

A 'Missed' Diagnosed Case of Christ-Siemens-Touraine Syndrome

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Abstract

Christ-Siemens-Touraine syndrome also known as Anhidrotic ectodermal dysplasia, is a rare genetic disorder, occurring in 1-100,000 live male births. A two year old male child with classical signs and symptoms of the disease, affecting skin, hairs, nails and teeth, misdiagnosed initially as a case of seborrhic dermatitis, and later as tuberculosis, is described here. Treatment includes counselling the family, skin care, avoiding high ambient temperature, and prosthodontic measures. Diagnosis as early as possible, based mainly on clinical features can prevent mortality due to hyperpyrexia and respiratory infections. Beyond early childhood, life expectancy is normal to slightly reduced.

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Introduction

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures, such as skin, hair, nails and teeth. Till date about 160 clinically and genetically distinct types of EDs have been described¹. Collectively, its prevalence is stated as seven cases per 10,000 births².

Anhidrotic Ectodermal Dysplasia, also known as Christ-Siemens-Touraine syndrome, is a rare genetic disorder, transmitted mostly as X linked recessive form. It is the most common type of EDs³. It was first described by Thurnam in 1848 and later in the 19th century by Darwin⁴. An autosomal recessive form has also been described⁵. The incidence of X linked recessive form is estimated at 1-10 in 100,000 live male births, and the carrier incidence is around 17.3 in 100,000 women³. It manifests as a triad of hypohidrosis or complete absence of sweat glands, hypotrichosis and anomalous dentition.

A two year old male child with classical signs and symptoms of the disease, but still misdiagnosed due to lack of awareness among clinicians, is being reported here.

The Case

Two year old male child, product of non-consanguineous marriage, presented to us in OPD with complaints of intermittent episodes of high fever, never recorded, developing on sun exposure and relieved on keeping the child indoor. Father gave definite history of lack of sweating since birth. Baby was born as full term, appropriate for gestational age, with plastic wrap appearance of skin at birth. First

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six months of life were uneventful, then parents noticed cracking and peeling of skin; child was then diagnosed as having seborrheic dermatitis and given topical medications. At 10 month of age, child was again shown to a doctor for fever, poor growth and pale appearance of skin. At that time, child was prescribed anti tubercular treatment which he took for six months. Milestones were appropriately achieved. As there was no improvement child was referred to our hospital. History of poor dentation was given. There was no similar family history. On examination we found pallor, sparse and blonde scalp hairs and eyebrows, low set ears, frontal bossing, thick everted lips, peg shaped upper two incisors, absent other teeth. Skin showed cracking, nails were small and dysplastic. Child weighed 8.6 Kg (<-3 SD according to WHO charts), Height was 79 cm (between -2 to -3 SD), Head circumference was 45.5 cm. Systemic examination was normal. Blood investigation showed microcytic hypochromic anaemia (Hb=9 gm/dl). Renal and liver function tests were normal. Dermatologist's opinion was taken who suggested coconut oil application and counselling of patient. Skin biopsy was done which showed thin and flattened epidermis with rudimentary sweat glands. Parents were counselled about the nature of the disease and avoidance of high ambient temperature. They were not ready for prosthodontic intervention, as suggested by dentists due to financial issues. Child was discharged after building up nutritional status.



Fig 1: Showing sparse and blonde scalp hairs and eyebrows, low set ears, frontal bossing, & thick everted lips



Fig 2: Shows peg shaped upper two incisors

Discussion

Ectodermal dysplasia term was coined by Weech in 1929². X linked recessive anhidrotic/ hypohidrotic ectodermal dysplasia is its most common type (80%)³. Its typical phenotypic features include soft, thin, dry skin, hyperkeratotic palms and sole, thin sparse, light colored hairs, lack of sweating, oligodontia, conical shaped teeth, wide midline diastema, frontal bossing, depressed nasal bridge, prominent supraorbital ridges, prominent and obliquely set ears, depressed midface, protuberant lips, dysplastic nails, and absent sweating. Growth failure is also common². Delay in dentition provides an early opportunity to diagnose the condition. The upper normal limit for the first tooth to appear is 15 months⁶. Carrier females do not show the complete syndrome⁴. Dental examination and sweat testing can help in identifying carrier females³. Genetic analysis, though not available everywhere can be used for identification of carrier females⁷. After initial few years, there is a general improvement in health status; but the quality of life later is badly affected by poor dentition and facial features³. Prosthodontic measures are indicated as early in life as possible, preferably before five years of life; which leads to better self-image, mastication and linguistic development⁸. Mortality rate as high as 30% has been reported in literature during first two years due to respiratory infections and hyperpyrexia^{2,9}. Beyond early childhood, the life span of a patient with one of the common types of ectodermal dysplasia is normal to slightly reduced^{2,3}.

Conclusion

CST syndrome, though rare can be diagnosed on the basis of clinical features. As clinicians we should be

aware of the condition so that mortality can be prevented due to hyperpyrexia and respiratory infections.

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