The Journal of Laryngology and Otology
April 1996, Vol. **110**, pp. 379–382

Persistent nasal crusting due to hypohidrotic ectodermal dysplasia

A. H. AL-JASSIM, A. C. SWIFT, CH.M., F.R.C.S.

Abstract
Hypohidrotic ectodermal dysplasia is an hereditary condition of the ectodermal tissues which may escape recognition because of lack of clinical awareness due to its rarity. Otorhinolaryngological features of this syndrome include chronic respiratory tract infections, persistent foul-smelling nasal discharge and crust formation, and hearing problems.

The condition is usually an X-linked recessive disease affecting mainly ectodermal tissue, although non-ectodermal tissue may also be affected. The notation 'ectodermal' is used because ectodermal tissues are always involved. The syndrome is characterized by complete or partial absence of sweat glands, sparse hair growth, absent or deformed peg teeth, sparse sebaceous glands, occasional absence of salivary and lacrimal glands, scanty mucous glands and deficient cilia.

We describe two children with this disorder with specific reference to the effect on the upper respiratory tract.

Key words: Ectodermal dysplasia

Introduction

Anhidria and anogontia was described in two Jewish boys by Danz in 1792 (Freire-Maia and Pinheiro, 1984) but the first known case of ectodermal dysplasia was reported in 1848 (Thurnam, 1848).

Darwin (1890) discussed an association between hair and teeth. He describes instances of deficient teeth in both hairless dogs and men with inherited baldness. Darwin specifically mentions 10 men in four generations of a Hindu family who had very little body hair and 'who suffer much during hot weather from excessive dryness of the skin'.

The term of 'hereditary ectodermal dysplasia' was suggested in 1929 (Weech, 1929) but the differentiation between the hypohidrotic and anhidrotic types was not made until 1939 (Clouston, 1939). The hypohidrotic form is five times more common in males (Upshaw and Montgomery, 1949).

Patients

Case 1

A five-year-old boy was referred to the ENT clinic with a three-year history of chronic nasal discharge and a foul odour from his nose (Table I). Hypohidrotic ectodermal dysplasia had been recognized when he was admitted at the age of three weeks with a history of poor feeding. Pregnancy and delivery had both been normal.

During his first three years he was frequently admitted to the children's ward with recurrent diarrhoea and vomiting, respiratory tract infections and asthma. He received glycerine and bicarbonate nasal drops for his foul-smelling crusts and Hypermellose drops for dry eyes. His teeth failed to develop and he was subsequently fitted with dentures (Figure 1). He also has aplastic lacrimal punctae affecting both eyes, intermittent aphonia and he has had one episode of parotitis and pyrexia.

Hypohidrotic ectodermal dysplasia was confirmed by genetic DNA testing. His mother is a carrier and has hypodontia and pointed teeth. Features of this syndrome were also present in both the maternal grandmother and great-grandmother.

He has undergone clearance of the nasal crusts on several occasions under general anaesthesia. *Haemophilus influenzae* and a heavy growth of *Staphylococcus aureus* have been cultured from the crusts on each occasion.

He has recently used a Grossan nasal irrigator to clear his nose and has been treated with Naseptin and long term erythromycin for three months, but the latter was unfortunately ineffective.

Case 2

A three and a half-year-old girl was referred with a

TABLE I
FEATURES OF HYPOHIDROTIC ECTODERMAL DYSPLASIA IN TWO CHILDREN

	Case 1	*Case 2*
Sex	Boy	Girl
Age	5 years	3.5 years
Family history	Positive	Negative
Nose	Foul-smelling odour	Foul-smelling odour
Teeth	Anodontia	Peg teeth
Hair	Thin and sparse	Thin
Eyebrows	Sparse	Very thin
Eyelashes	Absent	Very thin
Face	Mandibular hypoplasia	Maxillary hypoplasia
Ears	Normal	Normal
Mental status	Hyperactive	Normal

From the Department of Otorhinolaryngology, Aintree Hospitals (Walton), Rice Lane, Liverpool, UK.
Accepted for publication: 31 December 1995.

https://doi.org/10.1017/S0022215100133687 Published online by Cambridge University Press



FIG. 1
A five-year-old boy with hypohidrotic ectodermal dysplasia. He is wearing a full set of dentures.

history of chronic infection and crusting in both nasal cavities. She was also intolerant to heat and had poor hair and nail growth and peg-like teeth (Figure 2). The nasal crusts had a foul odour and although cleared under a general anaesthetic on three occasions, they reformed again very quickly. Simultaneous antral washouts were always clear. Cultures of the crusts grew lactose non-fermenting coliforms and β-haemolytic *Streptococcus* Lancefield Group A but fungal cultures were negative.

