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Case Report

Hypohidrotic ectodermal dysplasia: a case report and literature review

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Abstract Ectodermal dysplasia is a rare hereditary disorder involving two or more of the ectodermal structures, which include the skin, hair, nails, teeth, mucus and sweat glands. Different combination of defects may give rise to variable phenotypes of this syndromic disorder. We present a nine years old boy having dental, skin and sweat glands defect. A brief review of literature is also presented.

Key words
Ectodermal dysplasias, hypodontia, anodontia, hypohidrosis, anhidrosis

Introduction

Ectodermal dysplasia (ED) syndrome is a rare heterogeneous group of inherited disorders that share primary defects in the development of two or more tissues derived from ectoderm. These tissues primarily are the skin, hair, nails, eccrine glands, and teeth. Defects in tissues derived from other embryologic layers are not uncommon. The disorders are congenital, diffuse, and nonprogressive. To date, more than 150 distinctive syndromes have been described with all possible modes of inheritance. The most common syndromes within this group are hypohidrotic (anhidrotic) ED and hidrotic ED.[1,2] Hypohidrotic ED (also known as Christ-Siemens-Touraine syndrome) is the more common phenotype and is usually inherited as an X-linked recessive trait. It is characterized by several defects (e.g. hypohidrosis, anomalous dentition, onychodysplasia, hypotrichosis). Typical facies are characterized by frontal bossing; sunken cheeks; saddle nose; thick, everted lips; wrinkled, hyperpigmented skin around the eyes; and large, low-set ears. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth. Eccrine sweat glands may be absent or sparse and rudimentary, particularly in those with hypohidrotic EDS. In some cases, mucous glands are absent in the upper respiratory tract and in the bronchi, esophagus, and duodenum.[1-6] Scalp hair may be sparse, short, fine, dry or there may be complete absence of hair. Structural hair-shaft abnormalities like longitudinal grooving, hair-shaft torsion, and cuticle ruffling can also be seen.[7] The prevalence of atopic eczema is high. Other common signs are short stature, eye abnormalities, decreased tearing, and photophobia. Intelligence is normal. Nails are often brittle

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and thin or show abnormal ridging, but they may be grossly deformed especially in hidrotic type. The presence or absence of these abnormalities defines the different syndromes. Rare associated findings may be seen as breast hypoplasia, missing fingers or toes, deafness, cleft lip and/or palate, impaired immune system and photosensitivity.[2,3,7,8] ED results from dyshistogenesis of ectodermal derivatives in early embryonic life and gene responsible for X-linked hypohidrotic ED has been mapped in the proximal area of the long arm of band Xq-12-q13.1 and decreased expression of the epidermal growth factor receptor has been thought to play a causal role in phenotype of this condition.[9] Morbidity and mortality is related to the absence or presence of eccrine and mucous glands. In majority of cases, life expectancy is normal. Children with decreased sweating may have a mortality rate of up to 30% in infancy or early childhood because of intermittent hyperpyrexia.[6] Recurrent high fever may lead to seizures and neurological sequelae. In affected patients with severe dental abnormalities, feeding difficulties may result in malnutrition and failure to thrive. Abnormal eccrine and mucous glands may result in secondary infections, especially in the upper respiratory tract. X-linked hypohidrotic ED has full expression only in males and female carriers show little or no signs of the condition. Jaw radiographs are indicated for infants with fever of unknown origin and possible hypohidrotic ED; tooth buds may be absent. X-ray of hands, feet, or both may demonstrate specific deformities. Sweating response can be seen by applying yellow starch-iodine powder on palmar or dorsal skin in a sweating room (sauna) for 10 minutes. Skin biopsy is the most reliable test to demonstrate absence or hypoplasia of sweat glands.[2,3,10] Fetal skin biopsy may help identify decreased eccrine sweat glands for prenatal diagnosis of hypohidrotic ED. No definite pharmacological treatment is available and the management of affected patients depends on which structures are involved. Patients with hypohidrotic ED are advised light clothing, a cool-water spray bottle, air conditioning for environment, use of artificial tears and application of petrolatum for nasal mucosa protection. Overexposure to warm temperatures and vigorous physical activities are to be avoided.[10] Moisturizers are prescribed to prevent xerosis or eczema as necessary for dry skin. Patients with severe alopecia may wear wigs to improve their appearance.[3,10] Orthodontic treatment may be indicated for cosmetic reasons and to ensure adequate nutritional intake.[10-12] Cleft lip or palate and other midfacial defects may need corrections to improve function and reduce physical disfigurement.[8,10]

Case history

A 9 year-old boy, reported with dry skin, itchy scaly lesions over his face, dental abnormalities and history of repeated febrile illnesses. His symptoms became noticeable in 2nd year of life when his parents started noticing dryness of his skin, sensitivity to sun, high grade fever off and on. Hyperpyrexia was more common in hot humid season. His teeth, especially central ones, gradually became malformed and loose. The child felt more comfortable in winter months. His intelligence was normal and there was no family history of atopy or any similar findings in any of his other

