Ectodermal Dysplasia: Report of Four Cases and Review of Literature

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Abstract

We report four cases of ectodermal dysplasia. Three of them are hypohidrotic ectodermal dysplasia (HED) and other one is cleft lip/ palate syndrome. Each patient presented with hypohidrosis, sparse hair, oligodontia. Ectodermal dysplasias are genetic disorders that result in defective structure or function of two or more major derivatives of the ectoderm, which include the sweat glands, hair, teeth, and nails. Hypohidrotic ectodermal dysplasia represents a group of ectodermal dysplasias that are characterized by sparse or absent eccrine glands as well as by hypotrichosis and oligodontia with peg-shaped teeth. HED is caused by defects in the ectodysplasin signal transduction pathway. Cleft lip/ palate syndrome; the inheritance of this syndrome is probably determined by an autosomal recessive gene. The scalp hair is often fine, dry, sparse and light in colour; the nails are dystrophic and teeth are few and small. Other features are cleft lip and palate, hypohidrosis and defects of the external genitalia.

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Introduction

The ectodermal dysplasias (EDs) are a group of inherited disorders that share in common developmental defects involving at least two of the major structures classically held to derive from the embryonic ectoderm-hair, teeth, nails, sweat glands[1].

Cases

Case 1: A 18 year old girl, presented to our clinic for evaluation of hypotrichosis, hypohidrosis and teeth abnormalities. The patient had history of parchment like skin sheet at birth. Clinical examination of the patient revealed a fine, sparse scalp hair, eyebrows, and eyelashes. Her skin was smooth and dry, and periorbital, perioral hyperpigmentation and wrinkling were evident.

Sebaceous hyperplasia was apparent in various degrees on the medial aspects of the nose (Fig 1).

The teeth were reduced in number and conical in shape (Fig 2), the alveolar ridges were underdeveloped, and the lips were prominent.

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Additional facial features included frontal bossing, prominent supraorbital ridges, and midfacial hypoplasia with a depressed nasal root and bridge. She had no nail dystrophy. She had a history of recurrent high fevers during infancy, and her mother reported that she does not sweat. She had family history of hypohidrosis, hypotrichosis and dental abnormalities; her brother is affected too.

Case 2: A 7 year old boy, presented to our clinic for evaluation of hypotrichosis, hypohidrosis and teeth abnormalities. The patient had history of parchment like skin sheet at birth. Clinical examination of the patient revealed a fine, sparse scalp hair, eyebrows, and eyelashes (Fig 3).

His skin was smooth and dry, and periorbital, perioral hyperpigmentation and wrinkling were evident. Sebaceous hyperplasia was apparent in various degrees on the medial aspects of the nose. The teeth were reduced in number and conical in shape and the lips were prominent. Additional facial features included frontal bossing, prominent supraorbital ridges, and midfacial hypoplasia with a depressed nasal root and bridge. He had palmoplantar hyperkeratosis. He had no nail dystrophy. His medical history includes oligodontia, chronic nasal congestion and mild conductive hearing loss.

He had a history of recurrent high fevers during infancy, and his mother reported that he does not sweat. He had family history of hypohidrosis,

hypotrichosis and dental abnormalities; his sister is affected too.

Case 3: A 19 year old young female, presented to our clinic for evaluation of teeth abnormalities.

She had a history of recurrent high fevers during infancy, and the patient's mother reported that her daughter is not able to sweat she had to apply some precautions to protect her from overheating during warm weather and physical exertion. The patient's skin was smooth and dry. Mild periorbital and perioral hyperpigmentation were evident. She had oligodontia. The teeth were reduced in number and conical in shape.

She had no nail dystrophy. Other family members (including first degree cousins) were affected by hypohidrosis and dental abnormalities.

Her mother's medical history included four death births. Her family pedigree is represented at Figure 4.

Our diagnosis was hypohidrotic ED for these three patients (case1-2-3). The diagnosis had been made by clinical findings and positive family history.

Case 4: A 5 year old girl, was referred to our clinic for nail dystrophy, hypotrichosis and teeth abnormalities. Clinical examination of the patient revealed a fine, sparse scalp hair, eyebrows, and eyelashes. Scalp hair was light in color. The nails were dystrophic and teeth were few and small. She had an operation scar because of cleft lip and palate and defects of the external genitalia (absence of labia minor).

Her medical history includes lack of lacrimal duct and mild conductive hearing loss. All these clinical findings were significant for cleft lip/ palate syndrome that had been diagnosed in early childhood. The diagnosis had been made by clinical aspects.

