

A Rare Case Report of Anhidrotic Ectodermal Dysplasia in a Newborn

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Abstract: Ectodermal Dysplasias are a heritable group of disorders characterized by faulty development of two or more tissues derived from embryonic ectoderm. This disorder primarily involves skin, hair, nails, teeth and eccrine glands. The disease is usually diagnosed in late infancy and childhood but early diagnosis in a newborn baby tends to have good prognosis. We report a rare case of Anhidrotic Ectodermal Dysplasia in a 26 day old Newborn baby presenting with recurrent episodes of fever, hypotrichosis and anhidrosis.

Keywords: Ectodermal Dysplasia, Anhidrosis, Hypodontia, Hypotrichosis, Hyperthermia

INTRODUCTION

Ectodermal Dysplasias (ED) are a heterogeneous group of hereditary diseases characterized by classic triad of sparse hair (atrachosis or hypotrichosis); abnormal or missing teeth (anodontia or hypodontia); and inability to sweat (anhidrosis or hypohidrosis).

The frequency of Ectodermal Dysplasias is highly variable. In US, the prevalence of Anhidrotic Ectodermal Dysplasia is estimated to be 1 case per 100,000 births. Collectively the prevalence of Ectodermal Dysplasias is 7 cases per 10,000 births [1]. More than 192 distinct disorders have been described so far. Most common of them are X-linked recessive Anhidrotic Dysplasia (Christ –Siemens –Touraine syndrome) [2]. It has full expression in males, although female carriers outnumber affected male but show little (or) no signs [1]. Hidrotic Ectodermal Dysplasia (Clouston syndrome) is autosomal dominant in inheritance and was described by Clouston in 1929 & Lowry et al in 1966. This syndrome spares sweat glands [3]. The etiology of Ectodermal dysplasia is genetic in nature [4].

Ectodermal Dysplasias are classified as either Group A disorders, which are manifested by defects in at least two of four classic ectodermal structures with or without other defects and Group B disorders which are manifested by defect in one classic ectodermal structure in combination with a defect in other ectodermal structures (i.e.; ears, lips, dermatoglyphia). The four classic defects being trichodysplasias, dental abnormalities, onychodysplasias and dyshidrosis [1].

CASE REPORT

A Male baby born of consanguineous marriage, resident of Mahaboobnagar district of Telangana state belonging to tribal community presented at 26 days of life to our neonatal intensive care unit in the month of May during hot summer with complaints of high fever and two episodes of clonic seizures each lasted for 2 -3 minutes during the fever.

On Examination the baby had high fever of 103 °F, tachycardia, tachypnea & clonic movements of all four limbs lasting for 2 to 3 minutes and was treated with empirical antibiotics, tepid sponging, anticonvulsants. Workup for sepsis was done as fever persisted for 3 days. But Haemogram, CRP, Blood Culture, X Ray Chest, Lumbar puncture, CT Brain were within normal limits. On further history mother told she has noticed recurrent episodes of fever from fourth day of life with absence of sweat. On further examination the baby found to have sparse, thin, light brown lusterless scalp hair (Fig. 1). Skin was dry, pale, wrinkled giving an old man appearance with absent sweating and hair on the body (Fig. 2). The eye lashes and eye brows are absent with periorbital wrinkling and hyper pigmentation. Lips are thick, everted with dry oral mucosa. No other anomalies were detected in the baby and a skin biopsy was collected and sent for histopathology (Fig. 3). The skin biopsy showed epidermis with mild focal papillomatosis, acanthosis. The dermis showed a few perivascular lymphocytes and scattered vessels in dermis and absence of skin appendage structures like hair follicles, eccrine glands, sebaceous glands suggestive of Anhidrotic Ectodermal dysplasia.



Fig. 1: Sparse, thin, light brown lusterless scalp hair



Fig. 2: Skin with wrinkling and absent hair

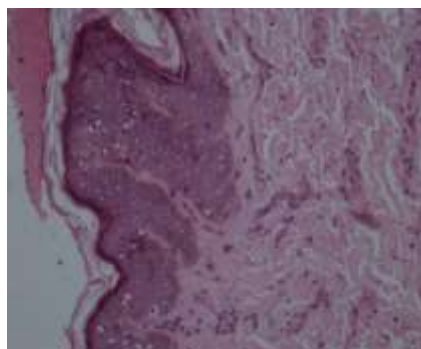


Fig. 3: Skin histopathology showing absent hair follicles, eccrine and sebaceous glands

Mother had no regular antenatal visits during her pregnancies, and she delivered three male children of which first male baby died at 45 days of life and second male baby is of 2 years age who is normal. The third baby is the affected one with Ectodermal dysplasia.

