Journal of Advanced Medical and Dental Sciences Research

@Society of Scientific Research and Studies

Journal home page: www.jamdsr.com doi: 10.21276/jamdsr

(e) ISSN Online: 2321-9599; (p) ISSN Print: 2348-6805

Case Report

Hereditary Ectodermal Dysplasia: Report of 3 Patients from a Family

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***ABSTRACT*:**
Ectodermal dysplasia (ED) is a group of rare, inherited disorders characterized by sparse hair, missing teeth and inability to sweat. Ectodermal dysplasia (ED) is characterized by dysplasia of ectodermal tissues and occasionally of mesodermal tissues of the developing embryo. This article represents three case reports of ED seen in family members.
Key words: Ectodermal dysplasia, hairs, nails, teeth.

Received: 18 February 2018 Revised: 28 February 2018 Accepted: 8 March 2018

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This article may be cited as: Bhasin M, Sreedevi, Kakkad A, Bhasin V. Hereditary Ectodermal Dysplasia: Report of 3 Patients from a Family. J Adv Med Dent Scie Res 2018;6(4):4-8.

INTRODUCTION

Ectodermal dysplasia as defined by Freire-Maia[1] are "congenital disorders characterized by alterations in two or more ectodermal structures, at least involving one in hair, teeth, nails, or sweat glands". It is essential to accentuate that an ED should have a genetic cause and that the four cited classical structures are affected in the following decreasing order of frequency: hair, teeth, nails, and sweat glands, associated or not with alterations in other ectodermal appendages.

According to Freire-Maia's classification, EDs are divided into two groups: group A which consists of all the entities with defects in two or more of the standard structures and group B comprises of those with disturbances in only one of these structures plus another ectodermal defect.

ED is thought to occur in approximately 1 of 1,00,000 live births with a mortality rate of 28% in males up to 3 years of age. They represent a large and complex group of diseases comprising more than 170 different clinical conditions.[2] There are two major types of ED depending on the number and functionality of the sweat glands; Hypohydrotic and Hydrotic. Hypohydrotic type, also called as anhydrotic type, is the most common ED (80%) and is often inherited as an X-linked disorder (XLEDA). It is characterized by the classical triad of hypodontia, hypohidrosis and hypotrichosis with characteristic dysmorphic facial features.[10] In this type, the sweat glands are either absent or significantly reduced in number. It is also termed as Christ-Siemens-Tauraine syndrome.[3] This article highlights case report of ED seen in three family members.

CASE REPORT 1

A 9 years old male patient reported to Oral Medicine & Radiology Department with a chief complaint of absence of lower front teeth and pointed upper front teeth since when they erupted (Figure:- 1). History of presenting illness revealed that patient's parents reported that milk teeth erupted late and were not in proper shape. Patient had less sweating and intolerance to heat during summer and complaints of smooth peeled skin and excessive thirst during summer and keeps water bottle along with him every time. There was no history of trauma or extraction of the teeth. Patients never visited any medical or dental practitioner for the correction of same. Family history revealed that patient's sister and father had the similar complaint of malformed and missing teeth.

Extra- oral examination showed presence of less than normal distribution of hair and hair was thin, sparse (Hypotrichosis) all over the body. Forehead showed fine periocular wrinkling with sparse eyelash and eye brow hairs (Figure:- 2). Intra- oral examination revealed that 11, 12, 53, 54, 21, 22, 63, 64, 31, 32, 73, 74, 41, 42, 83, 84 were missing. Cone shaped or peg shaped and over- retained 51 present. Palate was V shaped (Figure:- 3).

On basis of above history & clinical examination, provisional Diagnosis of hereditary ectodermal dysplasia was put forth. Digital IOPA of maxillary anterior region revealed presence of 11, 21 and 51. Physiological rootresorption of 51 was evident. 11 and 21 were screw driver shaped, with incomplete root formation and blunder buss canals (Figure:- 4). Panoramic radiograph revealed presence of maxilla and mandible with partial anodontia and malformed teeth. 12, 53, 54, 22, 63, 64, 31, 32, 73, 74, 41, 42, 83, 84 were missing. Permanent tooth buds of 13, 23, 31, 32, 33, 41, 42, 43, 14, 15, 24, 27, 37, 34, 44and 47 were also missing. Distal root of 75 and 85 showed lamina dura in furcation area. 36 revealed incomplete root formation (Figure:- 5). Lateral Cephalogram revealed presence of deficient maxilla, and mandible with missing teeth. Soft tissue shadow of lips showed everted thick lips. Partial anodontia was also evident (Figure:- 6). On basis of above history, clinical examination & radiological examination, a final diagnosis of hypohidrotic ectodermal dysplasia was put forth.

