

ECTODERMAL DYSPLASIA: REPORT OF A RARE CASE

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ABSTRACT

Ectodermal Dysplasia (ED) is a hereditary congenital disorder of ectodermal origin. It is characterized by lack of sweat glands, (hypohydrosis), Alopecia (Hypotrichosis), defective palms and soles (Palmoplantar hyper keratosis) and oral presentation of partial absence of teeth, (hypodontia) or complete absence of teeth (anodontia). Hypodontia of primary and permanent dentition is one of the most frequently occurring oral symptoms in ectodermal Dysplasia. We report an unusual case of ectodermal dysplasia effecting soles and palms along with oral manifestations.

KEY WORDS: Hypodontia, Anodontia, Hypohydrosis, Hypotrichosis, Palmoplantar Hyperkeratosis

INTRODUCTION

Ectodermal dysplasia (ED) have been described as a group of disorders of morphogenesis displaying two or more of the symptoms of trichodysplasia, dental anomalies, onychodysplasia, and dyshidrosis.¹ It is usually described as being hypohidrotic or hidrotic, depending upon the degree of sweat gland function. Congenital malformation of teeth, hair, nails, or sweat glands may occur either as single isolated malformations or as a part of an ectodermal dysplasia syndrome. These various types of disorders may be inherited in any one of genetic patterns including autosomal dominant, autosomal recessive and X linked. According to an estimate 150 different subtypes of ectodermal dysplasia can be defined with estimated frequency of 1 case occurring in every 10000 to 100000 birth.²

The new cases specify genetic mutation and chromosomal location identified, where group of disorders lead to abnormal development of two or more structures derived from ectodermal layer. The earliest case of ectodermal dysplasia was described in 1792. The ectoderm of one of the three germ layers present in developing embryo gives rise to central nervous system, peripheral nervous system, sweat glands, hair, nails, enamel and teeth³.

Case report

A 13 year old boy visited to us with the chief complaint decayed lower teeth. The subsequent complaint was regarding reduced size of all teeth in particularly incisors. Family history was insignificant. For past four generation there was no such case reported in the family. General examination revealed that he had one digit missing in both the hands (Fig.1).



Fig.1. Missing digit in both the hands

The skin was dry and rough. There was sparse hair on scalp, few hairs in the eye brow and eye lashes. There was slightly depressed saddle nose and everted lips. Linear wrinkles are present around the eyes and mouth. The skin of hands feet often exhibited hyper keratosis. Intraoral

examination revealed that the permanent teeth present were

13,14,16,21,23,26,33,36,37,46,47.

The deciduous teeth present were

55,71,72,74,75,81,82,83,84,85.

Carious Teeth were 85,36,37,46. Mobile Teeth 71,84. All the teeth present were smaller in size and malformed (Fig.2-4). There was mild amount of stains and calculus present, and there was profuse bleeding from the gums during probing. The patient did not have any masticatory problems

Discussion:

This case is unique with involvement of ectodermal structures; as well there is involvement of soles and palms with of symptoms like hypotrichosis, hypohydrosis and hypodontia. The following are the symptoms⁴.

1. Lower than normal number of teeth.
2. Delayed or absent tooth formation.
3. Inability to sweat.
4. Absent tears (Occasionally)
5. Thin skin
6. Decreased skin pigment
7. Foul smelling and nasal discharge
8. Poor temperature regulation
9. Heat intolerance
10. Scanty hair
11. Abnormal Nails
12. Low nasal bridge.

Pathophysiology: Ectodermal dysplasia results from the abnormal morphogenesis of cutaneous or oral embryonal ectoderm, with reduction in the number of hair follicles and with hair shaft abnormalities.⁵

Diagnosis: Orthopantomogram can be done at early age to rule out hypodontia and dental abnormalities⁶. Specific skeletal deformities can be noticed on hand and feet radiography.

Prenatal Diagnosis: The diagnosis has been made on fetal skin biopsies obtained by fetoscopy by 20th week of gestation after determination of sex of the fetus. By histological analysis they demonstrate either complete lack of or reduction in the number of pilosebaceous follicles and lack of sweat gland

primordial in multiple skin biopsies. The interpretation of the biopsies can be difficult if one does not appreciate the normal regional variability distribution of skin appendages of fetal skin. This is complicated and implies considerable risk to pregnancy.⁷ The identification of mutation in family will further improve the accuracy of prenatal diagnosis.⁸

Histopathological examination demonstrates a reduction in the number of sweat glands, hair follicles and sebaceous glands associated with the different EDs. In EDA, the epidermis is thin and flattened. Eccrine sweat glands are few or poorly developed or are very rudimentary. Salivary glands may show ectasia of ducts and inflammatory changes⁹.

Phenotypic Tests: Signs of ectodermal dysplasia are found in about 70 percent of obligate carriers. It is difficult to place much weightage upon subjective assessment of scalp, hair density, heat intolerance, breast feeding difficulties and appearance of eye brows. Two methods of assessment of sweating have been developed to identify possible female carrier. The first sweat test is performed on back of the carrier female and it gives a V shaped strikes that are referred as lines of Blaschko. The other method of assessing sweat pores in female carriers is to make counts of sweat pores along ridges on the finger tips or palms. There are methodological difficulties in performing such test. The gene mapping for X- LINKED ectodermal dysplasia has given possibility of detection of carriers by molecular genetics. Even if DNA diagnosis is feasible it is not available as a service in most centers, and will continue to be expensive and thus unattractive for foreseeable future.¹⁰

Genetic Counselling: Because of the importance of early diagnosis families with x – Linked EDA should be offered genetic counseling. This implies having a calculated risk of having an affected child. For genetic counseling the diagnosis of female carrier is very important. This helps in Optimization in neonatal and pediatric care of affected male infant. Where there is a substantial risk of death infancy, there is substantial mortality and morbidity in male infants with about 30 per cent in first 2 years of life because of fever or chest infection.¹¹

Complications: Febrile seizures, brain damage caused by increased body temperature.



Fig.1.



Fig.2.



Fig.3

Fig.1-3. Intraoral features

Treatment: The initial treatment includes oral prophylaxis and amalgam restoration. The patient was advised oral prophylaxis against caries. He had no masticatory problems. So he was reluctant for other dental treatments.

CONCLUSION

Ectodermal dysplasia is a genetically inherited disorder which affects the ectodermal structures, such as hair, nail, skin, sweat glands and dental anomalies such as hypodontia or microdontia. Dental Practitioners should consider ED as a differential diagnosis in patients having dental anomalies. Various diagnostic tests mentioned above can aid in the confirmation of the diagnosis. Although management of these patients may be challenging, the importance of basic prevention should be stressed.

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