

Autosomal recessive anhidrotic ectodermal dysplasia (Christ - Seimens - Touraine syndrome) in siblings

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ABSTRACT

Anhidrotic Ectodermal Dysplasia (Christ - Seimens - Touraine Syndrome) is a rare genodermatosis comprising triad of hypohidrosis (anhidrosis), hypotrichosis and anodontia. It is usually inherited as X-linked recessive. Autosomal dominant and recessive inheritance is very rare, here we report a case of autosomal recessive anhidrotic ectodermal dysplasia.

Key words: Anodontia; Genodermatoses; Hypotrichosis; Autosomal recessive; Ectodermal dysplasia; Christ-Siemens-Touraine syndrome

INTRODUCTION

Ectodermal dysplasias (EDs) are a heterogeneous group of hereditary disorders characterized by certain shared structural and functional abnormalities in tissues derived from the ectoderm [1]. They are characterized by deficient function of at least 2 ectodermal derivatives such as skin, hair, teeth and sweat glands. 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].

Ectodermal dysplasia is divided into two types based on the number and function of sweat glands: hidrotic ectodermal dysplasia (Clouston syndrome) and hypohidrotic (anhidrotic) ectodermal dysplasia (HED) (Christ-Siemens-Touraine syndrome) [3].

Hypohidrotic ED (Christ-Siemens-Touraine syndrome) is the most frequent form of ectodermal dysplasia, and genetic defects in ectodysplasin signal transduction pathways are the basis of this syndrome [4]. It is characterized by sparse hair, heat intolerance, and excessively dry skin due to the absence of sweat glands and abnormal spiky or absent teeth [5].

We report typical features of the CST syndrome in siblings, inherited in autosomal recessive pattern, which is a rare pattern of inheritance in this syndrome.

CASE REPORT

A 7-year old boy presented with the chief complaint of child being uncomfortable in warm weather and recurrent episodes of fever in summer. He also complained of necessity to take 6 to 8 baths per day with cold water in summer. There was similar history in the younger sibling, who was a 4 year old girl. The mother stated that her children had recurrent episodes of unexplained hyperpyrexia and thirst; and were not able to sweat, and she had to apply some precautions to protect them from overheating during physical exertion or warm weather. There was no similar history in the parents or any other family members and the children were a product of non-consanguineous marriage. On general examination both the siblings had sparse, thin, light, blond hair over the scalp, scanty eyebrow and eyelashes, depressed nasal bridge (Figs. 1a and 1b), frontal bossing (Fig. 2), and prominent supraorbital ridges. Lips were dry, everted, and prominent. The skin was dry and wrinkled. Hyper pigmentation was

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Figure 1: (a) Older Sibling-Sparse and light colored scalp hair with sparse eyebrows and eyelashes, Perioral and periorbital hyperpigmentation, Depressing nasal bridge and frontal bossing. (b) Younger sibling-with striking similar face as older sibling and similar features.



Figure 2: Sparse hair and frontal bossing.

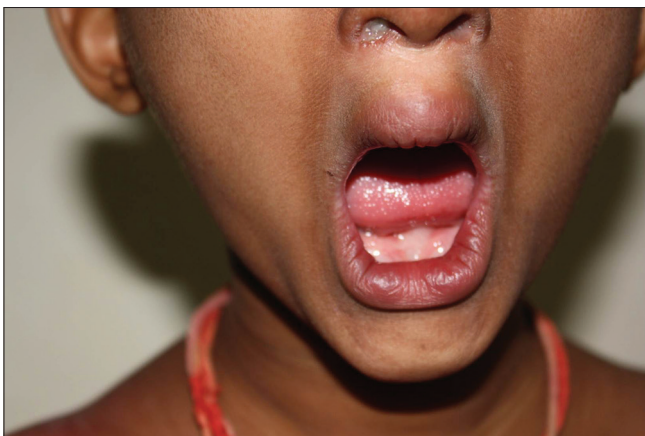


Figure 3: Complete anodontia.

