The Journal of Laryngology & Otology
July 2004, Vol. **118,** pp. 556–557

Anhidrotic ectodermal dysplasia presenting as atrophic rhinitis

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Abstract
Atrophic rhinitis is a chronic inflammatory disease of the nose. The aetiology of primary atrophic rhinitis is not yet known, although secondary atrophic rhinitis is known to be associated with chronic granulomatous diseases such as tuberculosis and leprosy. The authors report a case of atrophic rhinitis, which was a presenting feature of a rare genetic disorder known as Christ-Siemens-Touraine syndrome, also known as anhidrotic ectodermal dysplasia.

Key words: Atrophic Rhinitis; Anhidrotic Ectodermal Dysplasia

Introduction

Atrophic rhinitis is a common disorder affecting the nose in developing countries. It is mainly classified into primary and secondary atrophic rhinitis. The causes for secondary atrophic rhinitis are mainly post-surgery and granulomatous disorders of the nose. The authors report atrophic rhinitis as a presenting feature in an adult male with a rare genetic disorder called anhidrotic ectodermal dysplasia.

Case report

A 41-year-old male farmer presented with nasal bleeding and worms coming out of the nose of a month's duration. The bleeding was spontaneous and episodic and attendants complained of a foul smell emanating from the nose. There was no past medical history suggestive of leprosy, tuberculosis, syphilis or any other granulomatous disorder.

On examination, nasal fetor was apparent. The patient had a saddle nose deformity (Figure 1). Anterior rhinoscopy revealed the presence of live maggots and greenish crusts in both the nasal cavities. Gauze wicks impregnated with turpentine were inserted in the nasal cavities and maggots were removed. On removal wide nasal cavities and atrophic turbinates were noticed. Posterior rhinoscopy also revealed atrophic changes. Examination of the ears revealed a total absence of hair in the external auditory canals. The left tympanic membrane was dull and lustreless with impaired mobility. On examination of the oral cavity, it was seen that the patient had only two teeth in the upper jaw, which were conical and notched (Figure 1). The lower jaw had eight teeth and the posterior pharyngeal wall mucosa was dry and shiny. Fibre-optic laryngoscopy was normal. A general physical examination revealed dry, smooth skin, sparse scalp hair and total loss of eyebrows and eyelashes. Facial hair was also sparse, the patient's lips were thick and everted and fissures were noticed radiating from the angle of the mouth. Supraorbital margins were prominent. Body hair

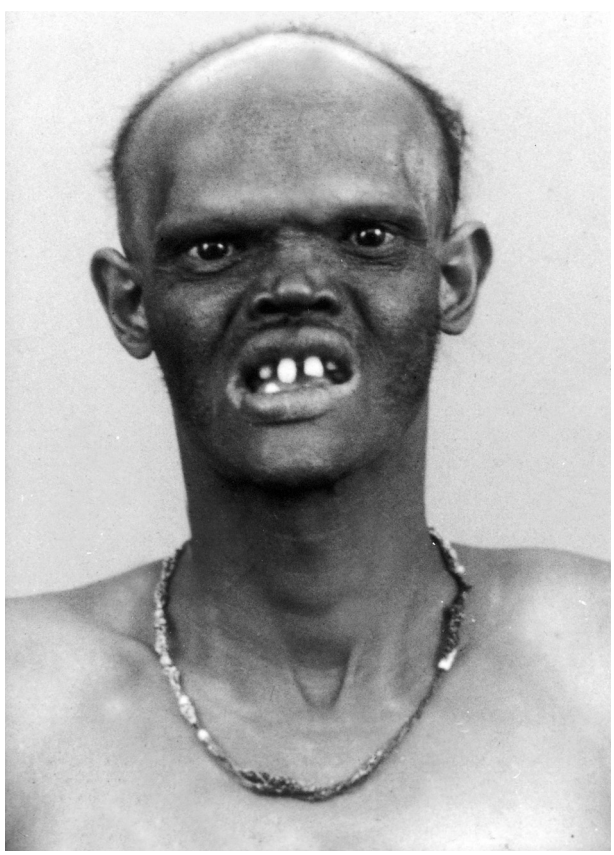

Fig. 1
Facial appearance in anhidrotic ectodermal dysplasia.

