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Photo Vignette

Hypohidrotic and hidrotic ectodermal dysplasia: a report of two cases

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ABSTRACT

Ectodermal dysplasias are a large group of syndromes characterized by anomalies in the structures of ectodermal origin. There are 2 major types of this disorder, based on clinical findings: hypohidrotic ectodermal dysplasia and hidrotic ectodermal dysplasia. This clinical classification is very important because clinical professionals involved with this disease need first a clear and practical method of diagnosis. The main oral manifestation of ectodermal dysplasia may be expressed as hypodontia. Thus, dental professionals may be the first to diagnose ectodermal dysplasia. The present article reports one case of each of the main types (hypohidrotic and hidrotic) of ectodermal dysplasia and the authors review the literature regarding the pathogenesis, clinical features, and therapeutic management of this condition.

INTRODUCTION

Ectodermal dysplasias (EDs) represent a large and complex nosological group of congenital diseases, which were first described by Thurnam in 1948 and later in the 19th century by Darwin [1]. It is a hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo [2]. There are 2 major types of this disorder: hypohidrotic ectodermal dysplasia (HED), in which sweat glands are either absent or significantly reduced in number, and hidrotic ectodermal dysplasia (HidED), in which sweat glands are normal. The condition is inherited in an autosomal dominant manner. The dentition and hair are affected similarly in both types, but the hereditary patterns and nail and sweat gland manifestations tend to differ [3,4]. The present article reports one case of each of the main types (hypohidrotic and hidrotic) of ectodermal dysplasia. In addition, the authors review the literature regarding the pathogenesis, clinical features, and therapeutic management of this condition.

CASE PRESENTATIONS

Case I

A 29-year-old man with missing teeth, heat intolerance, and absence of sweating since childhood came to the Center of Dermatological Studies of Recife (CEDER), Brazil. The physical extraoral examination showed low implantation of the ear, frontal bossing, saddle nose, hyperpigmentation in the periorbital region, sparse hair, thin and dry skin, and palmoplantar hyperkeratosis. In addition, the vertical dimension of his lower face was reduced and his lips were protuberant, leading to the characteristic appearance of old age (Figure 1). Intraoral examination revealed the absence of many permanent teeth (Figure 2). The hypodontia was also confirmed by panoramic radiography, as well as by the underdevelopment of alveolar ridges

(Figure 3). In addition, other members of the family shared similar alterations. Because hypohidrosis, hypotricosis, and hypodontia were very evident, the patient was diagnosed with HED. The patient was referred to a dermatologist and to a specialized dental rehabilitation center for orthodontic treatment and prosthetic rehabilitation.



Figure 1. The physical extraoral examination showing low implantation of the ear, frontal bossing, saddle nose, pigmentation in the periorbital region, sparse hair, the vertical dimension of lower face reduced and lips protuberant and keratosis palmoplantar.



Figure 2. Intraoral examination showing hypodontia.



Figure 3. Panoramic radiography showing hypodontia and underdevelopment of alveolar ridges

Case II

A 14-year-old boy with missing teeth came to the Center of Dermatological Studies of Recife (CEDER), Brazil. His mother reported the medical history of his lack of teeth since childhood. There was no family history of missing teeth. The physical extraoral examination showed low implantation of the ear, scanty eyebrows, and nail dystrophy (Figure 4). Intraoral examination revealed the absence of many permanent teeth (Figure 5), which was confirmed by panoramic radiography (Figure 6). Based on these clinical findings, he was diagnosed with HidED. The patient was referred to a dermatologist and to a specialized dental rehabilitation center for orthodontic treatment and prosthetic rehabilitation.



Figure 4. The physical extraoral examination showing low implantation of the ear, scanty eyebrows.



Figure 5. Intraoral examination showing hypodontia.



Figure 6. Panoramic radiography showing hypodontia.

DISCUSSION

EDs are a large group of syndromes and nearly 200 different conditions have been described under this term [2,5,6]. EDs are heterogenous concerning clinical and genetic aspects and are characterized by anomalies in the structures of ectodermal origin. They can be manifested in problems relating to hair, nails, teeth, sweat glands, and sebaceous glands, which may or may not be associated with alterations in other ectodermal appendages [2,5,6,7]. The incidence of EDs is estimated to be 7 in 10,000 live births [2,5].

Currently, the causative gene has been identified in about 30 different EDs [6,8,9,10]. With advances in the understanding of the genetic basis of the disease, newer classifications, which use molecular knowledge as the starting point and are based on the defects in cell-cell communication and signalling, adhesion, transcription regulation, or development are proposed, such as the Priolo and Lagana classification in 2001 [8] and the Lamartine classification in 2003 [8,9,10]. The classification based on molecular aspects is very important. However, these classification systems are not of much relevance from a clinical point of

view, particularly with respect to diagnosis and management because this classification is not yet available for all EDs. Furthermore, clinical professionals involved with this disease (pediatricians, dermatologists, ophthalmologists, dentists, physiotherapists, and other professionals) need first a clear and practical method of diagnosis before one can clarify information concerned with cause [4,7].

