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HYPOHIDROTIC ECTODERMAL DYSPLASIA REPORTED IN A FEMALE CHILD- A CASE REPORT

Dermatology

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ABSTRACT

Hypohidrotic ectodermal dysplasia is a rare X-linked recessive disorder which presents with the classical features of hypotrichosis, hypohidrosis and hypodontia. Females are usually carriers and here we report a young female child who presented with the characteristic features of this disease.

KEYWORDS

Christ-Siemens-Touraine syndrome, hypohidrosis, hypotrichosis, peg shaped teeth

Introduction:
Hypohidrotic ectodermal dysplasia also known by the names, Anhidrotic ectodermal dysplasia and Christ-Siemens-Touraine syndrome, is the commonest of the ectodermal dysplasias, affecting the skin, hair, nails and teeth. The disease is fully established in males and an affected female carrier will present with decreased sweating, hypodontia or anodontia, sparse hair. We present such a case with associated milia like papules over the nose.

Case report:
A 9 year old female child born of a consanguineous marriage, presented to the skin OPD with complaints of decreased to absent sweating and malformed teeth since birth. She also complained of raised lesions over the nose. She gave a positive history of heat intolerance and recurrent episodes of hyperpyrexia like episodes. Her mother gave a history of delayed eruption of few permanent teeth as well as decreased number and abnormal shape of the erupted teeth. No history of similar features in the family.

On examination from head to toe, the child had sparse, brittle hair on the scalp and eyebrows, high forehead with mild frontal bossing, a flat nose and a pointed chin. There were multiple skin-coloured to pigmented papules over the nose resembling milia. She had extremely dry and warm skin owing to the absent sweating. She was small in stature and thin built. On examination of the oral cavity, her upper two incisors and canines were peg shaped or conical, widely spaced with a lot of missing teeth.

Radiological study of the skull revealed mild frontal bossing and peg shaped incisors.

With the above hair, skin and teeth abnormalities, we came to a diagnosis of hypohidrotic ectodermal dysplasia.

Discussion:
Ectodermal dysplasias are a group of congenital disorders affecting the skin, hair, nails and teeth, all of which are of ectodermal origin1.

Christ-Siemens-Touraine syndrome, a rare X-linked recessively inherited, anhidrotic form of ectodermal dysplasia2 was first described by Thurnam3 and later by Darwin.

Christ-Siemens-Touraine syndrome presents with the classical triad of hypohidrosis (decreased sweating), hypotrichosis (sparse hair) and hypodontia (decreased teeth) 4.

Hypohidrosis results in extreme dryness of the skin, leading on to heat intolerance. Hypohidrosis is due to diminished or absent sweat glands resulting in an inability of the body to thermo regulate. Recurrent hyperthermia5 is a consequence of hypohidrosis, leading on to heat exhaustion and death. The reduced or absent sweating also results in dry scaly skin. At birth, the baby may have marked peeling of skin resembling a collodion membrane6.

The scalp hair and eyebrows are dry, sparse and extremely brittle, while the eyelashes may or may not be affected. Microscopy may show hair shaft defects like pili torti, pili canaliculi or trichorrhexis nodosa. Onychodysplasia is an uncommon nail abnormality5.

Dentition is abnormal and may be the presenting feature with delayed eruption of permanent teeth7. Hypodontia or anodontia are the manifestations of the disease, but peg shaped or conical teeth are the classical and common findings7.

In addition, the patient may also have absent or rudimentary dermatoglyphics8, high forehead, mild frontal bossing, saddle nose, prominent supra-orbital ridges, sunken cheeks, pointed chin, protuberant lips, peri-orbital pigmentation and wrinkling5. Rarely, the patient also presents with milia like papules over the face, cleft lip and palate, eczema, atrophic rhinitis, xerostomia and dry eye due to hypoplasia of the secretory glands5.

A biopsy will show thinned out epidermis with characteristically absent or reduced sweat glands. The sebaceous gland and hair shaft abnormalities are variable.

The diagnosis is mainly clinical but other methods like pilocarpine iontophoresis, sweat pore count, radiological studies to rule out anomalies9 can be carried out. Prenatal diagnosis9 and genetic testing for mutation in the ectodysplasin-A gene10 may also be employed to rule in a diagnosis of hypohidrotic ectodermal dysplasia.

Christ-Siemens-Touraine syndrome must be differentiated from incontinentia pigmenti11 congenital syphilis and other forms of ectodermal dysplasia.

Treatment is usually multidisciplinary and requires a lot of reassurance. Dentition can be corrected using an implant-supported prosthesis once the patient has attained the full craniofacial growth12. Treatment is aimed at managing hypohidrosis and adequate hydration is essential. Patient should avoid exposure to high temperatures and any physical activity that increases the body temperature. Supportive symptomatic management can be administered as and when needed.