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Accepted for publication: 7 March 2004.

was completely absent except for a few hairs in the axilla and pubic region. His full blood counts, urea and electrolytes were normal. VDRL test for syphilis and acid-fast staining for leprosy were negative. X-ray of the paranasal sinuses and skull revealed wide roomy nasal cavities and hypoplastic maxillary and right frontal sinuses, prominent supraorbital ridges, mandibular hypoplasia and hypodontia. Audiology investigations revealed mixed hearing loss in the left ear and high frequency sensory hearing loss in the right ear.

Anhidrotic ectodermal dysplasia should be considered as a differential diagnosis when investigating secondary atrophic rhinitis

He was further investigated with a skin biopsy, which revealed a total absence of the eccrine glands with absent dermal appendages. This confirmed the diagnosis of anhidrotic ectodermal dysplasia, Christ-Siemens-Touraine syndrome. Further questioning revealed that the patient also had intolerance to hot weather and suffered from hyperpyrexic episodes during fever. All immediate members of the family were examined and were found to be normal. He was regularly treated with alkaline nasal douches and regular cleaning of the nasal cavity with a marked reduction in nasal symptoms.

Discussion

Wedderburn first described anhidrotic ectodermal dysplasia in literature in 1838, he noticed 10 Hindu males over a span of four generations in Sindh, who had hypodontia, dry scaly skin and sparse body hair. Darwin later published this in 1875.[1] The genetic basis and nomenclature for this disease was first proposed by Weech in 1928.[2] This disorder is inherited in an X-linked recessive manner and the syndrome is fully expressed in males. Carrier females do not manifest the full-blown syndrome, but may show slight features of the disorder like teeth abnormalities.[3] Structures derived from the ectoderm are primarily affected giving rise to skin, teeth, eccrine and mucosal gland abnormality.

Clinically, the syndrome is characterized by absence of sweating with hyperpyrexic episodes during fever and hot weather. Teeth abnormalities are present with hypodontia or anodontia, the teeth being conical and inturned. The skin is thin, soft, dry and finely wrinkled, scalp hair is sparse and fine, at times giving rise to near total alopecia. The eyebrows may be sparse or totally absent. Body hair is also scanty. [4]

The characteristic ENT features are atrophic rhinitis, which results in poor development of mucous glands in the nose and is characterized by nasal fetor and crusting. The cartilaginous and bony dorsum of the nose is sunken giving rise to a saddle nose. The nostrils are large and conspicuous.[5,6] Satyr ears may be present with either conductive or neural deafness. [4] Lack of mucous glands in the oral cavity gives rise to dry mouth and dysphasia. The patient is also susceptible to repeated attacks of upper respiratory infections[7] due to lack of mucous glands. The diagnosis can be confirmed by the sweat-pore count method as described by Crump and Danks[8] and also by a skin biopsy.

Management of this condition is difficult due to a multitude of problems. Atrophic rhinitis is managed conservatively with regular nasal douching. Cosmetic appearance can be improved by rhinoplasty and otoplasty. Prosthodontic techniques will help to improve dental abnormalities. The patient should avoid hot climates and febrile episodes should be treated immediately by cooling of the body. Genetic counselling should be offered and attention should be paid to the patient's emotional needs.

This case report is presented because of its rarity. Although there have been three case reports in ENT literature describing the association between atrophic rhinitis and anhidrotic ectodermal dysplasia in the paediatric population,[5,6,7,8] none have been described in adults. (Medline search)

Conclusion

Patients who are investigated for secondary atrophic rhinitis should be investigated for features of anhidrotic ectodermal dysplasia as a differential diagnosis and the ENT surgeon should be aware of this condition and its management.

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Mr R. Sachidananda takes responsibility for the integrity of the content of the paper.
Competing interests: None declared