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HEREDITARY ECTODERMAL DYSPLASIA – A REPORT OF TWO CASES

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ABSTRACT:

Hereditary Ectodermal dysplasias are a group of X-linked recessive inherited disorders characterised by primary defects in the development of two or more tissues derived from embryonic ectoderm. The tissues which are affected include skin, hair, nails, eccrine glands and teeth. Here, we report two cases of hypohidrotic ectodermal dysplasia in male siblings.

Key words: hereditary ectodermal dysplasia, partial anodotia
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INTRODUCTION:

Hereditary ectodermal dysplasias (HEDs) are characterised by defective formation of one or more structures derived from ectoderm. It was first described by Thurman in 1848 [1] and was coined by Weech in 1929[2]. Freire-Maia redefined it as a developmental defect which at embryonic level affects the ectoderm and therefore the tissues and structures derived from it [3]. Thus it affects the development of keratinocytes and cause aberrations in the hair, sebaceous glands, eccrine and apocrine glands, nails, teeth, lenses and conjunctiva of the eyes, anterior pituitary gland, nipples and the ears [3].

CASE REPORT:

A 19 year old male and his male sibling of 17 years of age, both born of non-consanguineous marriage presented with complaint of missing teeth since childhood. Physical examination revealed recession of hair line all along with

sparse, thin light brown brittle scalp hair. On extra oral examination, prominent forehead, sunken cheeks with prominent supra orbital ridges, thin eyebrows, hyper pigmentation around eyes and saddled nose were noted. Lips were everted and prominent in both of them. Eye lashes appeared normal. Both of them gave the same clinical history of almost complete absence of sweating from birth. Their maternal grandfather had similar complaints. Dental examination revealed partial anodontia and maxillary incisors were conical in shape [Figure-1, Figure-2 and Figure-3]. They both complained of xerostomia. The combined clinical and dental findings pointed towards diagnosis of hypohidrotic ectodermal dysplasia.

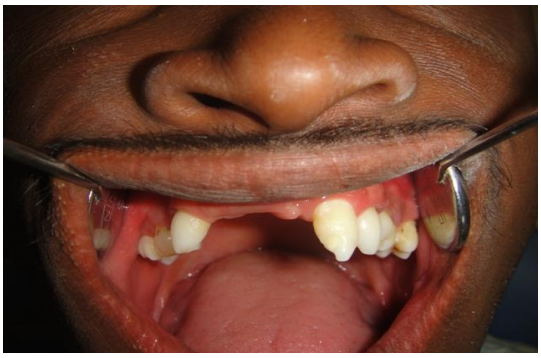

Figure 1: Partial anodontia and conical shaped anterior teeth in the elder brother

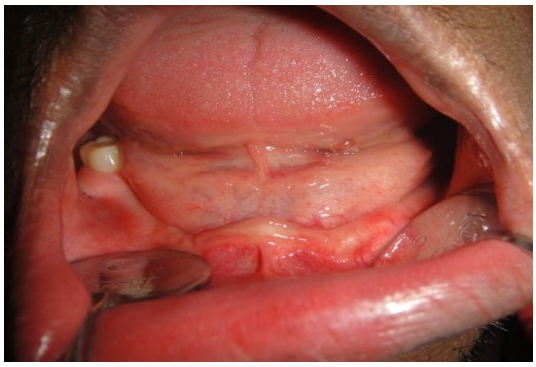

Figure 2: Only lower right second molar tooth present in the mandibular arch in the elder sibling

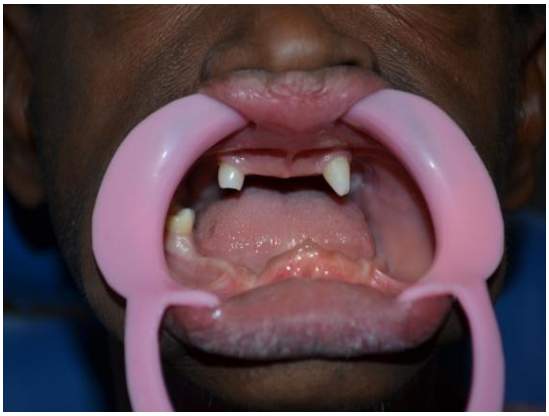

Figure 3: Partial anodontia. Only maxillary canines and right lower mandibular second molar tooth are present in the younger sibling

DISCUSSION:

The ectodermal dysplasias (EDs) are congenital, diffuse and non progressive disorders. More than 192 distinct disorders have been described till date [4]. It is typically inherited as a cross-linked recessive trait so that frequency and severity of the condition is more pronounced in males than in females.

