Hypohydrotic Ectodermal Dysplasia

TM Anoop*, S Simi*, PN Mini+, Manjula Ramachandran**, PK Jabbar***, PK Rajakumari#, P Sujathan##

Abstract
Hypohydrotic ectodermal Dysplasia (Christ - Siemens Touraine syndrome) is a rare genetic disorder that affect several ectodermal structures. The condition is usually inherited as X – linked recessive trait, in which gene is carried by females and manifested in males. The manifestations may vary in individuals and usually involves skin, hair, nail, sweat and sebaceous glands. Hypohydrotic Ectodermal Dysplasia with classical features in two siblings is reported here. ©

INTRODUCTION
Hypohydrotic ectodermal dysplasia is a rare genetic disorder characterized by faulty development of ectodermal structure; resulting most notably anhydrosis/ hypohydrosis, hypotrichosis and hypodontia. This condition is usually an X linked Recessive disorder affecting predominantly male and female are carriers.

CASE REPORT
Two siblings born of consanguineous marriage, aged 16 years and 12 years presented with decreased sweating and repeated history of intolerance to heat since childhood. Patients had history of collodion membrane at the time of birth; with absent scalp hair and eye brows. Younger brother had history of febrile seizures during childhood. The mile stones were delayed.

On examination, Younger sibling has sparse hair, everted lips, megalom pinna, palmo – plantar hyperkeratosis, hypodontia with peg shaped incisors (Fig. 1A). skin was dry. Nail were normal. Systemic examination was normal. The external genitalia and intelligence were normal. Elder sibling, has sparse hair, frontal bossing, everted lips, hypodontia (Fig. 1B) and palmo - plantar hyperkeratosis (Fig. 1C). Skin was dry. Nail were normal. Systemic examination was normal. The external genitalia and intelligence were normal.

The mother, has decreased sweating during heavy work and skin appears dry with out any abnormalities involving teeth, nail, hair. She also has brachydactyly with clefting between big toe and second toe.

Skin biopsy of younger sibling revealed skin with normal appearing epidermis. Dermis showed scattered chronic inflammatory cells. No hair follicles, sebaceous glands or eccrine gland seen (Fig. 1D).

Skin biopsy of elder sibling also revealed normal appearing epidermis and dermis with small collection of mononuclear inflammatory infiltrate. No hair follicles, sebaceous glands or eccrine glands (Fig. 1E).

DISCUSSION
The Ectodermal dysplasias (EDs) are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryogenic ectoderms – hair, teeth, nails, sweat glands.

To date, more than 192 distinct disorders have been described. Freire – Maia and Pinheiro published an exhaustive review and classification system for these disorders using a numeric system of 1 (hair), 2 (teeth), 3 (nail), 4 (sweat glands) for characterization.3

Hypohydrotic ectodermal dysplasia is characterized by partial or complete absence of sweat glands, hypotrichosis and hypodontia. The X - linked hypohydrotic ectodermal dysplasia, otherwise called Christ – Siemens – Touraine Syndrome was first described in 1848 by Thurnam. The incidence at birth is 1 in 100,000 males.4 The complete syndrome does not occur in females but female may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities.

Three genes, Ectodysplasin (EDA1), EDA receptor (EDAR) and EDAR associated death domains (EDARADD) have been described.

Both autosomal recessive and dominant mode of inheritance have been described.

The essential features are absent or reduced sweating, hypotrichosis and total or partial anodontia. The patient
with this disorder have facies suggestive of congenital syphilis. The cheek bone are high and wide, with prominent frontal ridges and chin, saddle nose, sunken cheeks, thick everted lips, large ears and sparse hair. The skin is smooth, soft, dry, wrinkled especially around eyes and appears prematurely aged. The temporary to permanent teeth may be absent or reduced. The conical pointed teeth are key feature of the syndrome and may be the only obvious abnormality. Usually incisors and/or canines are characteristically affected.

The scalp hair is usually sparse, fine and blonde. Alopecia is often the first feature to attract but it is seldom total. The affected males may present at birth with collodion membrane 5. Absent or reduced sweating causes heat intolerance and affected individuals may present with unexplained fever in infancy or childhood. Mental retardation reported in 30 – 50% of cases and is believed to be due to damage from prolonged fever and febrile seizures.

The nails are usually normal or may be brittle. Sexual development is usually normal. The otolaryngology manifestations include thick nasal secretion and impaction, sinusitis, Recurrent respiratory tract infection, pneumonia and increased frequency of asthma.

The characteristic facies is pathognomonic but may not be recognized in infancy. In partial forms, the pointed conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy.

Another variety, hydrotic ectodermal dysplasia (other wise called Clouston syndrome) is inherited in an autosomal dominant manner, was described by Clouston in 1929 and Lowrey et al in 1966, which is found in Canadian families of French descent. Clouston syndrome usually spares the sweat glands and can affect teeth, hair and nail.
Several ED syndrome may manifest in association with mid facial defects. The 3 most commonly recognized entities are:

1) EEC syndrome: Ectodermal dysplasia; Ectrodactyly cleft lip/cleft palate or both.
2) Hay – Wells syndrome (AEC Syndrome) – Ankyloblepharon, ED, Cleft lip/Palate.
3) Rapp – Hodgkin syndrome – Autosomal dominant with high fore head, cleft lip/cleft palate, maxillary hyperplasia, hypohydrosis, hypodontia.

REFERENCES


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**Announcement**

**5th Infectious Disease Certificate Course - IDCC - 2008**

PD Hinduja National Hospital and MRC, Mumbai, India and Henry Ford Health System, Detroit, USA. 24th Aug (Sunday) to 31st Aug (Sunday) 2008. Time : 8.30 am to 4.30 pm

**Format:** Ward Rounds, Archived Cases, Interactive Lectures, Microbiology Discussions, Work Mats, Visit to Infectious Disease Hospital etc.

**Eligibility:** Post graduates in Medicine/Pediatrics/Microbiology (Final year postgraduates may also be considered)

**Registration procedure:** Candidates to send short bio data with Demand Draft/Cheque of Rs. 3,500/- in favor of PD Hinduja National Hospital and Medical Research Centre payable at Mumbai. (Outstation cheques will not be accepted).

**Candidates to make their own arrangement for accommodation**

**Last date for registration:** 30th June, 2008

**Inquiries:** 022-24447204/5, marketing@hindujahospital.com

**Course Coordinations:** Dr FD Dastur/Dr Rajeev Soman/Dr Camilla Rodrigues/Dr Tanu Singhal

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**Announcement**

Office Bearer of API Gwalior Chapter Elections for the year 2008.

Chairman : PC Mathur
Vice Chairman : Niraj Pandeya
Secretary : J Mathur
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Treasurer : V Vaswani
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 : RK Gangil
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