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Anhidrotic ectodermal dysplasia: an ENT presentation in infancy

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Introduction

Anhidrotic ectodermal dysplasia (AED) is a rare genetic disorder characterized by absent or diminished numbers of structures derived from the ectoderm. It is recognized clinically by anhidrosis, hypotrichosis and anodontia or reduced numbers of deformed teeth (Weech, 1929; Reed *et al.*, 1970). As affected individuals cannot sweat, hyperpyrexia during infective illnesses to which they are predisposed is a potentially fatal complication. Two recent articles have stressed the nasal complications of this disorder in adults (Gil-Carcedo, 1982; Martini *et al.*, 1984). Atrophic rhinitis is common, often presenting with a chronic offensive nasal discharge. The case described here developed an offensive nasal discharge as the first manifestation of the disease at only four months of age. Early diagnosis is essential so that parents can be warned of the hazards of hyperpyrexia and offered genetic counselling. The purpose of this report is to familiarize otorhinolaryngologists with this disorder, report the very early onset of atrophic rhinitis in this case, highlight the physical signs of this disorder in infancy and outline simple investigations used to confirm the diagnosis.

Case report

The patient was the first son of unrelated English Caucasian parents. Following an uneventful pregnancy he appeared normal at birth. At the age of four months, his parents were concerned by a persistent, offensive bilateral nasal discharge associated with marked crusting. At that time, they were also worried by generalized pallor of the skin which contrasted with dark, wrinkled skin that was developing around his eyes. He contracted a chest infection at the age of eight months and required hospitalization. A diagnosis of cystic fibrosis was considered on the basis of respiratory infection, pallor and failure to thrive (both height and weight were below the third centile) but when a pilocarpine iontophoresis sweat test was performed, no sweat could be collected, and the diagnosis of anhidrotic ectodermal dysplasia was made.

The skin was pale, with a 'porcelain like' quality, and the superficial veins over the chest were easily seen. The scalp hair was sparse and fine, though not unusually so for a child of this age. The eyebrows however were sparse, being represented only by a few medial hairs, and the eyelashes were similarly diminished in number. The periocular skin was finely wrinkled and hyperpigmented (Fig. 1). Anterior rhinoscopy revealed marked crusting of the nasal mucosa with an associated bilateral purulent, foul-smelling nasal discharge. There was no family history of hair, dental or sweating abnormalities, or atopic diseases. Both parents' skin, hair and teeth were normal.

On direct questioning his parents had not considered that his hair was abnormal and they were unaware that he could not sweat.

The diagnosis of AED was confirmed by hand sweat pore studies using crystal violet in cellulose and acetone solution, as described by Crump and Danks (1971). The patient's impressions showed flattened fingerprint ridges and absent sweat pores (Fig. 2); his mother's impressions were normal. A radiograph of the skull at eight months showed unerupted conical upper incisors (Fig. 3); at the age of 13 months, they had erupted, and atopic eczema had developed in the antecubital and popliteal fossae.

Discussion

As its title suggests, the ectoderm expresses the main abnormal features in AED. Nasal symptoms are common in AED, and they usually consist of a fetid, purulent nasal discharge associated with often tightly adherent nasal crusts, both of which are features of atrophic rhinitis. Ozaena was a presenting complaint in two adults with previously undiagnosed AED reported by Martini *et al.* (1984). More detailed studies of nasal pathology in this condition are few. Gil-Carcedo (1982) described greatly expanded nasal cavities with striking atrophy of the turbinates in an affected boy, and nasal mucosal biopsy demonstrated squamous metaplasia with almost complete loss of ciliated cells. Recurrent nasal infections have been reported to progress to nasal septal destruction.

Structures of non-ectodermal derivation may also be involved, and the respiratory tract is one of the most important. Post-mortem studies of a case described by Reed *et al.* (1970)



Fig. 1

Fine wrinkling and pigmentation of the lower lid and sparse eyebrow and lash hairs evident at eight months of age.

