ORIGINAL RESEARCH PAPER

Oral Pathology

KEY WORDS: Hereditary Ectodermal dysplasia; ectoderm; genetic.

"Hereditary ectodermal dysplasia- Case report with Review of literature".

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ABSTRACT

Hereditary Ectodermal dysplasia (ED) comprises of a group of inherited conditions in which two or more ectodermally derived anatomic structures fail to develop. There are numerous subtypes of ED inherited in several genetic patterns. Depending upon the subtype different types of tissues are affected. As teeth are invariably affected in ectodermal dysplasia its knowledge is necessary for dental professionals. We present here a case of Hereditary Ectodermal dysplasia with various clinical and radiological manifestations. Also a brief account of ectodermal dysplasia and its current management techniques are discussed below.

INTRODUCTION:

Ectodermal dysplasia (ED) syndrome comprises of diverse group of inherited conditions in which development of two or more ectodermally derived tissues like skin, hair, nails, eccrine glands, and teeth are affected and also these disorders are congenital, diffuse, and non-progressive. Tissues derived from other embryologic layers are also affected. [1]

The first case of ectodermal dysplasia was published by Thurnam in 1848 and the term was coined by Weech in 1929.[2] The earliest documented cases of ED were in 1792, subsequently more than 200 different pathologic clinical conditions have been recognized and defined as ED showing incidence of about 1 in 10,000 to 100,000 births.[3]

ED causes significant problems in mastication, speech and psychological development so dental treatment will help to maintain self-esteem and social well-being of the affected individuals, [3] hence considerable awareness among health professionals is required. Two types of ED are seen, hypohidrotic and hidrotic. Hypohidrotic being more common. In this article we present a case of Hypohidrotic ED with brief note on its review and management.

CASE REPORT.

An 18 yrs. old male patient came to our OPD with a chief complaint of multiple missing teeth and wanted replacement of the same. The patient had no relevant family and past medical history. Patient had his upper anterior deciduous teeth exfoliated in past. Decreased sweating and heat intolerance were also experienced by the patient. On extra oral examination, the patient had prominent supraorbital ridges, periorbital pigmentation, mild mandibular hypoplasia, scarce hair on face and generalized body. Patient had dry scaly skin with dystrophic hand nails (Fig 1 and 2). Intra-oral examination revealed multiple missing permanent teeth and over-retained deciduous teeth in upper as well as lower jaw. Crowns of lower anterior teeth were conical in shape. Existing teeth were hypoplastic with spacing between them (Fig 3). Orthopantomogram (OPG) showed over-retained deciduous teeth and absence of numerous permanent teeth. Taurodontism was noted in deciduous as well as permanent molars. Roots of many teeth showed external resorption (Fig 4).

DISCUSSION:

The most common syndromes within ED group are hypohidrotic (anhidrotic) ED and hidrotic ED. Hypohidrotic ED also known as Christ-Siemens-Touraine syndrome shows classical triad of hypodontia, hypotrichosis, and anhidrosis along with other symptoms [4] and is usually inherited as an X-linked recessive trait which shows complete expression only in males and female carriers show little or no signs of the condition.[1] with carriers-incidence of 17.3 in 100,000 women. However autosomal recessive and autosomal dominant modes of transmission are infrequently seen. [5]

Hidrotic ED (Clouston syndrome) an autosomal dominant condition in which patients show normal facies, normal sweating and no specific dental defect.6 However some reported cases suggest involvement of teeth as anodontia or hypodontia [7,8]

Witkop's syndrome, or tooth and nail syndrome is an autosomal dominant condition with hypodontia, conical teeth and nail dysplasia. Nail disorders are present from birth and improves with age. Also sweating is normal and the hair, even though fine, is normal in distribution.[9]

Classification

According to Freire-Maia's classification, EDs are divided into two categories: Group A which comprises all the entities with disturbances in two or more of the classical structures, and Group B which includes those with alterations in only one of these structures plus another ectodermal defect. The ectodermal structures are represented as: 1- hair, 2- teeth, 3- nails, 4- sweat glands.

