Keywords: submandibular salivary glands; agenesis; Ectodermal Dysplasia

Introduction
The ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by a deficiency of ectoderm and mesoderm derived tissues, including hair, skin, teeth, and sweat glands. Inheritance patterns are variable, and the incidence of ED is estimated at 1:100,000 live births (2). The most common type is the X linked recessive Christ-Siemens-Touraine syndrome or anhidrotic/hypohidrotic ED. Patients affected with many other ED subtypes present with otolaryngologic manifestations (4, 5).

Only a few cases of ectodermal dysplasia with salivary gland aplasia are reported in the English literature (4, 6). We present our case of ectodermal dysplasia and bilateral nonfunctional submandibular glands with clinical and radiological findings.

Materials and Methods
Case report
A 22-year-old female with ED presented with complaining of dryness of her mouth, difficulty of chewing, dysphagia and recurrent upper respiratory tract infections. Her complaints had developed progressively over the last 5 years. She reported skin involvement, alteration in sweating or eye problems. Her medical history was unremarkable. On examination she had thin, sparse hair, eyebrows and eyelashes (Fig. 1). The nails on her hands were dystrophic presenting with ridging of the nail plates (Fig. 2). She revealed that her brother also had a similar problem. Bilateral angular cheilitis was present and there was a florid gingivitis. Her oral hygiene was fair. The gingivae had considerably receded and her dentition was heavily restored.

Further investigations consisted of an ultrasound scan that showed normal left and right parotid glands and small size submandibular salivary glands. Computed tomography confirmed that both submandibular glands were hypoplastic. A 99mTc-pertechnetate scintigraphy showed normal uptake in the parotid glands. However, no functioning submandibular glands were shown. Although the submandibular gland abnormalities can be rarely seen in ectodermal dysplasia, to our knowledge, this is the first report which presents nonfunctioning submandibular glands in an ectodermal dysplasia case.
submandibular salivary glands. Computed tomography was performed, revealing that both submandibular glands were hypoplastic (Fig. 3). A 99mTc-pertechnetate scintigraphy confirmed good uptake by the parotid salivary glands but absence of functioning submandibular salivary glands was noted (Fig. 4).

**Fig. 3.** Computed tomographic scan showing hypoplasia of both submandibular glands

**Fig. 4.** Salivary imaging with 99mTc-pertechnetate showing normal uptake of isotope by parotid and thyroid glands but no uptake submandibular glands

### Results and Discussion

The ectodermal dysplasias (ED) are heritable conditions in which there are abnormalities of two or more ectodermal structures. There are nearly 200 distinct disorders that comprise the EDs. The two major categories of ED, initially described by Weech (9) in 1928, are the hidrotic (Clouston syndrome) and the hypohidrotic/anhidrotic (Christ-Siemens-Touraine syndrome) forms, the latter being more common. They are differentiated by the mode of inheritance, different associated anomalies, and the presence or absence of sweat glands (1).

The most common type is the X linked recessive Christ-Siemens-Touraine syndrome or anhidrotic/hypohidrotic ED, in which affected patients have hypoplastic sweat glands with subsequent heat intolerance. They may present with deficient nasal cilia with subsequent chronic infections (rhinitis, pharyngitis, and otitis media), epistaxis, and ocular drying with corneal injury, dysphagia, hearing loss, bronchitis, dysphonia, eczematoid skin changes, unusual facies, hypodontia, and sparse hypopigmented scalp hair. Patients affected with many other ED subtypes also present with otolaryngologic manifestations (4, 5). Hair, nails and teeth are commonly affected in ED and it is clear that these structures have been compromised in our patient.

A report was made of a 28-year-old male with hypohidrotic ED who had atrophic submandibular salivary glands (6). In the present case report, ultrasound scanning and a 99mTc-pertechnetate scintigram confirmed the absence of functioning submandibular glands. This report and our case would support that submandibular glands are almost certainly of ectodermal origin.

Only a few links to salivary gland aplasia with ED have been reported (4, 6) One of them describes a man with bilateral submandibular gland aplasia as a rare feature of ED (6). The other one was characterized with unilateral submandibular gland aplasia (4). Although both submandibular glands were present, distinctly from the previous reports, they were nonfunctional in our case. To our knowledge the presence of submandibular...
glands that lack functional activity in ectodermal dysplasia makes our case unique.

Our patient was a female and had more than two complaints secondary to ectodermal tissue. She had complaints of xerostomia, difficulty in swallowing and chewing, nasal obstruction, recurrent upper respiratory tract infections. In addition, complaints not related with ear-nose-throat such as sparse hair and eyebrows, dystrophic nails, xerophthalmia, eczema of skin, tooth carries and defects of tooth were detected, too.

Clinically, patients may be asymptomatic or may present with dryness of the mouth, difficulty in chewing and swallowing, and dental carries (7). In addition, our case had burning sensation in mouth, recurrent laryngopharyngitis, generalized carries of tooth, decreased taste sensation.

Salivary gland scintigraphy is the most widely used and useful method for evaluation the functions of salivary glands (3). Salivary gland imaging techniques revealed hypoplasia of both submandibular salivary glands. Ultrasonarography is a non-invasive and rapid technique that requires minimal patient compliance and the results are easy to interpret. A 99mTc-pertechnetate scintigram was considered the most appropriate method to evaluate the function of the submandibular glands. Therefore we recommend ultrasound and salivary functional tests in patients with known or suspected EDs.

Salivary gland agenesis can cause xerostomia and there may be oral and upper respiratory tract sequel, such as dental carries, candidiasis, ascending sialadenitis, laryngitis and pharyngitis.

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

Conclusions
We present a case of ectodermal dysplasia in which both submandibular salivary glands were nonfunctional. We recommend ultrasound and salivary functional examinations in patients with known or suspected EDs. A 99mTc-pertechnetate scintigram was considered the most appropriate method to evaluate the function of the submandibular glands.

High dental caries status should alert the practitioner to investigate for developmental abnormalities of salivary glands and their function. Early diagnosis of nonfunction of salivary glands is essential to minimize infection of mouth and pharynx, tooth carries and gum disease.

REFERENCES