Conclusion
Ectodermal dysplasia when identified early has a good prognosis. We have reported this case of Christ-Siemens-Touraine syndrome because of its rarity.

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Conflict of interest: The authors declare that they have no conflict of interest.

Figure 1: High forehead, mild frontal bossing, sparse hair and eyebrows, flat nose and pointed chin.

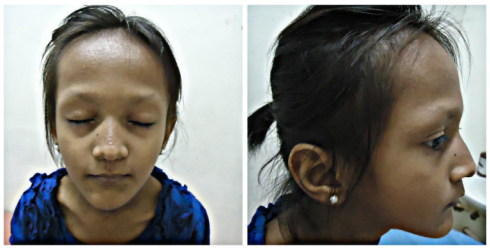


Figure 2: Conical/Peg shaped teeth and milia like lesions over the nose

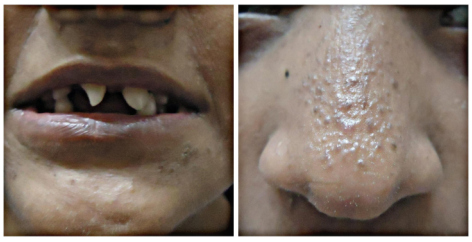
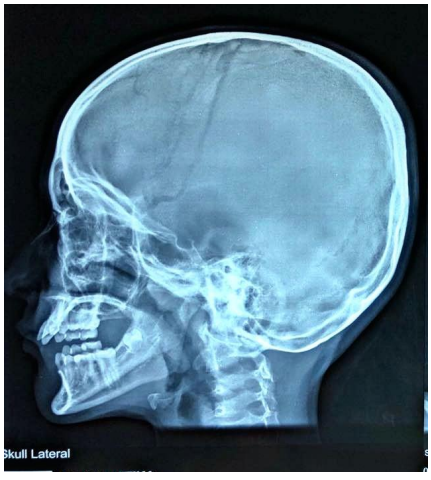


Figure 3: Radiological study of the skull shows mild frontal bossing and peg shaped teeth.



References:

1. Priolo M, Silengo M, Lerone M, Ravazzolo R. Ectodermal dysplasias: not only 'skin'deep. Clinical genetics. 2000 Dec; 58(6):415-30.
2. Zonana J, Elder ME, Schneider LC, Orlow SJ, Moss C, Golabi M, Shapira SK, Farndon PA, Wara DW, Emmal SA, Ferguson BM. A novel X-linked disorder of immune deficiency and hypohidrotic ectodermal dysplasia is allelic to incontinentia pigmenti and due to mutations in IKK-gamma (NEMO). The American Journal of Human Genetics. 2000 Dec 1; 67(6):1555-62.
3. Thurnam J. Two cases in which the skin, hair and teeth were very imperfectly developed. Medico-chirurgical transactions. 1848; 31:71.
4. Pinheiro M, Freire‐Maia N. Christ‐Siemens‐Touraine syndrome—a clinical and genetic analysis of a large Brazilian kindred: I. Affected females. American journal of medical genetics. 1979; 4(2):113-21.
5. Agarwal S, Gupta S. Hypohidrotic ectodermal dysplasia. Indian dermatology online journal. 2012 May; 3(2):125.
6. Shawky RM, Gamal R. Christ-Siemens-Touraine syndrome with cleft palate, absent nipples, gallstones and mild mental retardation in an Egyptian child. Egyptian Journal of Medical Human Genetics. 2016; 17(4):389-95.
7. Clarke A, Phillips DI, Brown R, Harper PS. Clinical aspects of X-linked hypohidrotic ectodermal dysplasia. Archives of disease in childhood. 1987 Oct 1; 62(10):989-96.
8. Wright JT, Grange DK, Richter MK. Hypohidrotic Ectodermal Dysplasia. 2003 Apr 28 [Updated 2013 Jun 13]. Gene ReviewsTM [Internet]. Seattle (WA): University of Washington, Seattle. 2014.
9. Sybert VP. Early diagnosis in the ectodermal dysplasias. Birth defects original article series. 1988; 24(2):277-8.
10. Drögemüller C, Distl O, Leeb T. X-linked anhidrotic ectodermal dysplasia (ED1) in men, mice, and cattle. Genetics Selection Evolution. 2003 Jun; 35(1):S137.
11. Phan TA, Wargon O, Turner AM. Incontinentia pigmenti case series: clinical spectrum of incontinentia pigmenti in 53 female patients and their relatives. Clinical and Experimental Dermatology: Clinical dermatology. 2005 Sep; 30(5):474-80.
12. Singh T, Singh R, Singh GP, pal Singh J. Hypohidrotic ectodermal dysplasia: a felicitous approach to esthetic and prosthetic management. International journal of clinical pediatric dentistry. 2013 May; 6(2):140.