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CASE STUDY

ECTODERMAL DYSPLASIA: A CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT

Ectodermal dysplasias is a rare hereditary disorder that results due to disturbances in the ectoderm of the developing embryo. The classic features that define this condition are partial or complete tooth agenesis which is a congenital lack of one or more teeth of primary and or permanent teeth, sparse hair, dystrophies of nails and lack of sweat glands. Since the pediatric dentists are the first to be consulted in case of loss of teeth, this article is a sincere effort to spread awareness about this rare entity. A case of eleven year old girl child is reported hereby who presented with a complaint of missing teeth, dry skin and brittle nails.

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INTRODUCTION

Ectodermal dysplasias is an umbrella term comprising of large, heterogeneous group of genetically inherited disorders that are characterised by defects in two or more ectodermal derivatives (Itin and Fistarol, 2004). Teeth, nails, hair, skin and eccrine glands are the most frequently affected tissues. Ectodermal dysplasias is a rare disease with a frequency ranging from 1/10,000 to 1/1, 00,000 live births (Mortier and Wackens, 2004; Johnson *et al*., 2002). In medical literature, it was first reported by Thurman in 1848 (Thurnam, 1848). Weech in 1929 coined the term Ectodermal dysplasia (Weech, 1929).

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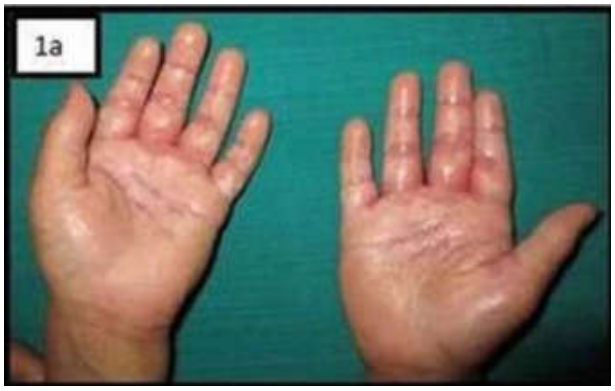
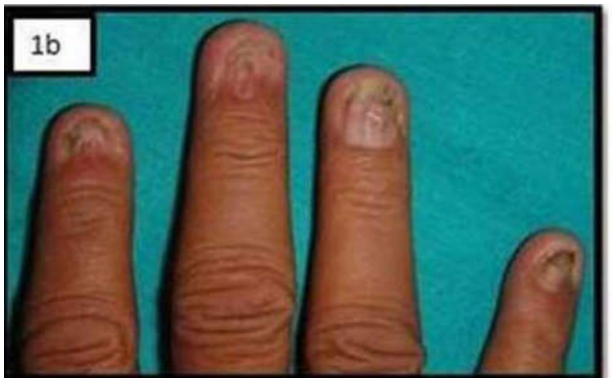
The characteristic features are hypodontia or anodontia(partial or complete absence of teeth); hypotrichosis (sparseness of scalp and body hair); Onchondysplasia (abnormal nails) and Hypohidrosis (reduced ability to sweat) (Itin and Fistarol, 2004; Mortier and Wackens, 2004).

CASE PRESENTATION

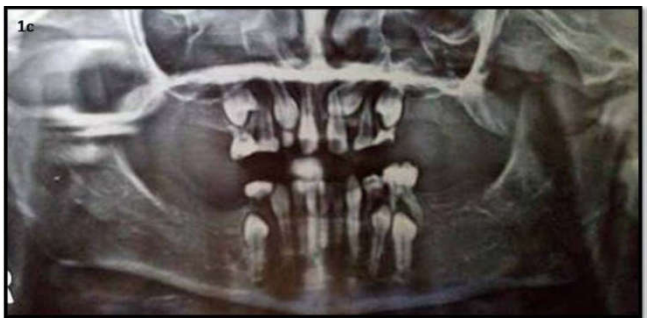
An eleven-year-old female patient reported to the outpatient department with a chief complaint of missing teeth since early childhood. Her parents gave history of delayed eruption of primary teeth and thin, weak nails in the patient. She was born at full term by normal delivery to healthy mother and father aged 27 and 30 years respectively. No history of consanguineous marriage of parents was present. No history of trauma or major illness was reported. No other person in the family had similar problems.



