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CASE REPORT

Aplasia of submandibular salivary glands associated with ectodermal dysplasia

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We describe a 28-year-old white Caucasian man displaying many of the physical signs of ectodermal dysplasia (ED). An unusual finding was his presentation with xerostomia. Salivary gland imaging techniques revealed aplasia of both submandibular salivary glands and relatively small parotids. The case highlights that hypoplasia and aplasia of exocrine glands could be rare features of ED. In the management of ED, early detection of xerostomia is important to limit any potential damage to the already hypodontic dentition.
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The ectodermal dysplasias (ED) are heritable conditions in which there are abnormalities of two or more ectodermal structures. At least 100 different types have been identified (1) some which are mild and others that are devastating. The structures affected are commonly hair, teeth, nails and sweat glands.

The main signs and symptoms are anihydrosis or hypohydrosis, thin and sparse hair, hypodontia (2) and nail dystrophy. Other symptoms of affected individuals may include deficient tears, poorly functioning mucous membranes, frequent respiratory infections, hearing or vision deficits, cleft lip and/or palate, immune dysfunction, sensitivity to light and lack of breast development.

Congenital absence of the major salivary glands in ED is extremely rare but minor salivary glands may fail to form (3). Major salivary gland aplasia has been known to occur in association with developmental anomalies such as hemifacial microsomia and Treacher Collins syndrome but it can also occur in isolation. Only a few links to salivary gland aplasia with ED have been reported (1) but no previous mention has been made of bilateral submandibular gland aplasia in ED.

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Case report

A 28-year-old white Caucasian male with ED from Le Mans, France currently residing in the UK presented complaining of dryness of his mouth. His oral symptoms had developed progressively over the last 1-year. He did not report any skin involvement, alteration in sweating or eye problems. His medical history was unremarkable. He had, however, undergone orthognathic surgery with a bilateral mandibular osteotomy at the age of 20 years.

On examination he had thin, sparse hair, frontal bossing and a depressed nasal bridge (Fig. 1). The nails on his hands (Fig. 2) were dystrophic presenting with ridging of the nail plates. He revealed that his mother and three of his sisters also had a similar problem. Clinodactyly was noted on the little finger on both hands (Fig. 2).

Bilateral angular chelitis was present, his dorsal tongue was lobulated and fissured and there was a florid gingivitis. His oral hygiene was fair but with substantial calculus deposits present. The gingivae had considerably receded and his dentition was heavily restored. Only 16 of his natural teeth were present virtually all root filled, five of these being reduced to root stumps. His osteotomy plates were still in place but there had been significant alveolar bone resorption. Large span bridges had been constructed at a young age in all quadrants but those on the left were mobile and recently removed (Fig. 3) and he now needed complete rehabilitation of his mouth.

His unstimulated whole mouth saliva was 0.05 ml/min confirming a low flow rate (normal >0.25 ml/min). *Staphylococcus aureus* was isolated from his inflamed oral commissures along with a significant count of *Candida* in an oral rinse. Further investigations consisted of an ultrasound scan that showed normal left and right parotid glands but possibly absent submandibular salivary glands. A right parotid sialogram with 1.4 ml Omnipaque 350 revealed normal duct anatomy but a small-sized parotid gland.

A 99mTc-pertechnetate scintigram was considered the most appropriate method to confirm the absence of functioning submandibular glands (4). The scan confirmed good uptake by the parotid salivary glands but