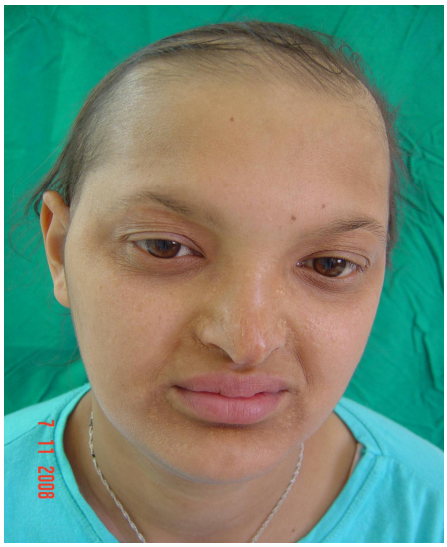

Fig.1 Periorbital, perioral hyperpigmentation and wrinkling were evident. Sebaceous hyperplasia was apparent on the medial aspects of the nose.

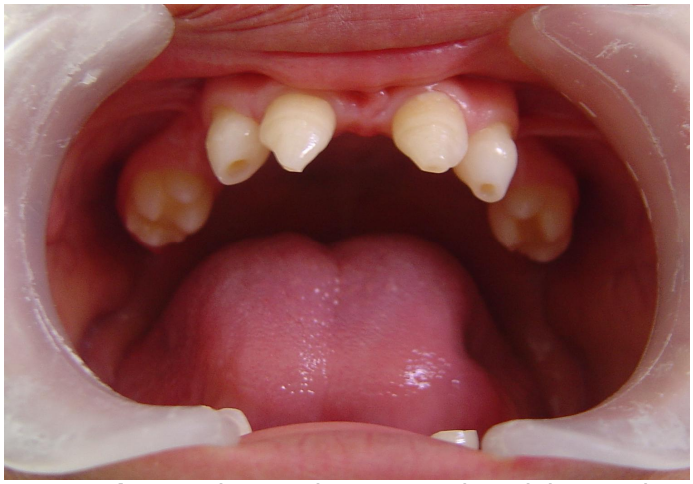

Fig. 2 The teeth were reduced in number and conical in shape.

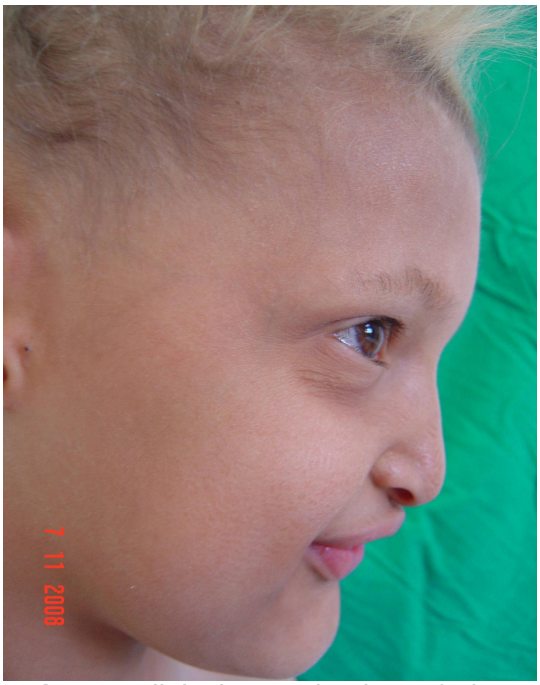

Fig. 3 Clinical examination of the patient revealed a fine, sparse scalp hair, eyebrows, and eyelashes.

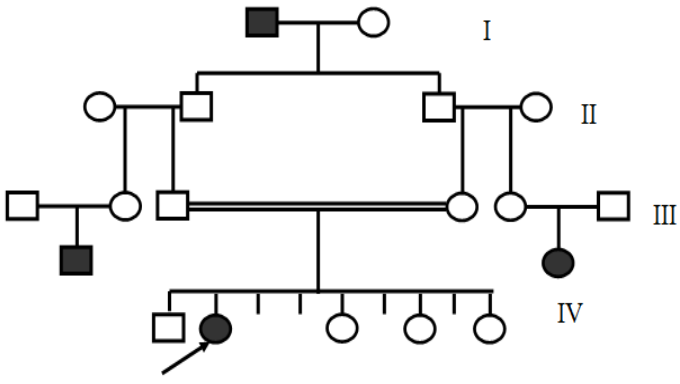

Fig. 4 Pedigree of case 3.