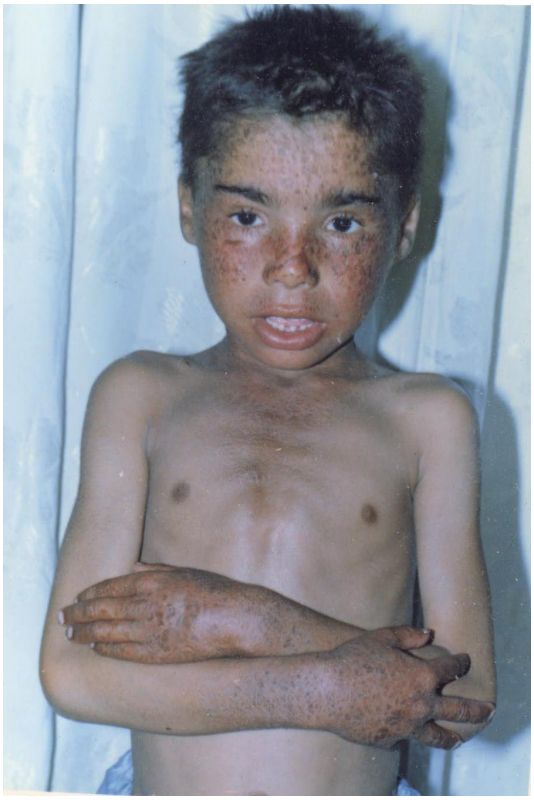

Figure1 Child showing atopic rash, xerosis more marked in photoexposed areas in addition to abnormal teeth.

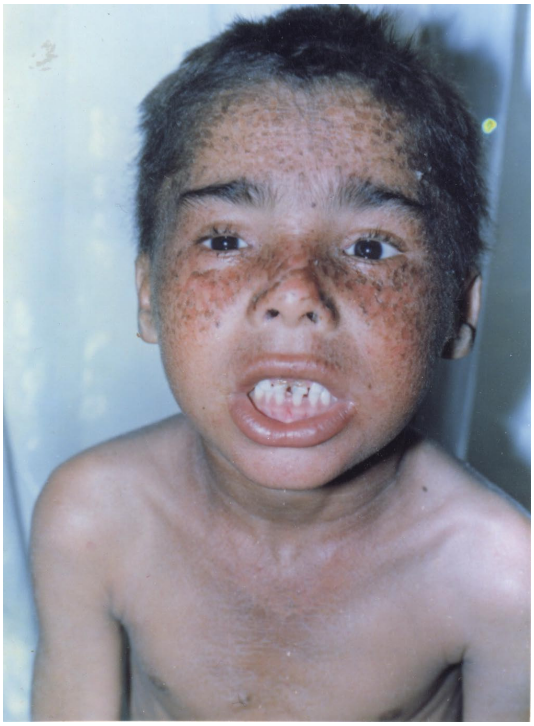

Figure 2 Central teeth are malformed (peg shaped).

family members. On examination, dry eczematous (atopic) rash was seen on his face and upper limbs and it was more marked in periorbital regions (**Figure 1**). Central incisors and canine teeth were malformed and widely spaced (**Figure 2**). Ears were somewhat low set and there was also an element of photosensitivity. Hair was dry and difficult to manage but they were not sparse or thinned out. Facial bony features wer e otherwise normal with no frontal bossing or depressed nasal bridge. His hair under light microscope showed longitudinal ridging but no established hair shaft disorder. Diagnosis was confirmed by skin biopsy, which revealed thin and flattened epidermis, reduction in the number and presence of rudimentary eccrine sweat glands. He was given emollient with topical steroid for his eczematous rash and sunscreen for use over sun exposed area before going under sun. Parents were educated about the disease and were advised to observe precautions regarding vigorous physical activities and exposure of the child to hot environment. He was also advised to keep good oral hygiene, to avoid sun exposure and to consult orthodontic specialist for expert dental management.

Discussion

Most common mode of inheritance of ED is X-linked. However, both recessive and dominant forms also exist. Autosomal recessive disorders are phenotypically indistinguishable from the X-linked forms. Some ED types are mild, while others are devastating. Each person with ectodermal dysplasia may have a different combination of defects. One may have hair and nails affected, while another may show

involvement of sweat glands and teeth. Each combination is considered a distinct type of ectodermal dysplasia. Obvious manifestations of the disorders are not clinically apparent in newborns and are normally evident during infancy or early childhood.[3,5] The patient reported here had involvement of sweat glands and teeth. Other ectodermal structures were largely unaffected. In addition, he had atopic eczema, xerosis, low set ears and photosensitivity. These clinical features were supportive in diagnosing ED, as these are described in association with this disorder.[3,5,10] This was not a very difficult to manage case and needed reassurance, education, and consultation for orthodontic treatment. Life expectancy in such cases, by observing necessary precautions, is almost as good as in other non affected children.

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