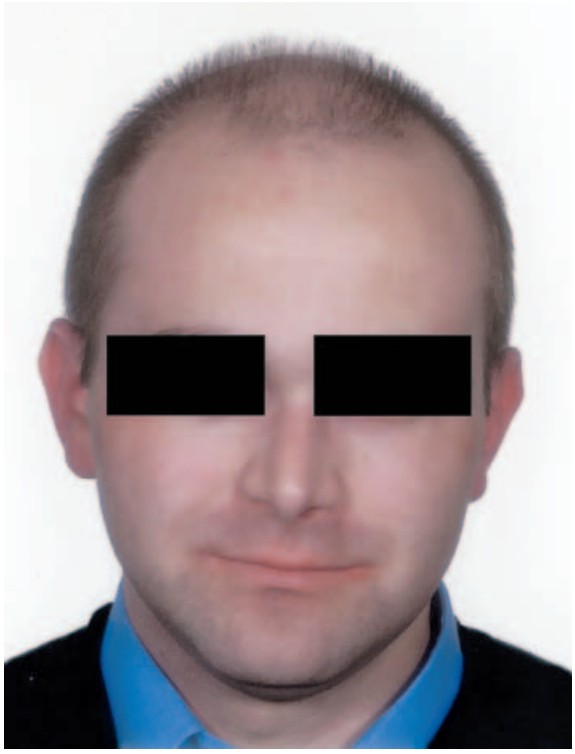

Figure 1 Sparse hair, frontal bossing and depressed nasal bridge.

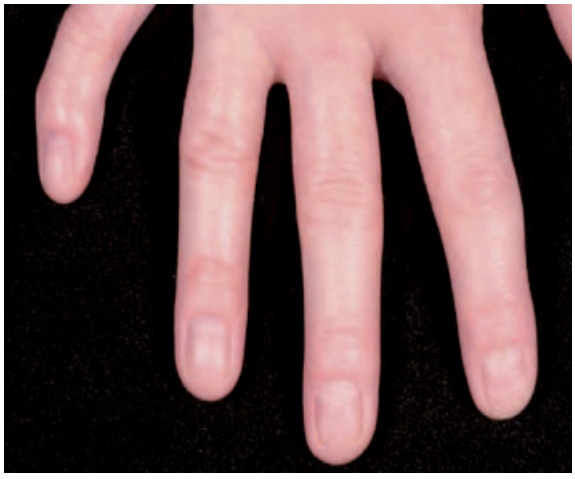

Figure 2 Vertical nail ridging and clinodactyly.

absence of functioning submandibular salivary glands was noted (Fig. 4).

Blood investigations revealed an anti-nuclear antibody titre of 1:40 and anti-smooth muscle antibody at 1:40. His extractable nuclear antigens (including anti-SSA and -SSB) were all negative and his full blood count, haematinics and liver function tests were all within the normal range. Sweat testing using iodine and starch (5) was performed on the patient's palm. This showed normal sweating when compared with a control.

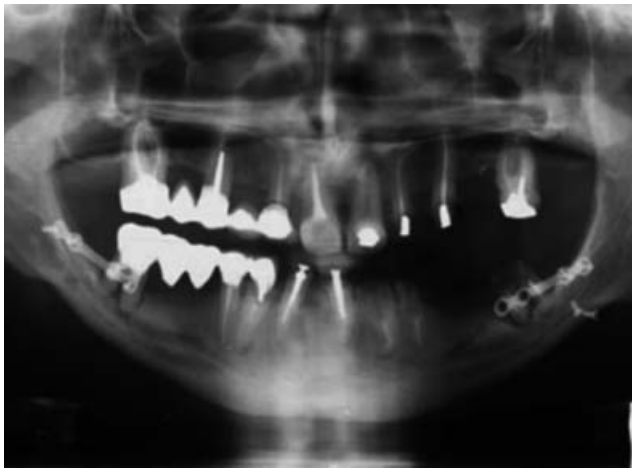

Figure 3 Panoramic image showing failing dentition and osteotomy plates.

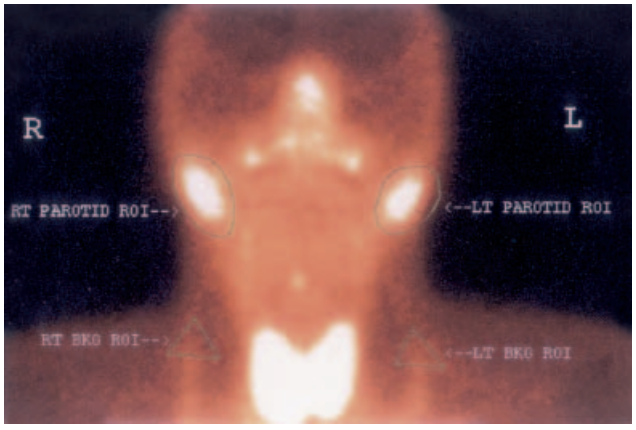

Figure 4 Salivary imaging with 99mTc-pertechnetate showing normal uptake of isotope by parotids and thyroid but no uptake in the region of the submandibular glands suggesting aplasia (ROI, region of interest).

