Hypohidrotic Ectodermal Dysplasia: A Case Report & Literature Review

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ABSTRACT: Ectodermal dysplasia is a syndrome of 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 (HED) is caused by defects in the ectodysplasin signal transduction pathway. HED represents a group of ectodermal dysplasia that are characterized by sparse or absent eccrine glands as well as by hypotrichosis and oligodontia with peg-shaped teeth. Different combination of defects may give rise to variable phenotypes of this syndromic disorder. We present a case of twenty one year old male patient having dental, skin and sweat glands defect. A brief review of literature is also presented.

KEYWORDS: Ectodermal dysplasia, Hypohidrosis, Hypodontia, Sparse hair.

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.¹² Ectodermal dysplasia might be inherited in any form of several genetic patterns including autosomal-dominant, autosomal-recessive, and X-linked modes. It is characterized by the triad of signs which comprises of sparse hair (atrlichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The etiology of ectodermal dysplasia appears to be genetic in nature.⁴

Ectodermal dysplasia as first described by Thurnam in 1848, and later in the 19th century by Darwin, but the term 'ectodermal dysplasia' was coined by Weech in 1929. Genetic studies regarding the etiology of ED reveal that mutations in the ectodysplasin-A and ectodysplasin-A receptor genes are responsible for X-linked and autosomal hypohidrotic ectodermal dysplasia.⁸ Although now more than 170 different subtypes of ectodermal dysplasia have been identified, still these disorders are considered to be relatively rare with an estimated incidence of ED is about 1 in 100,000 births and all mendelian modes of inheritance have been reported.⁵⁸

According to the state of sweat glands involvement, two major groups are distinguished: (1) X-linked anhidrotic or hypohidrotic, where sweat glands are either absent or significantly reduced in number (Christ-Siemens-Touraine syndrome), and (2) hidrotic, where sweat glands are normal and the condition is inherited as autosomal dominant (Clouston's syndrome).⁵⁶⁷ The dentition and hair are affected similarly in both types, but the hereditary patterns, nail and sweat gland manifestations tend to differ.⁸

Hypohidrotic ED is the more common phenotype and is usually inherited as an X-linked recessive trait with the gene mapping to Xq12-q13; and decreased expression of the epidermal growth factor receptor has been thought to play a causal role in phenotype of this condition,” 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, protruberant lips, wrinkled, hyperpigmented skin around the eyes, large, low set ears, fine and sparse hair on the body and scalp. Fingernails and toenails may also show faulty development. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth.¹⁰

Affected individuals typically display heat intolerance because of reduced number of eccrine sweat glands. Fever with unknown origin may lead to early diagnosis during infancy. The prevalence of atopic eczema is high. Other common signs are short stature, eye abnormalities, decreased tearing, and photophobia. Intelligence is generally normal.¹²

CASE HISTORY

A 21 year old male patient reported to our institution with the complaint of multiple missing teeth since childhood. The patient also gave a history of delay in the eruption of deciduous and permanent teeth, intolerance to heat and reportedly less sweat production. There was no
the patient has a short stature.

A history of consanguineous marriage between the parents. Physically the patient has a short stature.

On extra oral examination, the patient had dry skin with periocular area being hyperpigmented and wrinkled with sparse hair on the body and scalp. [Fig 1] Hairs present were fine in texture & lighter in color. Both upper and lower eyelids showed sparse eyelashes. Prominent supraorbital ridges, large and outwardly placed low set ears, flattened nasal bridge and protuberant lips were also present. [Fig 2]

Intra oral examination revealed multiple missing teeth and the teeth present were conical in shape in the maxillary arch and an edentulous mandibular arch was seen. [Fig 3] The similar findings were present in the other family member (including first degree cousins), as revealed by the patient.

An orthopantomograph was made which revealed multiple missing teeth in maxillary arch and an edentulous mandibular arch, thin alveolar ridges and reduced vertical bone height of maxillary and mandibular jaws. [Fig 4]

Based on clinical presentation and positive family history the patient was diagnosed as a case of hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome).

DISCUSSION

The Ectodermal dysplasia are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryonic ectoderm, the intimate origins of these diverse ectodermal structures account for the wide spectrum of dysplasia. 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. 13

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 altering the expression of an unknown number of target genes. The change in gene expression likely has an effect on both cellular proliferation and survival. 13,14

The major concern seen in these patients is the lack of teeth and the special appearance, as seen in our case. The most characteristic findings are the reduced number and abnormal shape of teeth. The delay in eruption of teeth is often the first step in the diagnosis. The men have an easily recognizable facies, also referred to as an old man facies, short stature and low set ears. 14 Some infants have a premature look with scaling of the skin. This can also form a clue to the diagnosis. 17 The extraoral features seen in this disorder are frontal bossing with the forehead appearing square in shape, prominent supra orbital ridge, with periocular area being hyperpigmented and wrinkled, depressed nasal bridge (saddle nose) as seen in our case. The other features include midface hypoplasia, pointed chin and protuberent and everted lips, as seen in this case. 18 Abnormalities of skin, teeth and thin and sparse hair are noted, but nail dystrophy is not seen in our case.

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. 18 In some EDs, the number of erupted teeth is reduced, the spacing of the teeth disrupted, and the periodontium 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. 18

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. The hypodontia varies in each case, but usually only 5 to 7 permanent teeth are present, the teeth are smaller than average, and the eruption of teeth is often delayed.

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. It is important to seek dental advice early, as maintenance of the alveolar ridge is important for later dental intervention.

CONCLUSION

Ectodermal dysplasia is a rare genetic disorder with involvement of various tissues in the body. A careful and thorough examination will lead to an accurate diagnosis. Restoration of normal function should be the main concern in these patients. Since oligodontia or complete anodontia leads to atrophy of the alveolar bone, prosthetic treatment is of great value to these patients from functional, psychological, and psychosocial stand points.
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REFERENCES

LIST OF PHOTOGRAPHS

Fig 1: Facial view showing sparse hair on face and scalp

Fig 2: Profile view showing large ear and protuberant lips

Fig 3: Intraoral photograph showing multiple missing teeth

Fig 4: Orthopantomograph showing multiple missing teeth