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Case Note

Hypohiderotic ectodermal dysplasia

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Abstract
We report a case of a six years age girl who presented to our out patients department with the history and findings suggestive of recurrent respiratory tract infections. She was also noticed to have; non homogeneous hyper-pigmented patches on the face since three months of age, sparse hair on the scalp and eyebrows, conical peg like teeth and delayed dentition, prominent and low set ears, perpetually flexed third toe bilaterally. Axillary skin biopsy showed adenexal structures and eccrine glands in subcutaneous fat which were reduced in number. The diagnosis made was: Anhydrotic/ Hypohidrotic type of ectodermal dysplasia.

Key words: Ectodermal Dysplasia, Anhidreotic, Hypohidrotic,

The ectodermal dysplasias (EDs) comprise a large, heterogeneous group of inherited disorders that share primary defects in the development of two or more tissues derived from embryonic ectoderm. The tissues primarily involved are the skin, hair, nails, eccrine glands, and teeth.

Six year old girl presented to our out patients department with the history of recurrent respiratory tract infections. She was born normally at full term out of a non- consanguineous marriage. Her developmental milestones were normal as per her age but had some physical features, which according to her mother she was noticed to be abnormal as compared to her siblings and peers. The mother also states that her daughter gets recurrent respiratory tract infections both upper and lower and that the temperature during that time goes very high and does not come down, in spite of antipyretics and tepid sponging. There were no such cases known in the family till date.

The abnormalities noticed were non homogeneous hyper-pigmented patches on the face since three months of age, sparse hair on the scalp and eyebrows, conical peg like teeth and delayed dentition, prominent and low set ears, perpetually flexed third toe bilaterally.

On examination: apart from the above abnormalities the rest of the body and the various systems were normal.

Histology: Dermis from the trunk consisted of thick collagen and few hairs without any eccrine glands. Axillary skin showed adenexal structures and eccrine glands in subcutaneous fat which were reduced in number. Diagnosis: Anhydrotic/ Hypohidrotic type of ectodermal dysplasia.

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Fig 1: Teeth and skin abnormality

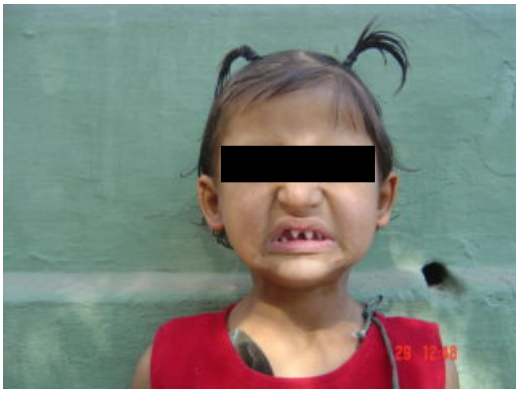
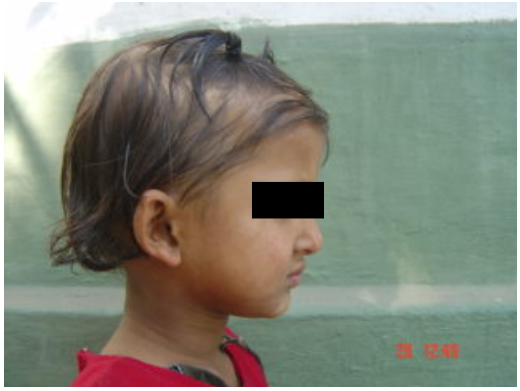


Fig 2: Hair abnormality



Discussion

Hypohiderotic Ectodermal Dysplasia (HED) syndrome is a condition in which there are defects in sweating, alopecia and hypodontia. Thurman first described this entity in 1848. Charles Darwin had also described a similar condition in 1875 in which he mentions about a Hindu Family suffering from a similar condition in which there was decreased number of teeth, early balding and heat intolerance. The same family had no daughters who were suffering from this disease.

It is thus clear now that HED syndrome is an X-linked recessive condition. The gene for it has been localized within the region Xq11-21.1 by linkage analysis. But an autosomal recessive type of HED Syndrome has also been described. The clinical features of this autosomal recessive type have not been distinguishable from the X-linked HED except that females are as severely affected as the males. The case that we have described probably falls in the autosomal recessive type of HED.

There are numerous and varied presentations of HED syndrome, some of which are as follows;

Skin: Hyperpigmentation, thinning and hypoplasia. Dry, fine and sparse to absent hair. Hypoplasia to absent sweat and sebaceous glands.
Mucus membranes: Hypoplasia, absent mucus glands in nasal, oral and bronchial mucosa.
Dental: Hypo to anodontia, conical peg like teeth.
Craniofacial: Depressed nasal bridge, small nose, hypoplastic ala masi, prominent supraorbital ridges.
Others: hoarse voice, hypo to absent mammary glands, absence of tears, failure to develop nasal turbinates, nail dystrophy, eczematous skin, asthmatic symptoms etc.

References

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