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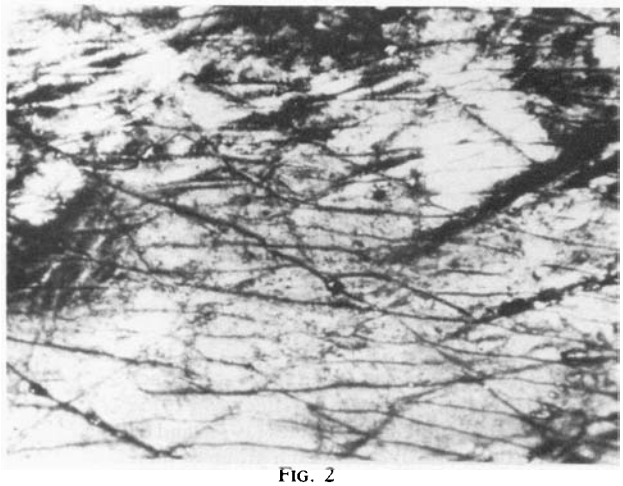


FIG. 2

Finger impressions showing flattened ridges and absent sweat pores in the propositus. (Crystal violet in cellulose acetate in acetone preparation × 10.)



Radiograph of the skull showing unerupted conical upper incisors.

showed absence of mucus-secreting glands from the tracheal mucosa, and similar absence of glands was noted in the oesophageal and gastric mucosae. The absence of mucus-secreting glands is felt to predispose to the increased incidence of infections of the respiratory tract; pharyngiti,s bronchitis and pneumonia being the main problems. The absence of sweat glands prevents normal thermostasis during these infective episodes, and hyperpyrexia may be alarming, with temperatures as high as 108 degrees Fahrenheit being recorded (Drago and Erenreich, 1961); this is obviously a potential cause of death and may be responsible for permanent brain damage resulting in the mental retardation occasionally complicating this condition (Capitanio *et al.*, 1968). One important aspect of this condition is the neccessity of alerting parents to this potential complication, so that frequent temperature recordings can be taken at times of ill health; should hyperpyrexia develop, then sponging with tepid water may help maintain a safe temperature.

The disorder is usually inherited in a sex-linked recessive fashion, manifesting fully in affected males; partial expression in carrier females is seen in some pedigrees (Kerr *et al.*, 1966). Autosomal dominant (Lipton and Roberts, 1950) and recessive (McDonald, 1949) inheritance patterns are reported, when full expression may be seen in females as well as males.

The scalp hair in affected children is usually fine, sparse in density, fair in colour and slow-growing, and seldom grows to a great length. The skin is pale, shiny and wrinkled, and atopic eczema has recently been noticed as a frequent association. Sweat glands are almost completely absent, and the absence of sweat ducts allows easy recognition of affected individuals by non-invasive tests. The nails are usually normal. The teeth, if present, are of reduced number, there seldom being more than four, they are severely dystrophic and usually represented by conical or peg-shaped incisors. Complete anodontia is not uncommon. In addition a number of other features are felt to be characteristic; the face shows frontal bossing and the lips are protruberant and everted, the ears are pointed and the nasal bridge saddle-shaped due to its depression and breadth (Gorlin and Pindborg, 1964). Amastia, sensorineural deafness and gonadal and skeletal abnormalities have all been associated with AED.

The characteristic features of the disease may not be apparent in the first year of life, as sparse hair is so common, anhidrosis seldom noted by parents and the abnormal teeth are yet to erupt. The fine wrinkling and pigmentation seen in the case reported here are some of the earliest signs of the disease. The eyebrows and lashes in the first year of life in normal children are usually well formed in contrast to their sparsity in AED, and both of these features should be sought.

The diagnosis may be easily confirmed by making finger print impressions, as described by Crump and Danks (1971). The procedure is painless and rapid, and relies on the finding of absent finger sweat pores in affected patients. It is much preferable to performing skin biopsies. Skull radiography will demonstrate unerupted dystrophic teeth or absent dentition and is a reliable indicator of the disease even at birth.

This case is unusual in that the first symptom noted by the parents at only four months of age was nasal discharge. It is therefore important for ENT specialists to be familiar not only with the typical findings of AED in older children, but with those seen in infants too.

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