

Christ–Siemens–Touraine syndrome with palmoplantar keratoderma: A rare association

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ABSTRACT

Christ–Siemens–Touraine syndrome is a form of anhidrotic ectodermal dysplasia (ED) characterized by triad of hypodontia, hypotrichosis, and hypohidrosis. Palmoplantar keratoderma is a characteristic feature of hidrotic forms of ED. Till date, only two cases have been reported of Christ–Siemens–Touraine syndrome with palmoplantar keratoderma; here we report a similar case emphasizing this rare association.

Key words: Anhidrotic ectodermal dysplasia, Christ–Siemens–Touraine syndrome, palmoplantar keratoderma

INTRODUCTION

Ectodermal dysplasias (ED) as defined by the national foundation for ED are a group of genetic disorders in which there are congenital birth defects involving two or more ectodermal structures. These disorders are nonprogressive and diffuse having all possible modes of Mendelian inheritance. EDs are mainly divided into two groups: The hidrotic form known as Clouston syndrome and anhidrotic or hypohidrotic form known as Christ–Siemens–Touraine syndrome. The diagnosis of Christ–Siemens–Touraine syndrome is mainly clinical, manifesting as absent or reduced sweating, sparse body hair, absent to abnormally shaped tooth along with specific facial features.

Here, we report a case of Christ–Siemens–Touraine syndrome with palmoplantar keratoderma, which is more common in hidrotic form of ED.

milestones and intelligence were normal. On examination, patient was moderately built with a characteristic facial appearance—depressed nasal bridge, increased intercanthal distance, periorificial hyperpigmentation and wrinkled skin, flat and wide nose, thick everted lips, malar hypoplasia, a bulging forehead, and pointed ears [Figure 2]. Eyebrows were scarce in their outer third. The incisors were conical, pointed and widely spaced [Figure 3]. In addition, there was mild, diffuse palmoplantar hyperkeratosis [Figure 4]. Nails were normal. These findings were consistent with the diagnosis of anhidrotic ED.

DISCUSSION

The EDs are a group of inherited disorders that share in common developmental defects involving at least two of the major ectodermal structures; hair, teeth, nails, or sweat glands.

The first case of ED was reported by Thurman in the year 1848,^[1] but the term ED was later

CASE REPORT

A 22-year-old male presented with a history of generalized dryness, absence of sweating with episodes of hyperpyrexia since early childhood. The patient also complained of loss of body hair, dental abnormalities, nonprogressive hoarseness of voice, and thickness of the palms and soles. He was born out of nonconsanguineous marriage and full-term vaginal delivery. There was a family history of similar illness in his younger brother [Figure 1]. Developmental

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Figure 1: Photograph showing characteristic facial features including frontal bossing, sparse scalp and eyelash hair, prominent supraorbital ridge, depressed nasal bridge, and periorbital hyperpigmentation



Figure 2: Photograph showing abnormally shaped conical teeth

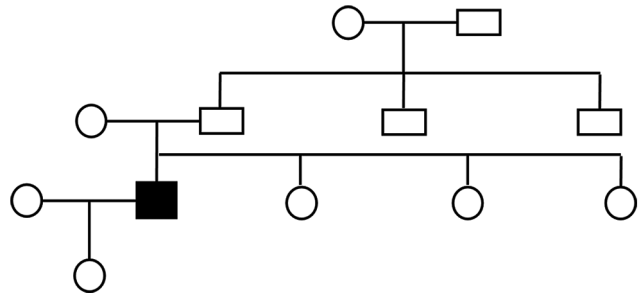


Figure 4: Photograph showing pedigree analysis



Figure 3: Photograph showing palmoplantar keratoderma

coined by Weeck in 1929. Different subgroups are created according to the presence or absence of the four primary ED defects:^[2] ED1: Trichodysplasia; ED2: Dental dysplasia; ED3: Onychodysplasia; ED4: Dyshidrosis. Based on the above, the 192 different types of EDs are categorized into one of the following subgroups made up from the primary ED defects.

Clinically, EDs are broadly divided into hypohidrotic or anhidrotic form, termed as Christ–Siemens–Touraine

syndrome, subgroup 1-2-3-4, which is X-linked recessive and characterized by the classical triad of hypodontia, hypotrichosis, and hypohidrosis.^[3] The other hidrotic form described by Clouston, subgroup 1-2-3, usually spares the sweat glands and can affect teeth, nails and hair.

Diagnosis of EDs is based mainly on the clinical grounds and other modalities include^[4]: Sweat pore counts, pilocarpine iontophoresis; radiographic evaluation of various anomalies; prenatal diagnosis using the genetic mutation analysis; and biopsy of the mucus membranes and skin. However, no such tests were done because of the patient's refusal.

Anhidrotic ED has been mapped to Xq12-q13, causing mutation in a novel transmembrane protein, ectodysplasin A, mainly involving ectodermal structures such as epidermis and its annexes (hair and nails), although nonectodermal tissue may also become involved.^[5]

The most common manifestation is the reduction or complete absence of eccrine sweat glands resulting in decreased or absent sweating, heat intolerance, and recurrent hyperpyrexia. Scalp hair is sparse, fine, lightly pigmented, and grows slowly

with scant or absent eyebrows. The eyelashes may be normal, sparse, or completely absent. Teeth abnormalities range from complete absence of teeth (anodontia) to sparse; abnormally shaped teeth.^[6] The nails are usually normal in most cases. Craniofacial features are square forehead with frontal bossing, prominent supraorbital ridge, concave midface, saddle nose, everted lips, and periorbital hyperpigmentation and fine wrinkling around the eyes.^[7] In our case these characteristic findings were present. Besides these, palmoplantar keratoderma was also present in our case, which is usually observed in hidrotic forms of ED. To the best of our knowledge, there are only two case reports of association of anhidrotic ED and palmoplantar keratoderma.^[8,9]

Neonates may demonstrate marked scaling or peeling simulating a collodion baby and in later age, eczema and xerosis are common especially over the flexures having a chronic course.

Other rare systemic manifestations include atrophic rhinitis, recurrent sinusitis and upper and lower respiratory tract infections, infantile feeding problems, hoarseness of voice, xerostomia, and impacted cerumen. These manifestations are due to diminished or absent mucous glands of the respective areas. However, no such manifestations were observed in our case.

A multidisciplinary approach is usually required for early diagnosis and prevention and management of several complications, some of which are fatal.

CONCLUSION

The presence of palmoplantar keratoderma in a case of ED supports the diagnosis of hidrotic EDs. But, in our case of anhidrotic ED, palmoplantar keratoderma was present, which is an unusual association. Till date, there are only two case reports of this association making our case an interesting one.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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