1a. Thick callous patches with extensive scaling over the palms of both the hands



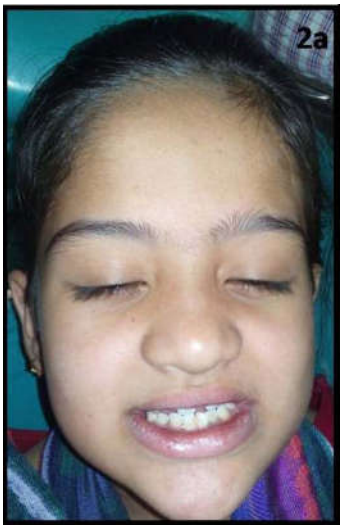
1b. yellowish-brown discoloration of nails with pitting and thinning of the nail plate along with longitudinal grooves and ridges



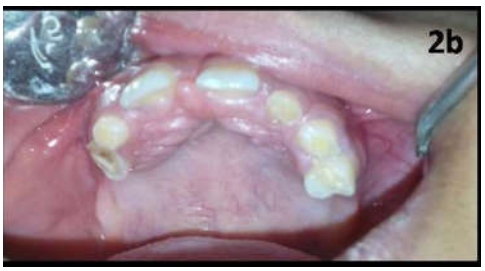
1c: Panoramic radiograph of 11 year old patient showing primary dentition and developing tooth buds of maxillary canine and first premolars on both sides; mandibular left first and second premolar and mandibular right first premolar tooth

FIG. 1. A young female presented with complaint of missing teeth, dry skin and brittle nails

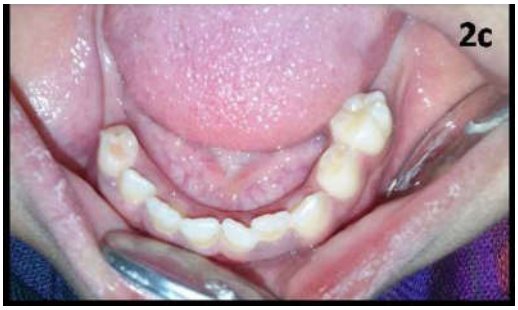
On general physical examination, she was well-oriented with time, place and things. Skin was dry, wrinkled and revealed callous patches with extensive scaling over the palms of both the hands. (Fig1a) Examination of nails revealed generalised yellowish-brown discoloration with pitting and thinning of the nail plate along with longitudinal grooves and ridges. (Fig 1b) Patient had a high hairline with broad forehead. Prominent supraorbital ridges, depressed nasal bridge and broad columella were noticed (Fig.2a). Intraoral examination revealed retained primary teeth in the oral cavity.



2a. Facial profile showing prominent supraorbital ridges, depressed nasal bridge, broad columella, high hair line and dry skin



2b. Maxillary arch with multiple missing teeth



2c. Mandibular arch with multiple missing teeth

(Fig 2b, 2c) No permanent teeth had erupted in the oral cavity as expected according to the chronological age of the patient. Further the patient was subjected to orthopantomography which demonstrated primary dentition with developing tooth buds in relation to right maxillary first premolar, canine, left maxillary canine and first premolar and mandibular left first and second premolar, right mandibular first premolar.(Fig.1c). Thus on the basis of oligodontia, palmo-plantar hyperkeratosis, nail dystrophies, patient was diagnosed with ectodermal dysplasia.

Investigations

Orthopantomography

which demonstrated primary dentition with developing tooth buds in relation to right maxillary first premolar, canine, left

DIFFERENTIAL DIAGNOSIS

Table 1. List of differential diagnosis

Conditions	Favouring Points	Contrasting Points
Papillon Lefevre Syndrome	Palmoplantar keratosis	Early onset severe periodontitis
Goltz syndrome	Multiple abnormalities of mesodermal and ectodermal tissue.	Reddish tan, atrophic linear cribiform patches. Ocular and skeletal defects
EEC syndrome	Ectodermal defect	Ectrodactyly, mild hypohidrosis and cleft lip/palate
Hay-Well syndrome/AEC	Ectodermal defect	Ankyloblepharon, cleft lip/palate, Hypospadias,Syndactyly,Short stature & Absent lacrimal puncta
Oro-facial-digital syndrome	Broadened nasal bridge	Facial asymmetry, Hypertelorism, Micrognathia & Pseudoclefting of upper lip

maxillary canine and first premolar and mandibular left first and second premolar, right mandibular first premolar. (Fig.2)

Treatment

- Psychological counselling of the parents and the patient was done to make them understand the nature of disease
- Patient was advised to avoid exposure to warm weather by staying indoors in air-conditioned environment and wear clothes made of cool fabric like cotton and linen.

Patient was kept under 6month regular follow-up to assess the growth of dentition.

DISCUSSION

National foundation of ectodermal dysplasia, Illinois, defines it as a group of genetic disorders in which there are congenital birth defects of two or more ectodermal structures. The ectodermal dysplasia is congenital, diffuse and non-progressive (Kramer *et al*., 2007). Till date about 160 clinically and genetically distinct hereditary ectodermal dysplasias have been reported. In 1994, Pinheiro and Freire-Maia categorised ectodermal dysplasia into two groups. Group A is characterised by defects in at least two of the four classic ectodermal structures with or without other defects.