During infancy she had been extremely ill due to a combination of cerebral and retropharyngeal abscesses, but she has since made a remarkable recovery. A sweat test was negative on three occasions. Immunoglobulins showed a raised IgA and low IgG, but IgG sub-classes were normal. Nasal brushings from the inferior and middle turbinates were reported as showing reduced numbers of cilia but the beat pattern was normal and the overall mean beat frequency was 13.4/second. Histology of the inferior turbinate showed a non-specific inflammatory change. A skin prick test to common allergens was positive to house-dust mite, *Cladosporium*, *Aspergillus*, grass pollen, cats and dogs. Anti-staphylolysin titre was: <2 units/ml (0–2). Cystic fibrosis gene mutations were investigated but were negative.

She has been treated with erythromycin and Naseptin for several months and this has been effective at reducing the foul odour and nasal discharge.

Discussion

The two children described clearly demonstrate the main clinical features of hypohidrotic ectodermal dysplasia and although the first child (*Case 1*) was diagnosed in infancy the condition was not recognized in the second child (*Case 2*) until she was three-years-old, in spite of her previous medical history. This delay arose purely from a lack of awareness of the condition due to its rarity.

One hundred and eight clinical variations of ectodermal dysplasia have so far been described (Freire-Maia and Pinheiro, 1984). The best known of these is Christ-Siemens-Touraine (CST) syndrome or X-linked hypohydrotic ectodermal dysplasia. The sex-linked nature of the inheritance in the hypohidrotic groups has been disputed (Weech, 1929) but two groups of patients with hypohidrosis have subsequently been described: one due to an X-linked gene, which may have mild to severe expression in heterozygous females (the CST syndrome), and the other due to a homozygous autosomal recessive gene (autosomal recessive hypohidrotic ectodermal dysplasia, Freire-Maia and Pinheiro, 1984). The two patients described in our report both have the CST syndrome or hypohidrotic X-linked ectodermal dysplasia which has the following features:

(1) Anhidrosis/hypohidrosis;
(2) Episodic hyperthermia;
(3) Apocrine gland agenesis;
(4) Thin, sparse hair;
(5) Nasal crusting;
(6) Deficient/anomalous dentition;
(7) Distinctive facial features;
(8) External ear anomalies;
(9) Upper respiratory tract infections.

Absent sweat glands may affect the whole body (Sunderman, 1941; Lipton and Roberts, 1950; Metson and Williams, 1952) but sometimes sweat glands may be present over certain regions of the body such as the chest and axilla (Felsher, 1944) or the forehead (Everett *et al.*, 1952).

Hair is often sparse and thin, eyebrows are usually absent and the eye lashes are fine or may be absent.

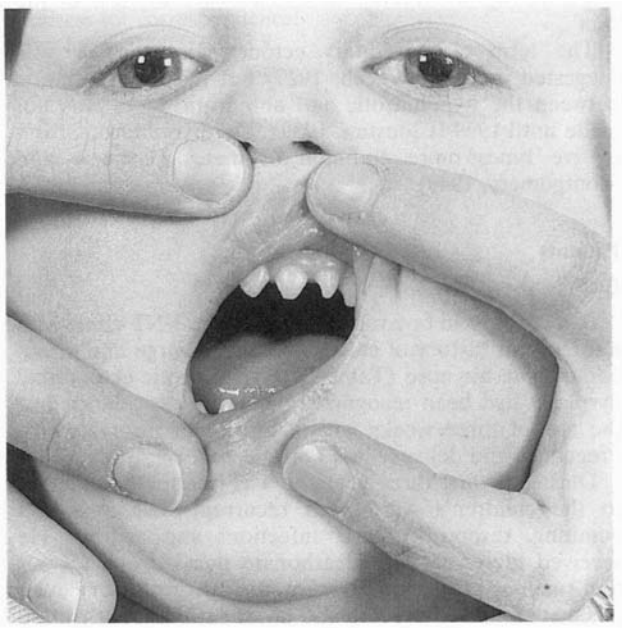


FIG. 2
Typically deformed peg teeth in a 3½ year-old girl with hypohidrotic ectodermal dysplasia.

https://doi.org/10.1017/S0022215100133687 Published online by Cambridge University Press

However, moustaches and beards are usually normal. The periorbital region is often more darkly pigmented than the rest of the body. The skin around the eyes is thin and wrinkled and photophobia may be present. The lacrimal glands may be hypoplastic or aplastic and dry eyes may lead to conjunctivitis (Reed *et al.*, 1970).