The A group is subdivided into 11 subgroups, according to the involved structures: 1-2-3-4; 1-2-3; 1-2-4; 1-3-4; 2-3-4; 1-2; 1-3; 1-4; 2-3; 2-4; 3-4. Similarly, entities belonging to Group B are classified with the same criteria into four subgroups with number 5 added at the end, indicating that another ectodermal defect is present: 1-5, 2-5, 3-5, and 4-5.10

Etiology

The responsible genetic defects for X-linked ED occur on the ectodysplasin (EDA1) gene, which is located at chromosome Xq12-q13.1 that encodes a collagenous transmembrane protein—ectodysplasin—and its two receptors, that are involved in epithelial-mesenchymal interactions, hair follicle morphogenesis, [11] ectodermal appendage formation and organogenesis during the initiation of development.5 In hidrotic ED the affected gene has been identified to be GJB6 on chromosome 13q, which encodes for connexin-30.[6]

Clinical features

Hypohidrotic ED

Forehead appears square, with frontal bossing, prominent supra-orbital ridge, depressed nasal bridge (saddle nose), hypoplastic and depressed midface, pointed chin, everted and protuberant lips. [5] malar hypoplasia and prominent low set ears.12 high arch palate and cleft palate.[2]

Anodontia or hypodontia involving lower incisors and premolars followed by the upper premolars and incisors. The upper cuspids and first upper and lower molars are usually formed, delayed teeth

eruption; radiographs show teeth with abnormality in shape and structure. Enamel is thin, rarely hypoplastic. Tooth crowns are small, abnormal in shape and constricted in cervical area. Upper incisors and cuspids are always conical or pointed.

Taurodontism commonly affecting second deciduous molars. Missing alveolar processes and if present is in form of very thin bony ridge. Poor development of mucous gland in the respiratory and gastro-intestinal tract may result in increased susceptibility to respiratory infections. Dry mouth, dry nose, reduced salivary secretions,[5] purulent rhinitis, dysphonia and diarrhoea.[3] Other oral findings seen are reduced salivary buffer capacity , increased number of bacterial cultures and increased predisposition to dental caries.[2]

The scalp, eyebrows and eyelashes show sparse hair, decreased body hair in pubic and axillary region. Nail dystrophy includes slow nail growth, split nails, longitudinal ridging, thinning and superficial peeling. Hypopigmentation, dry or smooth velvety skin texture is seen. Decreased sweating and heat intolerance which may lead to hyperthermia resulting in sudden infant death or brain damage. Episodes of hyperpyrexia and severe respiratory infections are life-threatening.[5]

Other common signs are short stature, eye abnormalities, decreased tearing, photophobia and atopic eczema, intelligence is normal, 1 hyperpigmentation and finely wrinkled, periorbital skin appearing prematurely aged, 3 incontinentia pigmenti and cardiomyopathy.5

Hidrotic ED

Hidrotic Ectodermal Dysplasia includes nail and hair defects, palmoplantar dyskeratoris with normal facies, normal sweating and no specific dental defect.[13]

Diagnosis

The presence of classical facies, anodontia, scanty hair and thin dry skin are characteristic enough for the hypohidrotic ED and investigative support for the diagnosis was not considered necessary. [9] Affected men present an easily recognizable facies, also referred to as an 'old man' facies, scaling of the skin, scalp and body hair is sparse. The carrier female has some phenotypic expressions which are same as those in affected males. One third of the carriers appears healthy, another third show mild symptoms, and the last third exhibits significant symptoms, but not as severe as the affected males.[5]

Some disorders like Ectrodactyly-ectodermal dysplasia clefting syndrome, Rapp-Hodgkin syndrome and Ellis-van Creveld syndrome14 have few overlapping features but presence of classical triad (hypodontia, hypotrichosis, and hypohidrosis) is sufficient to arrive at a diagnosis of ED.

