Anhidrotic Ectodermal Dysplasia – Report of Two Cases.

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ABSTRACT

Here we report two cases of anhidrotic ectodermal dysplasia in a family presented to us with intermittent fever, developmental delay, frontal bossing, hypohydrosis, sparse hair and oligodontia. Ectodermal dysplasias are a large hereditary group of disorders which are usually manifested as X-linked recessive hypohidrotic ectodermal dysplasia (HED) and has a full expression in males, whereas females show little to no signs of the disorder. Ectodermal dysplasia are group of rare genetic disorders characterized by faulty development of ectodermal structures and thought to be due to embryonic defect in ectodermal development. Patients frequently consult dentists for delay in tooth eruption so the appropriate awareness of the disease among dentists is essential for early diagnosis.

Keywords: Hypohidrotic Ectodermal Dysplasia, hypodontia, hypohydrosis, oral rehabilitation.

INTRODUCTION

Ectodermal dysplasias (ED) are groups of inherited genetic disorders due to abnormalities of the two or more ectodermal structures such as the hair, teeth, nails, sweat gland, craniofacial structure, digits and other part of the body.[1] Anhydrotic ED, also known as hypohydrotic ED (HED) and it might be inherited in any form of several genetic patterns including autosomal-dominant, autosomal-recessive, and X-linked nodes.[2]

Although more than 170 different subtypes of ectodermal dysplasia have been identified, these disorders are considered to be relatively rare with an estimated incidence of 1 case per 100,000.[2] The two most common types of ectodermal dysplasias are the X-linked recessive hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome).[3]

It is characterized by the triad of signs, which comprises of sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhydrosis or hypohydrosis). Patients may present with life threatening hyperpyrexia and heat intolerance. Patients also have characteristic facies with prominent frontal bossing, a saddle nose, sunken cheeks and protruding lips. Diagnosis is mainly clinical and molecular confirmation is desired but not necessary to establish the diagnosis.

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CASE-SERIES

Case 1: An 8-year-old underweight (16 Kg) male child reported to our institution for evaluation of growth retardation, hypotrychosis, hypohydrosis, anodontia, progressive facial changes, heat intolerance and intermittent hyperpyrexia since infancy. The cause of the fever had not been established on multiple occasional. For evaluation of fever, we admit the patient but no rise of temperature was recorded. There was no history of consanguineous marriage between the parents. On extra oral examination, the patient had dry, smooth skin with sparse hair on the body and scalp. Hairs were fine in texture, very small in length and lighter in colour. Frontal bossing, periorbital, perioral wrinkling, and hyperpigmentation were evident. Prominent supraorbital ridges, mid-facial hypoplasia with depressed nasal root and bridge, everted lips, small and outwardly placed ears were also present. Both upper and lower eyelids showed sparse eyelashes [Figure 1]. There were no nail changes. Oral examination revealed missing teeth in the maxillary and mandibular arches. Systemic examination was normal.

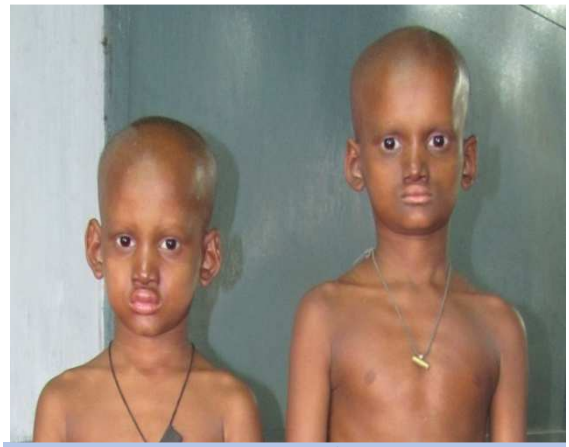

Figure 1: Characteristic appearance of HED.

Case 2: A 6-year-old underweight (14 Kg) male child, younger sibling of the previous child presented at the same time with similar complaints and clinical features except for the presence of conical teeth. Unlike to his elder sibling who had anodontia younger sibling had hypodontia (4 teeth in maxillary arch) and conical ''peg'' teeth [Figure 2].

