

Christ-Seimmens-Touraine Syndrome

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Abstract: Ectodermal dysplasia is a large heterogenous group of inherited disorders, defined by primary defect in development of two or more tissue derived from embryonic ectoderm. The tissues primarily involved are skin, its appendages (hair, eccrine glands, sebaceous gland & nails) and teeth. Ectodermal dysplasias are congenital, diffuse & nonprogressive. The genetic defects responsible for approximately 62 of the ectodermal dysplasias have been identified. The most common are X-linked hypohidrotic ectodermal dysplasia (Christ-Seimmens-Touraine Syndrome) which is caused by mutation of EDA gene which encodes the ectodysplasin protein, a soluble ligand that activates the NF-kappaB and JNK/c-fos/c-jun signaling pathways. Ectodysplasin is important in promoting cell survival, growth, and differentiation. Using specialized techniques, including confocal imaging, phototrichogram analysis, and pilocarpine iontophoresis, a complete absence of eccrine ducts, reduction in hair follicle units & hair follicle density and decreased growth rate of terminal hairs has been demonstrated in patients with XL-HED. In present case we have found classical features of X-linked hypohidrotic Ectodermal dysplasia also known as Christ-Seimmens-Touraine Syndrome.

Keywords: Dysplasia, Ectodermal, Hypohidrotic.

INTRODUCTION:

The ectodermal dysplasias (EDs) comprise a large, heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The tissues primarily involved are the skin, its appendages (hair follicles, eccrine glands, sebaceous glands & nails) and teeth. Thurnam published the first report of a patient with ectodermal dysplasia in 1848; the term ectodermal dysplasia was coined in 1929 by Weech [1].

Prevalence of X-linked hypohidrotic Ectodermal dysplasia is estimated at 1-9 cases per 10,00,000 births and most common in whites & occurs from birth to childhood depending on severity of symptoms [2].

Ectodermal dysplasias are congenital, diffuse & nonprogressive. The genetic defects responsible for approximately 62 of the ectodermal dysplasias have been identified; the most common are X-linked hypohidrotic ectodermal dysplasia [3].

Ectodermal dysplasia are characterized by dry hypo pigmented skin, absent or reduced sweating [4], sparse fair brittle scalp hair with alopecia, absent or diminished body hair and sparse or absent eyebrows & eyelashes due to reduction in hair follicles. Nail dystrophy, conical or pegged teeth, hypodontia or complete anodontia, delayed eruption of permanent

teeth, enamel defects and dental caries are found frequently [5].

CASE REPORT:

A 2 year old male child admitted in our hospital for raised body temperature and decrease sweating. Child requires repeated cold sponging to decrease body temperature since birth.

Child was born to a non-consanguineously married couple of 22 year old female and 25 year old male by full term normal vaginal hospital delivery. Antenatal period was normal with birth weight 2.5 kg. He was admitted at day third of life for high grade fever for 4 days. Child is developmentally normal and fully immunized as per NIP schedule. He has 7 month old female sibling with no similar complaints. His paternal uncle also had short stature, baldness and decrease sweating.

On general physical examination he has dry hypo pigmented skin, coarse hypo pigmented hair, absent eyebrows, prominent forehead with saddle nose, everted lips and pegged teeth. Systemic examination was normal.

On investigations his Hemoglobin was 5.3 gm/dl which is microcytic hypochromic. Serum calcium, electrolytes, vitamin D level, thyroid profile, renal function test and liver function tests were in normal limits. USG abdomen was also normal.

On the basis of history and physical findings diagnosis of Christ-Seimms-Touraine Syndrome (X-linked hypohidrotic Ectodermal dysplasia) was made.

Parents was advised air conditioning for home & school, encourage frequent consumption of cool liquids to maintain adequate hydration and thermoregulation, advised to wear cool & loose clothing. As antipyretics are not effective in the treatment of hyperpyrexia in these cases, parents were advised to do cold sponging whenever body temperature rises.



