

Hypohidrotic ectodermal dysplasia and finger clubbing – An unknown association

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Submitted: 26-12-2013

Revised: 21-02-2014

Published: 30-05-2014

ABSTRACT

The ectodermal dysplasias are heterogenous group of inherited disorders with primary defects in tissues derived from embryonic ectoderm such as hair, tooth, nail and sweat gland. This disorder is broadly divided into hypohidrotic or anhidrotic and hidrotic forms. Here, we present a classical x-linked hypohidrotic ectodermal dysplasia with finger clubbing, the association of which is not being reported earlier.

Key words: Hypohidrotic ectodermal dysplasia, Finger clubbing, Hidrotic ectodermal dysplasia

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INTRODUCTION

The ectodermal dysplasias (ED) are large, heterogenous group of inherited disorders that are defined by primary defects in the development of two or more tissues derived from embryonic ectoderm.¹ The tissues primarily involved are skin, hair, nail, sweat gland and tooth. These disorders are clinically divided into hypohidrotic or anhidrotic and hidrotic forms and each of these forms are subdivided into different syndromes, based on defects of different tissues. Here, we report a patient with classical hypohidrotic ectodermal dysplasia (HED) with grade two finger clubbing and such association is unknown.

CASE REPORT

A 45 years male attended outpatient department of our hospital with the complaint of no sweating, difficulty in tolerating hot environment and loss of teeth since childhood. On general examination, pulse and blood pressure were 92/min and 120/80 mmHg respectively, mild pallor and grade two finger clubbing were present. Examination of skin showed no evidence of sweat, even in hot and humid environment, thickened skin over palm and sole. The scalp and body hairs were thin, sparse, light in colour, curly and even twisted. The growth of the beards were poor. The eyelashes and eyebrows were absent, pubic

and axillary hair were sparse. The nails were thick, abnormal shaped with subungual hyperkeratosis. Total number of teeth present were three, two upper central incisor and one upper left lateral incisor and they were peg shaped or pointed. The appearance of head and face showed frontal bossing, broadened and depressed bridge of nose. There was impairment of hearing with partial sensorineural deafness in both ear. Systemic examination was normal.

His parents and one sister were healthy, but all of his three brothers were suffering from similar problems and there was no history of consanguineous marriage among parents. Laboratory examination revealed no abnormality. So, with all these findings, a diagnosis of hypohidrotic ectodermal dysplasia was made.

DISCUSSION

The ectodermal dysplasias (ED) are congenital, diffuse and non-progressive. More than 170 different syndromes have been identified.² The ectodermal dysplasia was first reported in 1848 by Thurnam,³ but term ectodermal dysplasia was not coined until 1929 by Weech.⁴ The current classification of ectodermal dysplasias are based on clinical features. Freire-Maia and Pinheiro proposed the first classification system of the ED in 1982 with additional updates in 1994 and 2001.^{5,6} Their original classification systems are based on

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Figure 1: Typical facial appearance of hypohidrotic ectodermal dysplasia with sparse hair, beard, eyelashes and eyebrows; depressed and broadened bridge of nose and reduced number of teeth

presence or absence of ectodermal structure abnormality. In this classification, '1' indicates hair abnormalities; '2' dental abnormalities; '3' nail abnormalities and '4' sweat gland dysfunction or dyhidrosis. The ectodermal dysplasias are classified into group A disorder, if defects are in at least 2 of the above 4 classic ectodermal structures and group B disorder, if defects are in one classic ectodermal structure with defect in other ectodermal structures like ear, lip etc (indicated by '5'). Since our patient had abnormality of hair, teeth, nail and sweat glands, his ectodermal dysplasia fits in group A and subgroup 1-2-3-4 with additional defect in ear. Ninety five percent of hypohidrotic ectodermal dysplasia (HED) have the X-linked recessive inheritance.⁷ The remainder five percent have either autosomal recessive or dominant inheritance. Since our patient, who was male and three of his brothers were affected, his lone sister being unaffected and there was no history of consanguineous marriage among parents, the probable mode of inheritance in this present case was X-linked recessive. This x-linked HED, which is also known as Christ-Siemens-Touraine syndrome is commonest form of ectodermal dysplasia. HED is caused by mutation of gene that encodes several proteins with roles in the ectodysplasin signal transduction pathway. Mutation in the ectodysplasin-A (EDA) gene is responsible for the X-linked recessive form of HED.⁸ Mutation in the gene encoding the EDA receptor and the adaptor protein that associates with EDA receptor's death result in autosomal dominant and autosomal recessive forms of HED respectively.⁹ Clinically, HED is characterised by sparse or absent sweat gland resulting in decreased or absent sweat production as well as hypotrichosis and oligodontia with peg shaped teeth, as seen in the present case also. Because of their diminished ability to sweat, patient with HED have a tendency to develop hyperthermia with physical exertion or exposure to a warm environment and often present with recurrent high



Figure 2: Grade two finger clubbing in hypohidrotic ectodermal dysplasia

fever. The scalp hairs, eye brows and eyelashes are sparse, fine and often lightly pigmented, which were also seen in our patient. Similar to our patient, HED patient usually have a characteristic facies with frontal bossing, a saddle nose and everted lips (Figure 1). There was presence of grade two finger clubbing in our patient, which is usually a feature of hidrotic ectodermal dysplasia (Figure 2). Hidrotic ectodermal dysplasia (HED2) or Clouston Syndrome is characterized by partial or total alopecia, dystrophy of the nails, hyperpigmentation of the skin (especially over the joints) and clubbing of the fingers.¹⁰ To the best of our knowledge, clubbing in hypohidrotic ectodermal dysplasia has not been reported earlier.

There is no specific treatment for this disorder. The things which can be done are to wear a wig and dentures to improve appearance, use artificial tears to prevent drying of eyes, spray the nostrils with saline nose spray, live in a cooler climate and take cooling water bath or use water spray to keep a normal body temperature.

CONCLUSION

We report this case because of unknown association between hypohidrotic ectodermal dysplasia and finger clubbing. There is no specific treatment for hypohidrotic ectodermal dysplasia. The protective measures and timely interventions of complications can prevent early death.

ACKNOWLEDGEMENTS

All authors are thankful to the concerned patient, for his written consent to publish this article.

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Authors Contribution:

AS – Concept, definition of intellectual content, literature search, participated in clinical data collection, manuscript preparation; **AB** – Definition of intellectual content, manuscript revision; **SKD** – definition of intellectual content, literature search, manuscript revision; **KM** – literature search, clinical data & photograph collection, manuscript preparation; **SKS** – literature search, clinical data & photograph collection, manuscript revision.

Source of Support: Nil, **Conflict of Interest:** None declared.