The disorder might occur during the first trimester of pregnancy. If it is severe, it appears before the sixth week of embryonic life and consequently the dentition will be affected. After eighth week other ectodermal structure may be affected [5]. Genetic studies of more than 300 cases have revealed X linked mode of inheritance with its gene locus being Xq11-21.1; the gene is carried by the female but manifested in male [3].

However, there are reports of multiple siblings being affected and of females suffering with this condition [3]. The gene that causes hidrotic ectodermal dysplasia (Clouston's syndrome) has been identified to be the GJB6, which encodes for connexin-30. GBJ-6 has been mapped to the pericentromeric region of chromosome 13q. Mutations of the gene PVRL1, encoding a cell-to-cell adhesion molecule/herpes virus receptor, have been reported in those with cleft lip/palate ectodermal dysplasia [6].

A definite classification of ectodermal dysplasia is difficult to formulate since many of the syndrome that involve (ED) have overlapping features. A simple attempt made by Nelson included five categories, namely Hypohidrotic (anhidrotic), Hidrotic (Clouston's syndrome), Ectodactyly ectodermal dysplasia (EEC) syndrome, Rapp-Hodgkin syndrome and Robinson's disease [7].

From the clinical point of view two main forms have been distinguished: Hypohidrotic form / Christ-Seimens-Tourian syndrome and Hidrotic form / Clouston syndrome.

The hypohidrotic form exhibits the classic triad-hypohidrosis, hypotrichosis and hypodontia. Usually X-linked recessive inheritance is seen. Males are affected severely, while females show only minor defects [6]. In the hidrotic form teeth, hair and nails are affected. The sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported [6, 8].

Table 1 - Difference between the hidrotic and hypohidrotic forms of ectodermal dysplasia [9]

	Hidrotic	Hypohidrotic
Mode of inheritance	Most often autosomal dominant	Most often autosomal recessive
Scalp hair	Soft, dawny ,colour is darker	Fine in texture, fair and short
Teeth	Anodontia to hypodontia	Anodontia to hypodontia
Lips	No abnormality	Protruding
Sweat glands	Active	Reduced to absent
Nasal bridge	No flattening	Underdeveloped
Nails	Dystrophic nails	No abnormality
Eyebrows	Frequently absent	Absent
Eyelashes /pubic/axillary hairs	Scanty /absent	Variably affected

The typical facies is characterized by frontal bossing, sunken cheeks, saddle nose, thick everted lips, wrinkled hyper pigmented periorbital skin and large low set ears. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia and delayed eruption of permanent teeth. Fine, sparse, lustreless fair hair over scalp is seen in most of the patients. Onychodystrophy may be seen but is not common. Extensive scaling of the skin and unexplained pyrexia and heat intolerance due to anhidrosis occurs. Intelligence is normal [4]. Encourage frequent consumption of cool liquids to maintain adequate hydration and thermoregulation and advised cool clothing. The treatment usually comprises of complete restoration of function and aesthetics to normalise the vertical dimension and provide adequate support to the facial soft tissues. The options may include fixed, removable or implant prosthesis, singly or in combination.

The presentation of facial deformity, dry skin, and sparse hair in this report is similar to previous reports. These features are due to anomalies of the skin appendages which include the hair follicles, sweat glands and sebaceous glands [10, 11, 12, 13].

The intolerance of heat and hyperthermia observed in our patients is similar to the previous reports which are again due to absence of sweat glands [11, 12, 13].

In developed countries diagnosis pertains to laboratory identification of genes and mode of inheritance of mutated genes associated with recessively X chromosome or autosomal dominant or recessive. This may be difficult in developing countries like India where such facilities are insufficient and it requires further probing through genetic analysis.

In our case, the patients showed typical facies with dental manifestations, sparse scalp hair and heat intolerance. As both male siblings and their maternal grandfather were affected, we can ascertain the mode of inheritance to be X-linked recessive.

The most important aspect to be considered in these patients is the psychological impact on the child and parents due to absence of teeth. The principal aim of dental treatment is to restore missing teeth and bone, since it provides good esthetics, phonetics and masticatory comfort. It also helps patients develop good psychological self-image. Treatment plan generally include removable or fixed partial prosthesis. Dental implants may also be successfully employed to support and retain teeth.

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