evident around the eyes (Figs. 1a and 1b). There was a striking similarity in the facial features of both the siblings. Oral examination revealed complete

anodontia. (Fig. 3) An ophthalmological examination revealed absence of lacrimal puncta. They had no nail dystrophy, and their intelligence was normal. With features of anhidrosis, anodontia, hypotrichosis and typical facial features both siblings were diagnosed as a case of Christ-Siemens-Touraine syndrome. Since there was no history of similar features in parents and both the siblings were involved and since they were of different sex, inheritance pattern was autosomal recessive. Parents were counselled about the inherited nature of the disease; however, they refused to undergo any genetic testing. Parents were counselled about maintenance of ambient cool temperature, application of sunscreen lotion, maintenance of hydration and prevention of children from exposure to excessive heat. We considered placement of removable partial denture as the best treatment option for anodontia for both children and referred them to a prosthodontist to receive professional prosthodontic procedures.

DISCUSSION

Ectodermal dysplasia is a rare disorder with defects in two or more of the following structures: the teeth and the skin and its appendages including hair, nails, eccrine, and sebaceous glands [4]. Anhidrotic ectodermal dysplasia (AED) is a rare disorder also known as Christ-Siemens-Touraine syndrome. Charles Darwin was one of the earliest observers to describe this condition. Subsequently, a full description of this disorder was given by Christ, Siemens and Touraine. It is commonly transmitted as an X linked recessive disorder. However, rarely autosomal recessive and autosomal dominant inheritance has also been seen [6]. Hypohidrotic ectodermal dysplasia manifests as a triad of defects, partial or complete absence of sweat glands, anomalous dentition and hypotrichosis. Affected children are unable to sweat, they may experience episodes of high fever in warm environment and may be mistakenly considered to have fever of unknown origin. The typical facies is characterized by frontal bossing, malar hypoplasia, flattened nasal bridge, recessed columella, thick and everted lips, wrinkled hyper pigmented periorbital skin and prominent low set ears. The skin over the entire body is dry, finely wrinkled and hypo pigmented. Anodontia or hypodontia with widely spaced conical teeth are consistent features. Poor development of mucous gland in the respiratory and gastro-intestinal tract may result in increased susceptibility to respiratory infections, purulent rhinitis, dysphonia and diarrhoea.

This disorder is non-progressive and life expectancy is normal or just below average. Mortality in early childhood is due to hyperthermia, failure to thrive and respiratory infections. Maintenance of cool, ambient temperature and psychological support is needed for the patient.

There is no specific treatment for this condition. Genetic counselling of involved families plays an important role.

Consent

The examination of the patient was conducted according to the Declaration of Helsinki principles.

REFERENCES

1. García-Martín P, Hernández-Martín A, Torrelo A. Ectodermal dysplasias: a clinical and molecular review. *Actas Dermosifiliogr*. 2013;104:451-70.
2. Vieira KA, Teixeira MS, Guirado CG, Gavião MB. Prosthodontic treatment of hypohidrotic ectodermal dysplasia with complete anodontia: case report. *Quintessence Int*. 2007;38:75–80.
3. Lesot H, Clauss F, Manie`re MC, Schmittbuhl M. Consequences of X-linked hypohidrotic ectodermal dysplasia for the human jaw bone. *Front Oral Biol*. 2009;13:93–9.
4. Mokhtari S1, Mokhtari S, Lotfi A. Christ-siemens-touraine syndrome: a case report and review of the literature. *Case Rep Dent*. 2012;2012:586418.
5. Mehta U, Brunworth J, Fete TJ, Sindwani R. Head and neck manifestations and quality of life of patients with ectodermal dysplasia. *Otolaryngol Head Neck Surg*. 2007;136:843–7.
6. Zimmla J, Elder ME, Schnieider LC, Orlow SJ, Moss C, Golabi M, et al. A novel X-linked disorder of immune deficiency and hypohidrotic: ectodermal dysplasia is allelic to incontinenti pigmenti and due to mutation in IKK-gamm. *Am J Hum Genet*. 2000;67:1555–62.

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