Atrophic rhinitis, crusting, discharge with ozaena, and a decreased sense of smell have also been reported (Weech, 1929). Histology of tissue from the muco-cutaneous junction of the nasal septum showed no gland formation (Metson and Williams, 1952). Dental anomalies may include anodontia, severe hypodontia or deformed peg-like teeth. Brodie and Sarnat (1942) reported a case of hypohidrotic ectodermal dysplasia in which both deciduous and permanent tooth buds were missing. Shaw (1990) reported a similar case with complete anodontia. Exfoliative cytology studies in patients with hypohidrotic ectodermal dysplasia show a reduction in the amount of keratinization in the buccal mucosa and this is thought to be due to anodontia (Bennett, 1963).

Severely affected patients have distinctive faces with prominent thick lips, a saddle nose, prominent frontal bossing, supra orbital ridges and hypoplasia of the maxilla. Oral dryness, hoarseness, intermittent aphonia, dysphagia, tonsillitis, pharyngitis and a pharyngeal abscess may also occur.

The pinna may show various anomalies and is often displaced anteriorly (Hartwell *et al.*, 1965). A conductive hearing loss may result from either blockage of the external canal by dry wax (Reed *et al.*, 1970) or Eustachian tube dysfunction (Lowry *et al.*, 1966; Clarke *et al.*, 1987). Late sensorineural deafness in adult patients with hypohidrotic ectodermal dysplasia has also been reported (Helweg-Larsen and Ludvigsen, 1946).

Beahrs *et al.* (1971) noted an association between chronic infection of the upper respiratory tract and hypohidrotic ectodermal dysplasia which they attribute to atrophic mucous glands and absent cilia. The incidence of atopy and asthma is high and is thought to be due to either a direct effect of the syndrome or increased permeability of the respiratory mucosa to inhaled extrinsic allergens (Reed *et al.*, 1970; Beahrs *et al.*, 1971; Clarke *et al.*, 1987). However, there is no evidence of an immunological deficiency (Clarke *et al.*, 1987).

In a study of 56 families with hypohidrotic ectodermal dysplasia, hyperthermic convulsions were much more common in children compared to adults (Clarke *et al.*, 1987). They suggested that bottle feeds should not be heated and older children should take cool drinks and wear T-shirts to overcome their heat intolerance.

Clarke *et al.* (1987) also suggested that making the diagnosis early in the child's life improves the outcome of the disease. The diagnosis is confirmed by the history, clinical features, a skin biopsy to confirm partial or total absence of sweat glands and genetic studies. Foetoscopy and foetal skin biopsy have also enabled the diagnosis to be made antenatally (Angelina Mariano and Blanchet Bardon, 1990). Women who carry the condition can be recognized by dental anomalies, an abnormal pattern of sweating or sparse scalp hair. Mental handicap is not considered to be a feature (Clarke *et al.*, 1987), although mental retardation has been reported (Upshaw and Montgomery, 1949).

The management of hypohidrotic ectodermal dysplasia should be multi-disciplinary and may require the expertise of paediatricians, orthodontists, speech therapists, plastic surgeons, geneticists, and counsellors as well as ENT surgeons. Children may need dentures to aid mastication and speech development. Nasal crusting can be improved by nasal douching or irrigation with a saline/bicarbonate solution but this is impractical in very young children. Similarly, topical corticosteroid nasal sprays may be helpful but they are not suitable until the child is older. Mitral and aortic valve replacement surgery in a patient with hypohidrotic ectodermal dysplasia has been recently described (Rozycka-Chrzanowska *et al.*, 1994).

In conclusion, otorhinolaryngologists should be aware of this rare, genetic condition which is likely to present with persistent infection and crusting within the nasal cavities. Regular nasal toilet and treatment with long-term antibiotics may help control the foul smell and nasal crusts, but these will be a long-term problem which will become easier to control with growth and maturity.

Acknowledgements

We are very grateful to Professor Valerie Lund for doing the studies on nasal ciliary beat, to Mr R. Hancock for the photography and Mrs Joy Olsen for typing the manuscript.

References

Angelina Mariano, M., Blanchet Bardon, C. L. (1990) Diagnostico antenatal E.M. dermatologia: contribuicao do estudo ultraestrutural. *Acta Medica Portuguesa* **2:** 101–107.

Beahrs, J. O., Lillington, G. A., Rosan, R. C., Russin, L., Lindgren, J. A., Rowley, P. T. (1971) Anhidrotic ectodermal dysplasia: Predisposition to bronchial disease. *Annals of Internal Medicine* **74:** 92–96.

