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Hypohidrotic Ectodermal Dysplasia–A Rare Case Report in Siblings

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Abstract: ***Hypohidrotic ectodermal dysplasia is a raregenetic disorder involving primordial germ cells of the hair, sweat glands, teeth and nails. Though the incidence is estimated to be I in one lakh, it is extremely rare in females due to X-linked inheritance. Hypohidrotic ectodermal dysplasia (HED) is a rare genetic disorder characterized by the faulty development of the ectodermal structure, resulting in most notably anhydrosis/hypohydrosis, hypotrichosis and hypodontia.[1] This condition is usually an X-linked recessive disorder affecting predominantly males.[2] Mutations in the gene encoding ligand ectodysplasin A (EDA) underlie classic, X-linked recessive HED, whereas mutations in the genes encoding the EDA receptor and (less frequently) the adaptor protein that associates with the EDA receptor's death domain result in autosomal dominant and autosomal recessive forms of HED. Here is a rare case of hypohidrotic ectodermal dysplasia in a girl and her younger brother.***

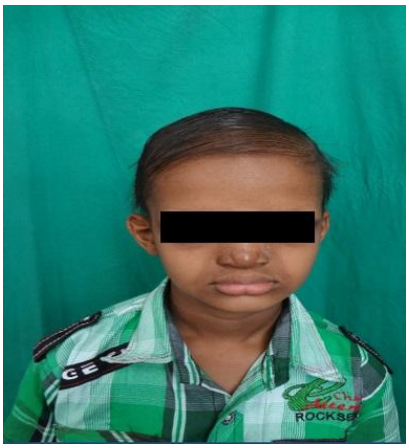
Keywords: ectodermal dysplasia, hypotrichosis, hypodontia, hypotrichosis, case report

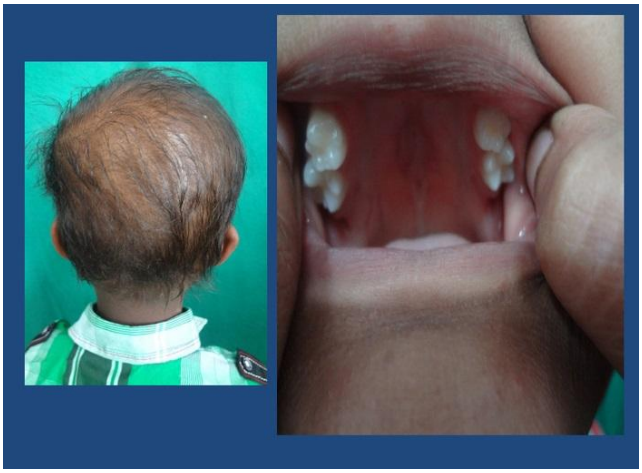
1. Case Report

11 year old girl and a 9 year old boy, siblings born through a second degree consanguineous marriage presented to our dermatology outpatient department with complaints of sparse hair and improper dentition. On clinical examination, the girl was adequately built for age. She had sparse scalp hair and absent body hair. She has ciliary and supraciliarymadarosis, depressed nasal bridge and pointed chin. Skin all over the body appeared dry and finely wrinkled. Nails were normal.

Dental examination revealed partial anodontia and peg teeth.

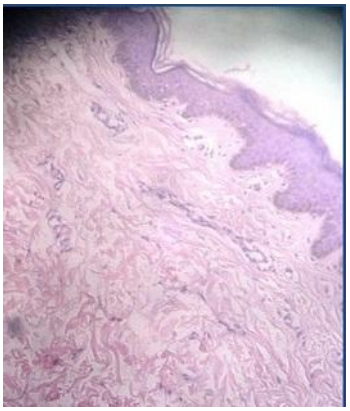
9 year old boy had very sparse brownish scalp hair, almost absent body hair. He too had ciliary and supraciliarymadarosis. Skin was very dry and wrinkled. Nasal bridge appeared depressed and lower lip was everted. Dental examination showed partial anodontia and peg teeth.





Both the children were examined by paediatrician, ophthalmologist and otorhinolaryngologists and found to have no abnormalities. Routine blood investigations were normal. Starch iodine test showed impaired sweating in both the kids. Biopsy was obtained from the arm of the boy and

H/P staining confirmed features of hypohidrotic ectodermal dysplasia.