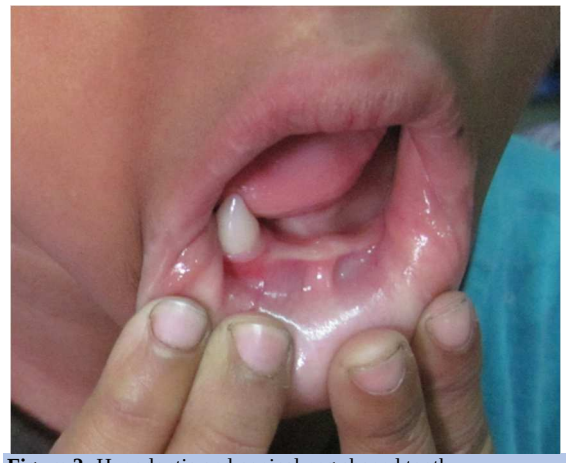

Figure 2: Hypodontia and conical peg shaped tooth.

All routine blood tests were normal. Skin biopsy was done in both cases and it showed thinning of epidermis, absent skin appendages like sweat glands, sebaceous glands and hair follicles [Figure 3]. Based on these clinical findings a diagnosis of hereditary hypohydrotic ectodermal dysplasia was made. Apart from some medical management, they were referred to dental surgeon for dental reconstruction.

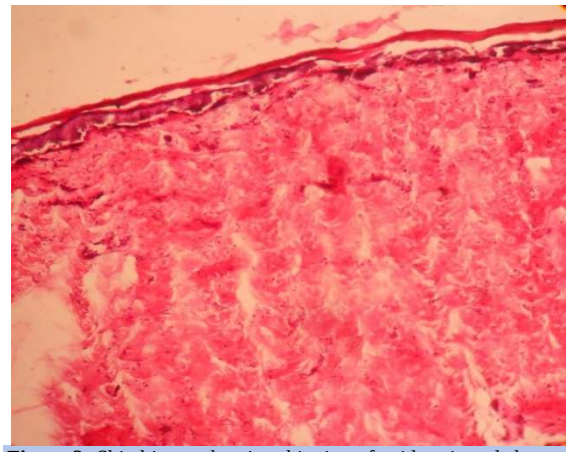

Figure 3: Skin biopsy showing thinning of epidermis and absent skin appendages.

DISCUSSION

The Ectodermal dysplasias are a group of inherited disorders that share in common developmental defects involving at least two of the major structure classically holds to derive from the embryonic ectoderms hair, teeth, nails, sweat glands.[4]

HED is typically inherited as an X- linked recessive disorder (gene locus Xq 13.1) so the frequency and severity of the condition are more pronounced in males than in the females. The disorder has been associated with mutations in at least three genes that also are involved in the activation of the transcription factor NF-kappa B, which regulates the expression of the genes that control immune and stress responses, inflammatory reaction, cell adhesion and protection against apoptosis.[5]

Patients with HED generally have prominent supraorbital ridges, frontal bossing, and a saddle nose. The maxilla may be underdeveloped and the lips are thick and prominent. The nose may appear pinched and the alliance appears hypoplastic. The patient may resemble like an old edentulous person.[6,7] The skin is usually dry, scaly, and easily irritated as a result of poorly developed or absent sebaceous glands. Sweat glands may also be absent or few in number or non-functioning which may result in increased body temperature. Scalp hair may be absent, sparse, very fine pigmented, or abnormal in texture. Eyebrows, eyelashes, and other body hair may also be sparse or absent. When hairs are present, they may be fragile, dry, and generally disorderly because of the lack of sebaceous glands. Finger and toe nails are usually normal.[7] Orofacial characteristics of this syndrome include anodontia or hypodontia, hypoplastic conical teeth, underdevelopment of the alveolar ridges, frontal bossing and depressed nasal bridge. Teeth are generally small, conical, tapered (peg shaped), and widely spaced. The complete syndrome does not occur in females but females may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities.[8] The major concern seen in these patients is the lack of teeth hyperpyrexia and the special appearance[4], 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. Some infants have a premature look with scaling of the skin. This can give a clue to the diagnosis.[4] The oral and extra oral manifestations of our cases are in accordance with the exiting literature.

Medical care depends on involved ectodermal structure. In our patients they had hyperpyrexia. So, they should be advised to take frequent cooled water to maintain hydration and thermoregulation. Xerostomia and reduced lacrimation should be treated with artificial saliva and tears respectively. For dental reconstruction, they should be referred to dental surgeon. Prognosis of ED is very good and life expectancy is generally normal except associated immunodeficiency.[9]

Bullet point

Patients with HED often first consult to paediatric dentist for delay in eruption of teeth so the awareness regarding the peculiarity of disease is essential in dentistry for early diagnosis and restoration of normal function as much as possible.

CONCLUSION

Awareness about Ectodermal dysplasias, a rare group of genetic disorders among dentists and physicians is essential for early diagnosis of the disease.

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