Case Report

Severe ocular surface abnormalities in a child and ectodermal dysplasia: a case report

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ABSTRACT

Ectodermal Dysplasia is a disorder that occurs due to abnormal development of at least two major ectodermal derivatives in the developing embryo. Author report the case of a 10 year old male child who was referred to our department with complaints of absent sweating, foreign body sensation and watering in both eyes for past few months. The family history could be traced to four generations and there was an observed trend of increase in severity of signs and symptoms occurring at younger age. The purpose of this case report is to create awareness in the Ophthalmic community about the diagnosis and clinical manifestations of the disorder. This case highlights the role of multidisciplinary approach for management of systemic disease, genetic evolution of affected individual and carriers and genetic counseling.

Keywords: Dry Eye, Ectodermal dysplasia, Meibomian Glands

INTRODUCTION

Ectodermal Dysplasia is a disorder that occurs due to abnormal development of at least two major ectodermal derivatives in the developing embryo.¹ The ectoderm of the embryo forms the skin, hair, nails, sweat glands, teeth and part of the eyes. Hypohidrotic ectodermal dysplasia (HED) is the most common subtype.² We report a case of a child with ocular and dermatological signs of HED along with severe involvement of other multiple organ systems. The family history could be traced to four generations and there was an observed trend of increase in severity of signs and symptoms occurring at younger age. The purpose of this case report is to create awareness in ophthalmic community about the diagnosis and clinical manifestations of the disorder. This case highlights the role of multidisciplinary approach for management of systemic disease, genetic evolution of affected individual and carriers and genetic counseling.

CASE REPORT

A 10 years old male child was referred to the Department of Ophthalmology, DRPGMC Tanda, with complaints of absent sweating, foreign body sensation and watering in both eyes for past few months. On examination, the child had features of dry eyes with conjunctival congestion, lid margins were dry and showed irregular corneal surface with positive fluorescein staining. The Schirmer’s test-1 was 20 mm and 15 mm in 5 minutes in RE and LE respectively, however the TBUT was 3 seconds and 6
seconds only. The eyelashes and eyebrows were absent (Figure 1).

![Figure 1: Absent eyebrows and eyelashes.](image)

Fundus examination was normal. Periorbital hyperpigmentation, dry cracked skin and dry lips, complete loss of nasal hair, malformed and sparse teeth were present (Figure 2).

![Figure 2: Peri-orbital hyperpigmentation, dry cracked skin and dry lips, complete loss of nasal hair, malformed and sparse teeth.](image)

The scalp hair was thin, brittle and sparse. Infrared photography of the everted upper and lower eyelids revealed the absence of meibomian glands (Figure 3). He had ichthyosis and hyper linearity of palms (Figure 4).

![Figure 3: Absence of meibomian glands on Infrared photography.](image)

![Figure 4: Icthyosis and hyper-linearity of palms.](image)

He also had micrognathia and mild scoliosis. The IQ of the patient was normal as per the report of Psychiatry Department. Eye symptoms were managed by prescribing dark glasses and lubricating eye drops to the patient. Lubricating gel was prescribed for the nighttime and hydrocortisone cream was advised for the relief of eczema around eyelids.

The patient was also referred to dermatology and dental department of the college. The patient was educated about the absence of sweat glands and was asked to protect eyes from sunlight.

![Figure 5: The patient with thin, brittle and sparse scalp hair and absent eyebrows/eyelashes.](image)

**DISCUSSION**

HED mainly presents with features of dry eyes, corneal vascularization and pannus. Complications may include
The diagnosis of HED is mainly clinical and based on family history. Carrier testing is possible for the X-linked and autosomal recessive forms if the disease causing mutation is known. Skin biopsy of hypothenar eminence may demonstrate absence or hypoplasia of eccrine sweat glands. Prenatal diagnosis using genetic mutation analysis is possible for pregnancies at increased risk if the disease-causing mutation in the family is known. Infrared imaging may reveal absence of meibomian glands in both the upper and lower eyelids. Ectodermal Dysplasias manifest as defects in ectodermal structures, while ED Syndromes present as combination of ectodermal defects and other systemic anomalies. The most common EDs are X-linked recessive HED (Christ-Siemens-Tourne Syndrome) and Hidrotic ED (Clouston Syndrome). HED is estimated to affect at least 1 in 17000 people worldwide. Mutations in the EDA, EDAR and EDARADD genes cause HED. HED is a heterogeneous group of disorders and so far, more than 192 distinct disorders have been described.

Most cases are caused by mutations in EDA gene, which are inherited in an X-linked recessive pattern. Males are affected by X-linked recessive disorders much more frequently than females as males have only one X chromosome and one altered copy of the gene in each cell is sufficient to cause the condition. In females a mutation must be present in both copies of the gene to cause the disorder1. In X-linked recessive inheritance, females with one altered copy of the gene in each cell is a carrier. Female carriers in HED have mild features as few missing or abnormal teeth, sparse hair and some anomalies with sweat gland function. Female carriers may display a blashkoid distribution of hypohidrosis (the lesions arranged in whorls and streaks corresponding to the lines of Blaschko which represent pathways of epidermal cell migration and proliferation during the development of the fetus) as a result of lyonisation and somatic mosaicism for the abnormal X chromosome.

CONCLUSION

It is important to diagnose patients with Ectodermal dysplasia and treat them for their eye manifestations. Such patients should also be referred to other departments for consultation liaison.

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