There is a history of consanguineous marriage in family and a history of maternal side uncle being affected with this same disorder.

The baby was discharged after seven days and counseling was given on temperature control methods, use of air-conditioners and preventing from exposure to hot environment, use of emollients, nasal saline drops, artificial tears and regarding the possible tooth abnormalities.

DISCUSSION

The most common type of Ectodermal Dysplasias is X-linked recessive anhidrotic Ectodermal dysplasia. The typical facies is characterized by frontal bossing, sunken cheeks, saddle nose, thick everted lips, wrinkled hyper pigmented periorbital skin & large low set ears [5]. Dental manifestations include conical (or) pegged teeth, hypodontia, or complete anodontia and delayed eruption of permanent teeth. Fine sparse, lusterless hair is seen over scalp along with absent eyebrows, eyelashes and absent sweating and tears which were evident in our case. Onychodystrophy is seen but is not common. Extensive scaling of skin & unexplained pyrexia & heat intolerances is seen, but intelligence is normal [1].

Palmoplantar keratoderma is a component of Hidrotic Ectodermal dysplasia but also reported in anhidrotic Ectodermal dysplasia [6].

The patients may have chronic nasal infections with foul swelling discharge and increased lung infections [7]. Diminished or absent salivary glands & mucous glands of the nose, mouth & ears cause numerous otolarygological complications including nasal obstruction caused by thick, fetid nasal discharge & adherent nasal crusts, sinusitis, recurrent upper respiratory tract infection, feeding problem in infancy, xerostomia, hoarse voice and impacted cerumen. Diminished production of tear film from the lacrimal glands may cause dry eyes, photophobia and corneal damage [8]. Febrile seizures, brain damage and death in early life may result from exposure to hot environment [9].

The age at clinical recognition of Ectodermal Dysplasias varies from birth to childhood. Most of the cases being detected in late infancy and childhood for their presentation with delayed development of tooth, alopecia and nail changes. A family history of similar features and affection of male siblings is helpful to ascertain the mode of inheritance to be x linked recessive [10]. Thurman reported first case of Ectodermal dysplasia involving hair, skin, and teeth in 1848, but Weech named it as Ectodermal dysplasia with absent sweat glands [11]. Freire -Maia and Pinheiro proposed first classification of Ectodermal dysplasia in 1982 [12].with additional updates in 1994 and 2001[13, 14].

There are various classifications proposed for Ectodermal Dysplasias based on clinical presentation, genetic pathways, knowledge of molecular defects, association with or without other syndromes. The recent identification of causative genetic defect lead to a newer classification of Ectodermal dysplasia. In 2003, Lamartine reclassified the Ectodermal Dysplasias in to

four functional groups based on pathophysiologic defect. (a) Cell to cell communication and signaling, (b) Adhesion, (c) Development, and (d) Other [15]. In 2009, sixty four genes and three chromosomal loci are identified to be associated with sixty two Ectodermal Dysplasias [16]. X linked recessive anhidrotic Ectodermal dysplasia is caused by mutations in EDA, which encodes Ectodysplaslin protein, a soluble ligand that activates NF-Kappa B and JNK / C-fos / C-jun signaling pathways [1]. Autosomal dominant and Autosomal recessive Ectodermal Dysplasias are caused by mutations in DL gene which encodes EDA receptor. Hidrotic Ectodermal dysplasia, autosomal dominant disorder caused by mutations in GJBL that encodes connexin 30. The role of alternative EDA receptor localized on X Chromosome (XEDAR) in the control of differentiation of the skin appendages is also studied.

Prenatal diagnosis of Ectodermal dysplasia is done by fetal skin biopsy obtained by fetoscopy at 20 weeks of gestation showing decreased number of eccrine sweat glands. Genetic mutation analysis can also be performed prenatally.

Affected infants with scaling skin may be misdiagnosed as collodion babies with lamellar ichthyosis. Fried tooth and Nail syndrome manifests hypotrichosis, hypodontia & prominent everted lips but the sweating is normal. Basin syndrome is characteristic hypotrichosis, hypodontia, hypohidrosis, but also by severe nail dystrophy & congenital absence of dermatoglyphics.

CONCLUSION

Anhidrotic Ectodermal dysplasia a rare genetic disease has to be included in the differential diagnosis of fever of unknown origin in new born period so that early diagnosis and adequate treatment avoids recurrent hyper pyrexia and its neurological consequences .It also avoids unnecessary use of antibiotics and early counseling regarding temperature control methods gives a better prognosis.

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