2. Discussion

Ectodermal dysplasias are a group of inherited disorders that share common developmental defects involving at least two of the major structures classically hold to derive from the embryogenic ectoderms – hair, teeth, nails and sweat glands. HED is characterized by partial or complete absence of sweat glands, hypotrichosis, and hypodontia. The X-linked HED, otherwise called Christ–Siemens–Touraine Syndrome, was first described in 1848 by Thurnam. The incidence at birth is 1 in 100,000 males.[3] The complete syndrome does not occur in females but a female may show dental defects, sparse hair, reduced sweating, and dermatoglyphic abnormalities.

HED is characterized by sparse or absent eccrinesweat glands as well as by hypotrichosis and oligodontia with peg-shaped teeth as seen in the present case also.[4] The conical and pointed teeth are key features of the syndrome and may be the only obvious abnormality. As these patients cannot sweat, patients with HED have a propensity to develop hyperthermia with physical exertion or exposure to a warm environment, and affected infants often present with recurrent high fevers. The scalp hair, eyebrows, and eyelashes are sparse, fine, and oftentimes lightly pigmented. Our patients had loss of eyebrows and eyelashes, and sparse, thin, lightly pigmented scalp hair. In contrast to several other types of ectodermal dysplasia, nails were normal.

HED patients have a characteristic facies with frontal bossing, a saddle nose, and full, everted lips as seen in this case also. Abnormal mucous glands result in extremely thick nasal secretions and a propensity to develop respiratory tract infections.

About 90% of HED are Xlinked recessive which is an abnormality in EDA – ectodysplasin protein that activated NF-kappa B and essential nodulators which have a role in development of ectodermal structures. Autosomal dominant varients are due to DL gene abnormaities affecting EDA protein. Autosomal recessive varients are due to EDARADD protein that interacts with EDA gene.

The management of children and adults with HED can be hard because of their heat intolerance (especially during febrile illness or physical activities and in warm climate) and because of their susceptibility to pulmonary infections. During hot weather, affected individuals must have access to an adequate supply of water and a cool environment, which may mean “cooling vests,” air conditioning, a wet T-shirt, and/or a spray bottle of water. However, external cooling is less effective in these patients because their heat transfer from the core to the skin is also reduced, presumably due to poor capillary dilatation.[7]

Psychological support and counselling plays an important role in development of these children. Oral hygiene and dental prosthesis can be offered to children over the age of 13. Skin has to be kept moist with repeated application of emollients. This condition demands multispeciality management from a team of dermatologists, paediatricians, ENT surgeons, ophthalmologists and dentists.

References

[1] Soloman LH, Kener EJ. The ectodermal dysplasia. Arch Dermatol. 1980;116:295–9.
[2] Reed WB, Lopez DA, Lauding B. Clinical spectrum of anhydrotic ectodermal dysplasia. Arch Dermatol. 1970;102:134–43. [PubMed]
[3] Stevenson AC, Kerr CB. On the distribution of the frequencies of mutation to genes determining harmful traits in man. Mutat Res. 1967;4:339–52. [PubMed]
[4] Cui CY, Schlessinger D. EDA signaling and skin appendage development. Cell Cycle. 2006;5:2477. [PMC free article] [PubMed]
[5] Kere J, Srivastava AK, Montonen O, Zonana J, Thomas N, Ferguson B, et al. X-linked anhidrotic (hypohidrotic) ectodermal dysplasia is caused by mutation in a novel transmembrane protein. Nat Genet. 1996;13:409–16. [PubMed]
[6] Bal E, Baala L, Cluzeau C, El Kerch F, Ouldim K, Hadj-Rabia S, et al. Autosomal dominant anhidrotic ectodermal dysplasias at the EDARADD locus. Hum Mutat. 2007;28:703–9. [PubMed]
[7] Brengelmann GL, Freund PR, Rowell LB, Olerud JE, Kraning KK. Absence of active cutaneous vasodilationassociated with congenital absence of sweat glands in humans. Am J Physiol. 1981;240:H571–5. [PubMed]
[8] 8.Agarwal S, Gupta S. Hypohidrotic ectodermal dysplasia. *Indian Dermatology Online Journal*. 2012;3(2):125-127. doi:10.4103/2229-5178.96711.