Discussion

EDs represent a large group of hereditary conditions characterized by congenital defects of one or more ectodermal structures including skin appendages. The original constructional theme encoded in ectoderm diverges into epidermis, hair, sweat and milk glands, and the mineralized crystalline anvils of teeth, under the direction of local signals emanating from the underlying mesoderm.

The intimate origins of these diverse ectodermal structures account for the wide spectrum of dysplasias. Clinically the hair (hypotrichosis, partial or total alopecia), nails (dystrophic, hypertrophic, or abnormally keratinized), teeth (enamel defects or absence), and sweat glands (hypoplastic or aplastic) are usually affected[2].

An estimated incidence of ED is about 7 in 10,000 births and all mendelian modes of inheritance have been reported[3]. Of more than 190 EDs described, the molecular basis has been elucidated for more than 30 of them[4].

Dental defects represent a core clinical feature of many EDs: anodontia, polydontia, dysplastic teeth, retained primary teeth, deficient enamel development (amelogenesis imperfecta), dentine deficiency (dentinogenesis imperfecta), and underdevelopment of the alveolar ridge[5]. In some EDs, the number of erupted teeth is reduced, the spacing of the teeth disrupted, and the peridontium affected. Disturbance of the enamel matrix may occur, making the teeth more susceptible to caries, and altering the shape of the teeth, leading to a pegged appearance and additional accessory cusps[3].

X-linked hypohidrotic ED (Christ-Siemens-Touraine syndrome) is the most frequent form was first described in 1848 by Thurnam and later in the 19 th century by Darwin. Both autosomal dominant and autosomal recessive inheritance of clinically similar conditions have now been demonstrated and the molecular defects defined[6].

The HEDs are caused by genetic defects in ectodysplasin signal transduction pathways. Epithelial cells in developing tooth, hair follicle an eccrine sweat gland utilize this pathway during morphogenesis, and genetic defects in the pathway results in aplasia, hypoplasia or dysplasia of these structures. The pathway is activated at a critical time during development in a specific group of epithelial cells. With activation, a transcription factor, NF-κB, is translocated into the nucleus of these cells and alters the expression of an unknown number of target genes. The change in gene expression likely has an affect on both cellular proliferation and survival[1,6].

The diagnosis is usually made with the identification of hypotrichosis, characteristic facial features, hypohidrosis (and more rarely anhidrosis), and teeth abnormalities.

The nails are usually normal. Abnormalities in the development of tooth buds result in hypodontia and peg-shaped or pointed teeth[7]. The hypodontia varies in each case, but usually only 5 to permanent teeth are present, the teeth are smaller than average, and the eruption of teeth is often delayed[8].

The extent of hypodontia may be useful in assessing the severity of the disease, and is best done with dental radiographs[3]. It is extremely important for children with ED to be seen by a dentist early. Radiographs after 2 years reveal the state of the permanent teeth and allow children to be fitted with a plate early; this is helpful for both aesthetic reasons, and to maintain the alveolar ridge, allowing for later tooth implantation[3].

Cleft lip/ palate syndrome; the inheritance of this syndrome is probably determined by an autosomal recessive gene. Hypohidrosis accompanies slight frontal bossing and some depression of the nasal bridge. The scalp hair is often fine, dry, sparse and light in colour; the nails are dystrophic and teeth are few and small. Other features are cleft lip and palate, syndactyly and defects of the external genitalia. Lacrimal gland abnormalities are frequently[1].

The disorder was assigned to chromosome 11q23 by linkage mapping[9].

Tooth and nail syndrome (Witkop syndrome) is another type of autosomal-dominant ED associated with specific dental findings. The primary dentition is usually unaffected, although the teeth may be small or peg-shaped, whereas the secondary dentition is often partially or completely absent[10].

Oculodentodigital dysplasia is a rare autosomal dominant subtype of ED, characterized by bilateral microphthalmos, nose malformations, hypotrichosis, syndactyly, and dental abnormalities particularly enamel hypoplasia[3].

Dental treatment is often necessary in patients with some forms of ED and some children may need dentures as early as 2 years of age[11]. It is important to seek dental advice early as maintenance of the alveolar ridge is important for later dental intervention[3,13,15].

Prosthetic teeth are implanted in adults for mastication and speech. Importantly, aesthetic dental interventions in patients with ED and malformed teeth and malocclusion helps with the development of a positive self-image and overall oral health[12-16].

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