Classification of EDs based on clinical findings is more relevant and may be divided into two broad categories [3,4]:

1. Hypohidrotic ectodermal dysplasia (X-linked hypohidrotic ectodermal dysplasia or Christ-Siemens-Touraine syndrome).
2. Hidrotic ectodermal dysplasia (Clouston's syndrome)

The most frequent of these two is HED, caused by mutations in the Ectodysplasin gene (EDA), located at Xq12–q13.1, which is more observed in males than females [2,11]. The EDA gene encodes the transmembrane type II protein ectodysplasin, which belongs to the tumor necrosis factor superfamily. Normally EDA is expressed in ectodermal tissues; it is implicated in epithelial-mesenchymal interactions during ectodermal morphogenesis and odontogenesis [2,11].

HED is characterized by the classical triad of hypotrichosis, hypohidrosis, and hypodontia. Scalp hair, eyebrows, and eyelashes may be sparse or absent (hypotrichosis). Hypoplasia of the sweat and submucous glands is cause of the hypohidrosis; sweating, although present, is greatly deficient leading to episodes of heat intolerance and hyperthermia [2,11,12]. Undiagnosed infant boys with HED syndrome can die of hyperthermia during minor febrile infections owing to absent or severely decreased sweating. Glandular hypoplasia also affects the respiratory tract and the middle ear where it may lead to recurrent infections [12].

Oral traits of ED may be expressed as dry mucosa and mainly hypodontia. Sometimes, diagnosis is delayed until the teeth fail to erupt at the expected age. As a result of the hypodontia and lack of alveolar ridge, the vertical dimension of the lower face is reduced and the lips become protuberant. The face of an affected child usually has the appearance of old age [3,13]. Not only number but also shape anomalies are often conspicuous both in the primary and permanent dentition, such as root and crown dysmorphies, mainly conical shaped incisors, and slightly abnormal molar crown shape [11,13].

Other signs of classic HED include periorbital hyperpigmentation, periorbital wrinkle, frontal bossing, prominent supraorbital, depressed nasal bridge (saddle nose deformity), a prominent chin, and hyperkeratosis of the palms and soles. Additional anomalies, such as chronic rhinitis, pharyngitis, laryngitis, and disorders of mental development may accompany the above-mentioned symptoms in some cases [13,14].

The classical triad of hypotrichosis, hypohidrosis, hypodontia, and almost all other features were found in case 1. Patient 1 reported that other male members of his family (uncles and cousins) showed the same characteristics, which is suggestive of an X-linked recessive inheritance.

HidED has an autosomal-dominant inheritance with complete penetrance and variable expressivity [2,7,15]. Although HidED has been described in families of various ethnic origins, it is particularly common in persons of French-Canadian ancestry, suggesting a founder effect for the HidED mutation in this population [4,16]. It's caused by mutations in GJB6, located at 13q12, which encodes connexin 30, a component of intercellular gap junctions [2,7,15].

The main features of HidED are nail deformities frequently associated with paronychia infections, hair defects that range from brittleness and slow growth rate to partial or total alopecia and hypodontia that occurs in varying degrees of severity [3,15,16]. Most of the clinical features are similar to that seen in the HED, e.g. the dentition and hair are affected similarly in both types. However, nail and sweat gland manifestations tend to differ. In HidED the sweat glands are normal [3,4,16].

Patient 2 showed hypodontia, sparse eyebrows, and nail dystrophy. His mother reported that the patient had never had episodes of heat intolerance or hyperthermia. In addition the family stated that there was no such disease in other members. Thus, we concluded that the patient is a carrier of HidED.

The clinical manifestations of ED cause considerable social problems in individuals affected by the condition. Medical management is symptomatic, mainly for patients with hypohidrosis (carried with HED). Patients are required to stay in a cool environment (air conditioning), to wear clothes that help keep them cool, and to drink cool liquids often. Emollients for dry skin, artificial tears for patients with decreased lacrimation, saline sprays for nasal mucosa, and antibiotics if any infection occurs may be required [4].

Dental professionals may be the first to diagnose ED- mainly because of hypodontia- in a patient. Thus, dentists have a responsibility in the detection of carrier status since hypodontia in either of the dentitions may be the only sign of what can be a potentially life-threatening condition [13,16].

Early orthodontic treatment and prosthetic rehabilitation are important for children and young patients with hypodontia because they may suffer from social isolation [17]. Follow-up by a multidisciplinary team involving pediatric dentistry, orthodontics, prosthodontics, and oral-maxillofacial surgery specialists is advocated to be the most appropriate approach in such cases [3]. Dental management is necessary not only to protect alveolar bone continuity and obtain regular phonation but also to solve esthetic and psychological problems [4,13]. In the present cases, the patients were referred to a specialized dental rehabilitation center.

HED and HidED are anomalies of great interest to dental clinicians because of the patients' absence of teeth. In this case report, 2 patients diagnosed with HED and HidED associated with hypodontia are described. The early detection of such disorders is of great value to these patients from the functional, psychological, and psychosocial standpoints.

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