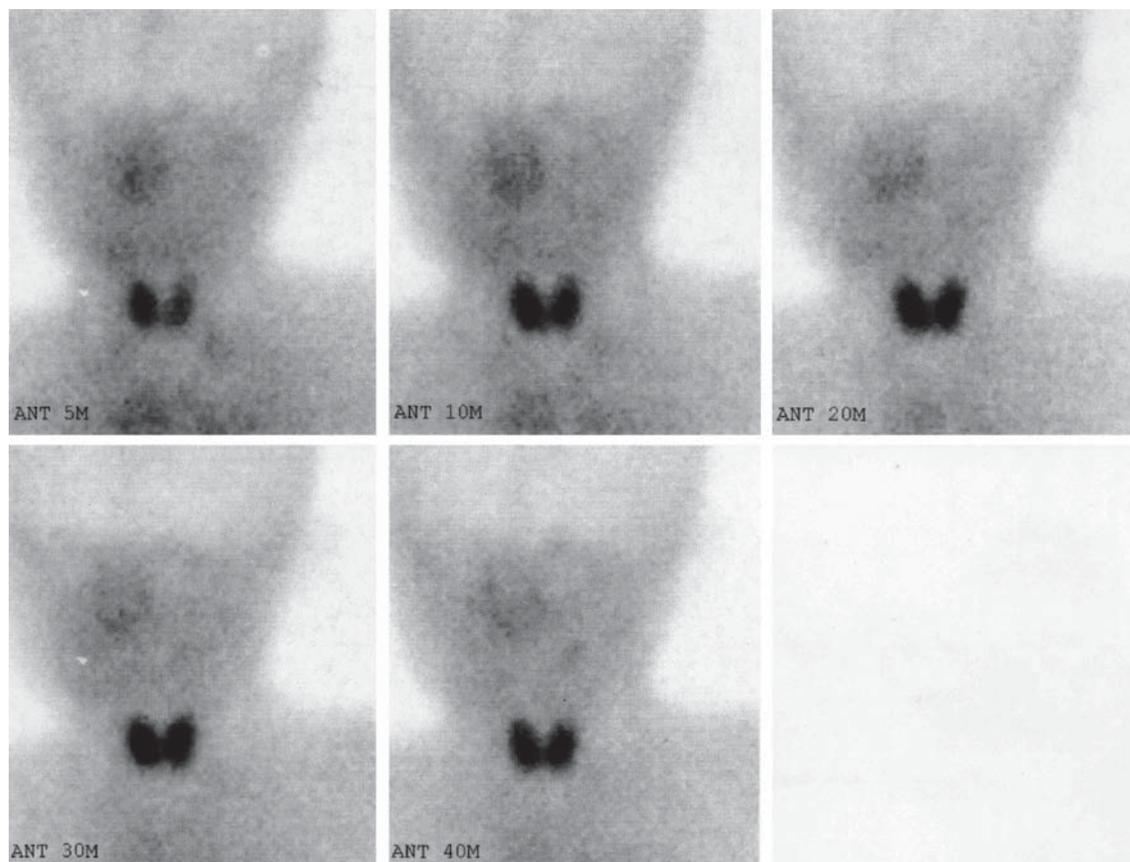


Figure 3. Salivary gland scintigraphy ( $^{99m}\text{TcO}_4$ ). There was no uptake of technetium after 5, 10, 20, 30 (stimulant administration), and 40 min in all major salivary glands.

Table I. Summary of cases of congenital major salivary gland agenesis with lacrimal abnormalities reported in the literature.

Type of salivary gland agenesis	No. of cases	Accompanying abnormality	Family history	Authors
Congenital absence of major salivary glands	1	Absence of the lacrimal puncta	No	Blackmar 1925 [2]
Congenital absence of all four salivary glands	3	Defects of the lacrimal apparatus	No	Smith & Smith 1977 [3]
Aplasia of all the major salivary glands	1	Lacrimal gland hypoplasia and enamel hypoplasia.	No	McDonald et al. 1986 [4]
Congenital absence of all major salivary glands	1	Absence of all four lacrimal puncta	No	Higashino et al. 1987 [5]
Congenital absence of parotid glands	1	Absence of lacrimal puncta	No	Gomez et al. 1998 [6]
Congenital absence of the major salivary glands	1	Impaired lacrimal puncta	No	Myers et al. 1994 [7]
Congenital absence of salivary glands	1	Absence of lacrimal puncta	Five family members affected	Ferreira et al. 2000 [8]
Absence of both parotid and submandibular glands	2	Absence of both lacrimal glands; two upper lacrimal puncta were completely occluded	No	Kim et al. 2005 [9]
Major salivary gland agenesis	1	Lacrimal gland agenesis	Mother had less severe symptoms	Kwon et al. 2006 [10]

Table I. (Continued).

Type of salivary gland agenesis	No. of cases	Accompanying abnormality	Family history	Authors
Complete congenital agenesis of all major salivary glands	1	Lacrimal puncta were all present	No	Pham Dang et al. 2010 [1]
Absence of salivary glands	1	Absence of lacrimal glands accompanied by growth and development retardation	No	Gok et al. 2010 [11]

we reported here was an isolated form, since no family history was found. The management is mainly symptomatic. Early diagnosis and preventive management are essential.

**Declaration of interest:** The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

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