While group B is characterised by defect in one classic ectodermal structure in combination with a defect in one other ectodermal structure (i.e., ears, lips, dermatoglyphics) (Pinheiro *et al*., 1994; Freire-Maia *et al*., 2001). Thus our case falls under group A. Or ectodermal dysplasia can be classified on the basis of inheritance. The subdivisions are X-linked hypohidrotic ectodermal dysplasia (the most common form), autosomal-recessive hypohydrotic Christ-Siemens-Touraine syndrome and autosomal-dominant hidrotic Clouston syndrome (Priolo and Lagana, 2001; Cobourne, 2007). The etiopathogenesis suggests it to be due to genetic defects in ectodysplasin signal transduction pathways. Since the epithelial cells in developing tooth, hair follicle and sweat gland utilize this pathway during morphogenesis, any genetic disturbance of the pathway leads to hypoplasia, aplasia or dysplasia of the associated structures (Cobourne, 2007). The most common complaint for which patient seeks medical care is either the absence of teeth or their atypical shape as seen in our case. Characteristic dental findings are conical shaped teeth, microdontia, spacing between teeth and partial or complete loss of primary and or permanent dentition. The lack of teeth leads to dietary difficulties in the patients and add up to poor nourishment, irritability, early fatigue and low self-esteem (Nordgarden, 2001). Patient suffering from X linked ectodermal dysplasia have characteristic "old man facies" because of dry, wrinkled, scalded skin. The texture of skin is comparatively thinner as per the age. Other features include frontal bossing, prominent supra orbital ridge and broad square shaped forehead. Depressed nasal bridge thereby appearing as saddle nose is also present. About 45-50% population presents with dystrophies of the nails. Brown discoloration of nails with slow growth and split nails are commonly reported. In some cases, longitudinal ridging, thinning and superficial peeling can also be noticed. Nails of the fingers are more frequently involved than toenails. All the above features were present in our case (Cobourne, 2007; Nordgarden *et al*., 2001; Bani *et al*., 2010). Abnormalities of hair are present in few affected individuals as sparse, fine-textured, slowly growing and thin scalp hair. Complete baldness has been reported in some cases by mid-teenage.

Thinning of eyebrows and eyelashes is most routinely seen. Decreased body hair, pubic hair, and/or axillary hair may also be present. However, beard and moustache hair are almost always normal. Heat intolerance is another major concern (Nordgarden *et al*., 2001; Bani *et al*., 2010). Due to the reduced number of sweat glands, the body temperature is not maintained which leads to hyperthermia that can be fatal in infants. In adults, hyperthermia may lead to recurrent episodes of high fever, severe respiratory infections and brain damage, and is probably the cause of the rare cases of ectodermal dysplasia reported with mental retardation (Nordgarden *et al*., 2001; Ranjan, 2013). Hair and sweat glands were normal in our case. Thus ectodermal dysplasia can be diagnosed on the basis of hypodontia, Misshapen teeth (i.e conical shaped, knife-edge, taurodontism) of teeth, characteristic facies, skin and nail dystrophies, hair abnormalities and heat intolerance. The list of conditions which present with similar signs and symptoms is summarised in Table 1 (Nordgarden *et al*., 2001; Bani *et al*., 2010; Ranjan, 2013; Patel *et al*., 2014; Vieira *et al*., 2007; Sinha *et al*., 2014). The key to effective management lies in the initial diagnosis of the disease. Thus families in whom ectodermal dysplasia is present should mandatorily go for genetic counselling. This would be helpful in assessing the risk of having an affected child (Bani *et al*., 2010). Thus, optimisation of neonatal and paediatric care of the same can be ensured. Dental radiographs can provide useful additional information and can be a simple screening test for the carrier

status. There are two methods commonly employed to assess the sweating (Bani *et al*., 2010; Ranjan, 2013). One is to count sweat pores along ridges on the fingertips or palms. In another sweat test is performed on the backs of the carrier female which gives a characteristic V-shaped pattern of streaks that refers to the lines of Blaschk (Bani *et al*., 2010; Patel *et al*., 2014). Managing such patients is a tedious and life-long task. The aim of the treatment is to restore the function and the aesthetics of the teeth and thus aid in psycho-social rehabilitation of the patient. Early placement of partial or full dentures is commonly recommended from the age of two or three years onwards. The denture must be periodically modified as the physical development occurs (Vieira *et al*., 2007). Counselling should be done repetitively at every visit. Wigs or customized hair care treatment to manage sparse hair. During hot weather, patient should restrict exposure to outside and must have access to cool environment like coolers, air-conditioners or by wearing wet T-shirt (Sinha *et al*., 2014).

Learning Points

- Impairment caused by ectodermal dysplasia is not limited to structural or functional disturbances but it hampers the psychological and social development of the young growing children.
- Apart from definite dental care, psychological counselling forms the mainstay of the treatment.
- Thus dental and medical fraternity should club together to provide best possible treatment to these patients.

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