Bennett, C. G. (1963) Study of exfoliative cytology of oral mucosa of children exhibiting clinical evidence of ectodermal dysplasia. *Journal of Dental Research* **4:** 943–949.

Brodie, A. G., Sarnat, B. G. (1942) Ectodermal dysplasia (anhidrotic type) with complete anodontia. A serial roentgenographic cephalometric appraisal. *American Journal of Diseases of Children* **64:** 1046–1054.

Clarke, A., Philips, D. I. M., Brown, R., Harper, P. S. (1987) Clinical aspects of X-linked hypohidrotic ectodermal dysplasia. *Archives of Disease in Childhood* **62:** 989–996.

Clouston, H. R. (1939) The major forms of hereditary ectodermal dysplasia. *Journal of the Canadian Medical Association* **40:** 1.

Darwin, C. (1890) *The Variations of Plants and Animals Under Domestications.* vol 2, D. Appleton and Company Inc., New York, p 319.

Everett, F. G., Jump, E. B., Sutherland, W. F., Savara, B. S., Suher, T. (1952) Anhidrotic ectodermal dysplasia with anodontia. A study of two families. *Journal of the American Dental Association* **44:** 173–186.

Felsher, Z. (1944) Hereditary ectodermal dysplasia. Report of a case with experimental study. *Archives of Dermatology and Syphilology* **60:** 410–414.

Freire-Maia, N., Pinheiro, M. (1984) *Ectodermal Dysplasia. A Taxonomic Approach in Ectodermal Dysplasias: A Clinical and Genetic Study.* 1st Edition, Liss, A. R., Inc., New York, p 22–27.

Hartwell, S. W. Jr., Pickrell, K., Quinn, G. (1965) Congenital anhidrotic ectodermal dysplasia. A report of two cases. *Clinical Pediatrics* **4:** 383–386.

Helweg-Larsen, H. J., Ludvigsen, K. (1946) Congenital familial anhidrosis and neurolabyrinthitis. *Acta Dermato-Venereologica (Stockholm)* **26:** 489–505.

Lipton, I., Roberts, M. H. (1950) Hereditary ectodermal dysplasia of the anhidrotic type. *American Journal of Diseases in Children* **79:** 504–509.

Lowry, B. B., Robinson, G. G., Miller, J. R. (1966) Hereditary ectodermal dysplasia symptoms, inheritance patterns, differential diagnosis, management. *Clinical Pediatrics* **5:** 395–402.

Metson, B. F., Williams, B. K. (1952) Hereditary ectodermal dysplasia of the anhidrotic type. Report of a case in a negro. *Journal of Pediatrics* **40:** 303–309.

Reed, W. B., Lopez, D. A., Landing, B. (1970) Clinical spectrum of anhidrotic ectodermal dysplasia. *Archives of Dermatology* **102:** 134–143.

https://doi.org/10.1017/S0022215100133687 Published online by Cambridge University Press

Rozycka-Chrzanowska, B., Hryniewiecki, T., Solik-Tomassi, A., Rawczynska-Englert, I., Sliwinski, M., Pajewska-Klimczak, M. (1994) Mitral and aortic valve replacement in a patient with ectodermal anhydrotic dysplasia. A case report. *Journal of Heart Valve Disease* **3:** 224–225.

Shaw, R. M. (1990) Prosthetic management of hypohydrotic ectodermal dysplasia with anodontia. Case report. *Australian Dental Journal* **35:** 113–116.

Sunderman, F. W. (1941) Persons lacking sweat glands. A hereditary ectodermal dysplasia of the anhidrotic type. *Archives of Internal Medicine* **67:** 846–854.

Thurnam, J. (1848) Two cases in which the skin, hair and teeth were very imperfectly developed. *Medicochirurgical Transactions* **31:** 71–82.

Upshaw, B. Y., Montgomery, H. (1949) Hereditary anhidrotic ectodermal dysplasia. A clinical and pathological study. *Archives of Dermatology and Syphilology* **60:** 1170–1183.

Weech, A. A. (1929) Hereditary ectodermal dysplasia. (Congenital ectodermal defect). A report of two cases. *American Journal of Diseases in Children* **37:** 766–789.

Address for correspondence:
A. C. Swift, Ch.M., F.R.C.S., F.R.C.S.Ed.,
Consultant Otorhinolaryngologist,
Department of ENT Surgery,
Aintree Hospitals (Walton),
Rice Lane,
Liverpool L9 1AE.

Fax: 0151 529 4782

https://doi.org/10.1017/S0022215100133687 Published online by Cambridge University Press