

Case Report

A 4-year-old male child with hypohidrotic ectodermal dysplasia

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Abstract A 4-year-old male child presented with history of dry skin, scanty scalp hair, and decreased sweating since birth. On examination scalp hair were scanty, thin, dry and brownish in color. Facial features were frontal bossing, saddle shaped nose, thick everted lips and low set large ears. There was partial adontia with only one conical incisor tooth. There was generalized dryness of the skin. Mucosae and nails were normal. Histopathology of the skin showed rudimentary and decreased number of eccrine sweat glands, which confirmed the diagnosis of hypohidrotic ectodermal dysplasia.

Key words

Hypohidrotic ectodermal dysplasia.

Case report

A 4-year-old male child presented with the history of thin and scanty scalp hair, dry skin, decreased sweating and intolerance to heat since birth. There was also history of erythematous scaly rash on face arms and legs off and on. He did not have any other medical problem and the growth was normal. His parents had consanguineous marriage, but there was no family history of similar disorder or atopy.

On examination scalp hair were thin, sparse, dry and brownish in color. Facial features were frontal bossing, saddle shaped nose, thick everted lips and low set large ears (**Figure 1**) Oral cavity showed partial adontia, with only one incisor tooth present in the upper jaw (**Figure 2**). There was also loss of eyebrows and eyelashes. Generalized xerosis was also present.

Mucosae and nails were normal. Microscopic examination of scalp hair revealed no hair shaft abnormalities. Histopathology of the skin showed thin and flattened epidermis. Eccrine sweat glands were few in number and rudimentary. On the basis of abovementioned findings diagnosis of hypohidrotic ectodermal dysplasia was made.

Discussion

Ectodermal dysplasia (ED) is an inherited disorder in which two or more ectodermally derived structures fail to develop, or are abnormal in development.¹ To date, more than 170 distinct disorders have been described.² The more common are hypohidrotic ED (Christ-Siemens-Touraine syndrome) and hidrotic ED (Clouston syndrome).³

Hypohidrotic variety of ED is more common. It is inherited as X-linked recessive trait, therefore, more common in males.⁴ Its incidence is 1/10,000 to 1/100,000 births. Each person with ED has different combination of ectodermal

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Figure 1 Child showing thin and scanty scalp hair, frontal bossing, saddle shaped nose, thick everted lips and low set large ears.



Figure 2 Oral cavity showing conical incisor teeth in the upper jaw.

defects. The tissues primarily involved are skin, hair, eccrine sweat glands, teeth and nails. Pure EDs are manifested by defects in ectodermal structures alone, while ED syndromes are defined by the combination of ectodermal defects in association with other anomalies.

Our patient had involvement of skin, hair and teeth with features of atopic eczema. No other associated anomalies like cleft lip and/or cleft palate were found.⁵ He had no nail involvement, which is the main feature of hidrotic ED having normal facial features. Management of such cases is not difficult; life expectancy in such patients, by observing certain precautions, is as good as in normal children.⁶ It includes use of

emollients and mild topical steroids for erythematous scaly rash, education of the parents about the disease and advice to avoid excessive heat exposure and vigorous physical activity, frequent consumption of cool liquids to maintain hydration and thermoregulation. For dental defects referral is made to dental surgeon for evaluation and early intervention, in order to restore their appearance and develop positive self-image.⁷ Maintenance of oral hygiene and use of fluoride treatment is also essential. Growth abnormalities may be present in children with ED like weight and height deficit.⁸ Therefore, careful evaluation and monitoring of growth is necessary.

References

1. Sarvan I, Naidoo S, Norval EJ. Hypohidrotic ectodermal dysplasia: an unusual presentation and management in an 11-year-old Xhosa boy. *SAJD* 2000; **55**: 34-7.
2. Priolo M, Silengo M, Lerone M, Ravazzolo R. Ectodermal dysplasias: not only skin deep. *Clinic Genet* 2000; **58**: 415-30.
3. Solomon LM, Keuer EJ. The ectodermal dysplasias. Problems of classification and some newer syndromes. *Arch Dermatol* 1980; **116**: 1259-9.
4. Kere J, Srivastava AK, Montonon O. X-linked anhidrotic (hypohidrotic) ectodermal dysplasia is caused by mutation in a novel transmembrane protein. *Nat Genet* 1996; **13**: 409-16.
5. Fosko SW, Stenn KS, Bolognia JL. Ectodermal dysplasias associated with clefting: significance of scalp dermatitis. *J Am Acad Dermatol* 1992; **27**: 249-56.
6. Bari AU, Rahman SB. Hypohidrotic ectodermal dysplasia: a case report and literature review. *J Pak Ass Dermatol* 2007; **17**: 52-5.
7. Dhanrajani PJ, Jiffry AD. Management of ectodermal dysplasia: a literature review. *Dent Update* 1998; **25**: 73-5.
8. Motil KJ, Fete TJ, Fraley JK *et al.* Growth characteristics of children with ectodermal dysplasia syndromes. *Pediatrics* 2005; **116**: 229-34.