Comments

The hair, nails and teeth are commonly affected in ED and it is clear that these structures have been compromised in this patient. The extent to which missing teeth in his dentition were due either to hypodontia or acquired tooth loss could not be ascertained as the patient could not recall which teeth never developed and which had been extracted. It was not possible to elicit if enamel or dentine had been hypoplastic since all tooth surfaces were restored.

The EDs are considered often as of x-linked inheritance, the females being the carriers and the males being affected. Female carriers may display some phenotypic characteristics of the condition, which would explain the patient's mother and three of his sisters all demonstrating vertical nail ridging. One sister had two sons but they displayed no signs of the condition. A presentation of x-linked recessive hypohidrotic ED would, however, result in abnormal sweating. The family history and normal sweating in the index case suggests a mild form

of one of many ED syndromes compatible with autosomal dominant inheritance. In mild forms of ED syndromes enamel hypoplasia is frequent which could explain in part, the severely restored dentition in this case.

Congenital absence of submandibular glands is extremely rare and it has not always been known to be symptomatic. Fracaro et al. (6) found in a healthy 13-year-old female both submandibular glands to be missing and she was unaware of any subjective xerostomia. Another report was made of a 38-year-old male with hypohidrotic ED who had hypoplastic salivary glands with absence of one submandibular gland (1). In the present case report, ultrasound scanning and a 99mTc-pertechnetate scintigram confirmed the absence of functioning submandibular glands. This case would support that submandibular glands are almost certainly of ectodermal origin.

Salivary gland aplasia can have varying clinical presentations depending on the type and number of glands absent. The symptoms may range from being negligible to persistent dry mouth causing masticatory and speech difficulty. In many adults, gland aplasia is less recognized as a cause for xerostomia. Rampant caries and severe periodontal disease may occur and reflects lack of cleansing, buffering and anti-microbial properties of saliva. The pattern of decay usually seen in patients with aplasia of the submandibular glands is caries in the mandibular incisor teeth (6) but because of the extensive restorative treatment received by the patient in this report, this could not be ascertained.

His management of dry mouth included advice to chew sugar-free gum and the use of saliva-stimulating tablets (SST), which the patient found helpful. The angular chelitis caused by *Candida* and *S. aureus* was treated with Miconazole oral gel and Neomycin sulphate ointment. Regular use of a fluoride mouth rinse was recommended as a measure of prophylaxis to reduce future dental caries.

This case report highlights that aplasia of one or more major salivary gland could be a feature of ED. As it is not always symptomatic, unless investigated by salivary gland imaging techniques their absence may pass unnoticed until adult life. An abnormally high dental caries status should alert the practitioner to investigate for developmental abnormalities of salivary glands and their function. Early diagnosis of hypoplasia or aplasia of salivary glands is essential to minimize tooth decay and gum disease with the use of meticulous oral hygiene, diet advice and the use of local fluoride delivery methods.

References

1. Nordgarden H, Johannessen S, Storhaug K, Jensen JL. Salivary gland involvement in hypohidrotic ectodermal dysplasia. *Oral Dis* 1998; **4**: 152–4.
2. Pindborg JJ. *Pathology of the dental hard tissues*. Copenhagen: Munksgaard, 1970; 21–2.
3. Scully C, Cawson RA. *Medical problems in dentistry*, 4th edn. Bristol: Wright, 1998; 245.
4. Van den Akker HP, Busemann-Sokole E. Absolute indications for salivary gland scintigraphy with 99mTc-pertechnetate. *Oral Surg Oral Med Oral Pathol* 1985; **60**: 440–7.
5. Berg D, Weingold DH, Abson KG, Olsen EA. Sweating in ectodermal dysplasia syndromes. *Arch Dermatol* 1990; **126**: 1075–9.
6. Fracaro MS, Linnett VM, Kallett KB, Savage NW. Submandibular gland aplasia and progressive dental caries: a case report. *Aust Dent J* 2002; **47**: 347–50.