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Hypohidrotic Ectodermal Dysplasia with Arachnodactyl and Palmoplantar Keratoderma

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Abstract

Hypohidrotic ectodermal dysplasia is a rare disorder characterized by hypohidrosis hypotrichosis, and hypodontia. We present an 8 year old female child with clinical picture suggestive of autosomal recessive hypohidrotic ectodermal dysplasia with an unusual manifestation including arachnodactyl and palmoplantar keratoderma.

Introduction

Hypohidrotic ectodermal dysplasia, also known as Christ-Siemen-Touraine syndrome is an X-linked recessive disorder. The full expression of the disorder occurs mainly in males; females serve as carrier of the genetic defect with a small fraction presenting with minimal to significant clinical findings [1]. The existence of autosomal recessive form of the disorder has been proposed on the basis of the presence of severely affected females with unaffected parents in highly consanguineous families [2]. The syndrome is characterized by congenital dysplasia of one or more ectodermal structures and includes defective hair follicles, anodontia or hypodontia with conical teeth; dry skin and unexplained high fever due to deficiency of sweat glands.

Case report

An 8 year old girl born of consanguineous marriage presented to the outpatient department of Dermatology, STD & Leprosy of SMHS Hospital (Associated teaching hospital of Government Medical College Srinagar) with poor hair growth and increased hair fragility since infancy. She was not able to tolerate the heat of summer and had recurrent episodes of unexplained high fevers. There was no history of convulsions, recurrent upper respiratory tract infection and dysphagia. There was no evidence of physical and mental retardation.

On examination there was diffuse alopecia with twisted dry lusterless and brittle short hair (**Fig.1**). The eye brows and eyelashes were scanty and there was hypotrichosis on rest of the body. There was ectropion of both eyes with epiphora (**Fig.2**). Hypodontia with conical lower incisors

were present (**Fig.3**). There was no evidence of dental caries but there was generalized xerosis with fine dry scaling which was more prominent on the shins. Hands and feet showed arachnodactyl with diffuse palmoplantar keratoderma (**Fig.4 and 5**). Nails were thin and ridged. There was no evidence of visual and hearing impairment. Her parents and two older male sibs were normal. The palmer skin biopsy confirmed the absence of sweat and sebaceous glands and revealed orthokeratotic hyperkeratosis and acanthosis of the epidermis. The Starch Iodine test on the back demonstrated lack of sweating.



Fig 1: Diffuse alopecia on the vertex with dry lusterless hair at the margins of the scalp.



Fig 2: Sparse eyebrows and eyelashes with ectropion of both eyes.



Fig 3: Hypodontia with conical lower incisors.



Fig 4: Diffuse palmer keratoderma with arachnodactyly of hands and feet.



Fig 5: Diffuse planter keratoderma.

Discussion

Hypohidrotic ectodermal dysplasia (HED) was first described by Thurman in 1848 [3]. Majority of reported cases of HED have been males with an X-linked recessive mode of inheritance and are known as Christ-Siemens-Touraine syndrome [1]. Affected females are heterozygous due to one normal and one affected X-chromosomes [1]. These carrier females are usually asymptomatic or may express a variable phenotype of this condition in view of random inactivation of one of the X-chromosome during early embryogenesis [4]. There is another less common form of HED with autosomal recessive mode of inheritance with clinical resemblance to Christ- Siemens-Touraine [1,5].

The syndrome is characterized by impaired or absent sweating, hypotrichosis and hypodontia. The classical facial features described include prominent forehead and chin, sunken cheeks with saddle nose, thick everted lips and large ears. The sparse lusterless fragile short hair is a striking feature of the disorder. The eyebrows, eyelashes and rest of the body hair are also sparse or totally absent. The dental abnormalities may present as hypodontia or andontia. The presence of conical

incisors and canines are characteristic. The gums are hypoplastic with normally developed jaws. Nails are thin, ridged and brittle.

Heat intolerance or unexplained hyperthermia occurs due to reduced or absent sweating. The Starch Iodine test demonstrates total absence or decrease in sweating. Affected females with X-linked recessive heterozygous inheritance, carrying one normal X chromosomes and one affected chromosome; demonstrate sweating along the Blasckho's lines, a phenomenon based on lyonization [6].

The otorhinolaryngological features include atrophic rhinitis, crusted nasal secretion, ozaena, recurrent upper respiratory infection, hoarseness, asthma and hearing defect [7]. The ocular manifestations include corneal and lenticular opacities [8]. The poor development of lacrimal, salivary and mucus glands of the gastrointestinal tract results in conjunctivitis, xerostomia and dysphagia. Diffuse palmoplantar keratoderma has been recent addition to the syndrome [9].

There is retardation in physical and mental development in one third of cases.

Our patient did not have the typical facies of the syndrome but had an additional feature of ectropion, arachnodactyl and diffuse palmoplantar keratoderma which are unusual manifestation of this genodermatosis.

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