Investigations

Extraoral radiographs usually shows absent tooth buds, specific deformities like frontal bossing. Starch powder test is used to evaluate sweating response. Skin biopsy demonstrates absence or hypoplasia of sweat glands and reduction in the number of sebaceous glands and hair follicles [4] which can be useful for prenatal diagnosis of Hereditary ED.[1] Linkage analysis is used for prenatal and early neonatal diagnosis.5 Identification of female carriers and to distinguish between the X-linked form and recessive form genetic testing is a suitable tool.[14]

Hereditary ED accompanying immunodeficiency shows hypogammaglobulinemia, impaired lymphocyte proliferation and cell-mediated immunity. Hypohidrosis and reduction in the number of eccrine glands can be identified by performing sweat pore counts, pilocarpine iontophoresis, and skin biopsy.[15] Decrease in number of sweat pores leads to disruption of dermal ridges that can be seen by dermatoglyphic studies. ED cases with known genetic mutation, karyotyping can be performed to identify the genetic defects.[16]

Prognosis

About 30% of patients die during the first 2 years of life due to hyperpyrexia or fulminant respiratory infections.12 However, normal life expectancy and a normal intelligence is seen in majority of hereditary ED cases.[5]

MANAGEMENT

Hereditary ED should be managed by team comprising of pediatrician, pediatric dentist, prosthodontist, dermatologist, otolaryngologist, speech therapist, psychologist, and social worker. The entire family can be assisted by psychotherapy or counseling .[5] The role of pediatric dentist in patient counseling is extremely vital to boost the self-confidence of the patient.

Conventional prosthodontics treatment for ED has consisted for various combinations of overdentures, complete or partial removable dentures, or fixed partial dentures. Along with conventional endosseous implants, zygomatic implants can also be used in cases of severely resorbed maxilla.[17]

Prosthetic treatment is required for preserving alveolar bone, mastication, speech, development of a positive self-image and overall oral health.[15] Dentures require periodic changes to accommodate alveolar growth, erupting teeth and rotational jaw growth. Removable prosthesis may get lost or broken; also bone destruction of an already hypoplastic alveolar may occur because of removable prosthesis, so crowns and bridges are preferred. Impaired salivary secretion rates, increased caries incidence, friable oral mucosa may affect the ability to wear removable prosthesis.[5] Salivary substitutes, topical fluorides and nutritional support may be helpful in such cases.

Osseo-integrated implants should be placed after cessation of growth.[11] Multiple issues must be considered before implant placement like position of the maxilla and the mandible relative to one another, the remaining teeth in the arch and their positions, the volume of bone present, and the age of the patient before implant placement.[18] The new modalities, like implant supported overdentures can be preferred over conventional removable dentures as they show excellent stability and retention.[14]

Light clothing, cool-water spray bottle, air conditioning, artificial tears and application of petrolatum for nasal mucosa protection are suggested for ED patients. They are advised against overexposure to warm temperatures and vigorous physical activities. Moisturizers are prescribed to prevent xerosis or eczema. Toupees may serve to improve the appearance of patients with severe hair deficiency. Orthodontic treatment is required in some cases for cosmetic and nutritional purpose. Cleft lip or palate and other midfacial defects may need surgical corrections to improve form and function.[1]

Excellent oral hygiene is important to achieve desired results in such patients. Periodic oral check-up and follow-up appointments are essential for future management.

Conclusions:

Dentists are often the first who diagnose these patients .When dental professionals come across patients with multiple dental agenesis, they should look for other signs of hereditary ectodermal dysplasia to help in diagnosis and further manage them appropriately so as to enable the patients to develop physically, emotionally, and socially like other healthy individuals.

Functional and prosthetic appliances help to enhance the facial growth [11], hence early dental evaluation and management is necessary.[12] Dental treatments is crucial for the psychological development of such patients as their look can be dramatically changed. The social anxiety associated with partial or full dentures in young patients can be eliminated by using dental implants.[3]

IMAGE LEGENDS

Fig 1- Patient of hereditary ectodermal dysplasia presenting with prominent supraorbital ridges, periorbital pigmentation and sparse facial hair.

Fig 2- Extensor surface of upper limb showing dry, scaly skin with sparse hair and dystrophic nails.

Fig 3- Clinical picture of partial anodontia. Note conical crown of lower anterior teeth. Generalised hypoplasia and spacing is also seen.

Fig 4- OPG showing partial anodontia with conical shaped anterior teeth. Over retention of deciduous teeth is seen. Also note external resorption and Taurodontism

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