Fig-1: Sparse hypo pigmented hairs, absent eyebrows, prominent forehead with saddle nose, everted lips, dry hypo pigmented skin

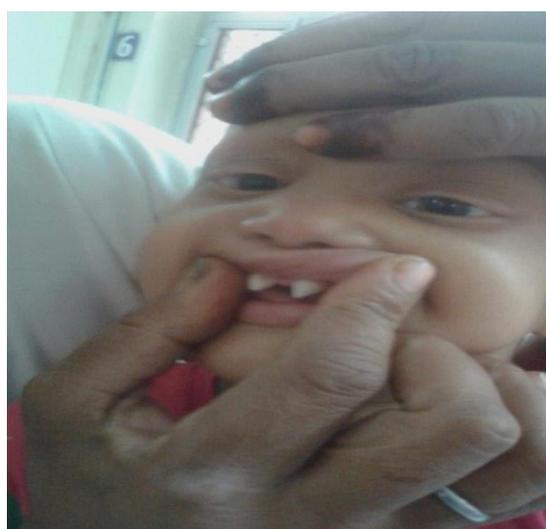


Fig-2: Pegged shaped teeth

DISCUSSION:

The ectodermal dysplasias (EDs) comprise a large, heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The tissues primarily involved are the skin, its appendages (hair follicles, eccrine glands, sebaceous glands & nails) and teeth. Thurnam published the first report of a patient with ectodermal dysplasia in 1848; the term ectodermal dysplasia was coined in 1929 by Weech.

The ectodermal dysplasias are congenital, diffuse & nonprogressive. To date, more than 62 disorders with genetic defects have been described. The most common ectodermal dysplasias are x-linked recessive hypohidrotic ectodermal dysplasia (christ-siemens-touraine syndrome). Key transcription factors and intracellular signaling pathways that have been implicated in the ectodermal dysplasias include the tumor necrosis factor (TNF)-like/TNF receptor signaling pathway, which involves ectodysplasin (EDA), the EDR receptor, the EDAR-associated death domain, the WNT signaling pathway, the NF-KB signally pathway, which involves the NF-KB essential modulator and the transcription factor p63 [6].

X-linked hypohidrotic ectodermal dysplasia (christ-siemens-touraine syndrome) which is caused by mutation of EDA gene which encodes the ectodysplasin protein, a soluble ligand that activates the nf-kappab and jnk/c-fos/c-jun signaling pathways [7]. Ectodysplasin is important in promoting cell survival, growth and differentiation. Using specialized techniques, including confocal imaging, phototrichogram analysis and pilocarpine ineiontophoresis, a complete absence of eccrine ducts, a reduction in hair follicle units & hair follicle density and decreased growth rate of terminal hairs has been demonstrated in patients with X-linked hypohidrotic ectodermal dysplasia [8, 9].

Ectodermal dysplasia are characterized by dry hypo pigmented skin, absent or reduced sweating, sparse fair brittle scalp hair with alopecia, absent or diminished body hair and sparse or absent eyebrows & eyelashes due to reduction in hair follicles. Microscopically medulla is often discontinuous, when medullation is present; a "bar code" appearance is often seen. Nail dystrophy, conical or pegged teeth, hypodontia or complete anodontia, delayed eruption of permanent teeth, enamel defects and dental caries are found frequently. Diminished lacrimation & salivation, dysmorphic facies, cleft lip and palate, frontal bossing, sunken cheeks, saddle nose, thick everted lips, wrinkled hyperpigmented periorbital skin and large low-set ears can be present. Wrinkled hyperpigmented skin around the eyes and everted lips are typical characteristics of anhidrotic/hypohidrotic ectodermal dysplasia syndrome.

Treatment includes air conditioning for home & school, encourage frequent consumption of cool liquids to maintain adequate hydration and thermoregulation, to wear cool & loose clothing. As antipyretics are not effective in the treatment of hyperpyrexia in these cases, advise to do cold sponging whenever body temperature rises.

Topical emollients should be frequently used to prevent dryness of skin. Topical minoxidil with or without a topical tretinoin can be used to improve hair growth [10, 11]. Artificial tears should be used to prevent corneal damage. Dental implants for dental abnormality can be used [12, 13]. No dietary restrictions are indicated. At present no pharmacological treatment is available.

CONCLUSION:

We present this rare case of X-linked hypohidrotic ectodermal dysplasia (christ-siemens-touraine syndrome) with the aim that such cases should be identified and treated at early stage to minimize cosmetic and social stigmata to them and to their families.

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