CASE REPORT 2

A 12 year old female patient reported with missing lower front teeth. History revealed that patients had missing lower front teeth since childhood. Patient father denied any past history of extraction of teeth or trauma. Past medical and dental history was non- contributory. Extra- oral examination showed presence of less than normal distribution of hair which was thin, sparse all over the body (Figure:- 7). Intra- oral examination showed midline diastema w.r.t upper anterior region. There was missing 12 22 31 32 41 42, retained 553 and 63. Labial frenum was high and thick (Figure:- 8). Considering the history and clinical examination a diagnosis of hereditary ectodermal dysplasia was given.

CASE REPORT 3

52 years father of above patients reported with missing all lower teeth and upper front teeth (Figure:- 9). History of presenting illness revealed that patient had missing front teeth since childhood and few lower teeth which got lost itself. Patient had visited the dentist in the past which advised him artificial prostheses for the same. Past medical history was non contributory. Family history revealed that patient's son and daughter had similar complaint of missing teeth since childhood. Extra- oral examination showed presence of thin and sparse hair all over the body. Scalp showed presence of few, thin hairs (Figure:- 10). Considering history, clinical examination and positive family history, a final diagnosis of hereditary ectodermal dysplasia was given.

All the patients underwent extraction of retained deciduous teeth followed by artificial prosthesis. Patient was recalled routinely for follow up.

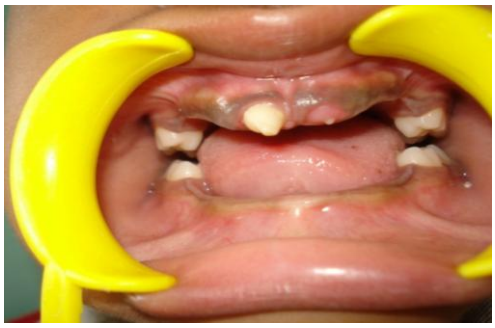

Figure:- 1 Absence of lower & upper front teeth

Figure:- 2 Forehead showed fine periocular wrinkling with sparse eyelash and eye brow hairs

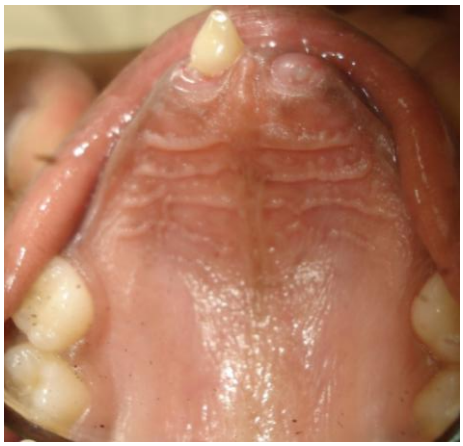

Figure:- 3 Cone or peg shaped overetained 51 present, V shaped palate

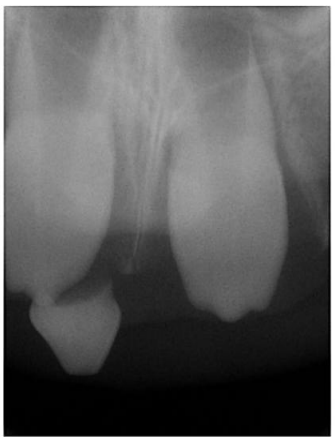

Figure:- 4 Digital IOPA of maxillary anterior region reveals presence of 11, 21, 51

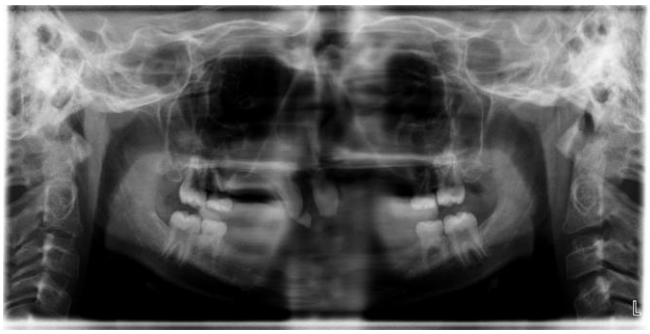

Figure:- 5 Panoramic radiograph revealed presence of maxilla and mandible with partial anodontia and malformed teeth

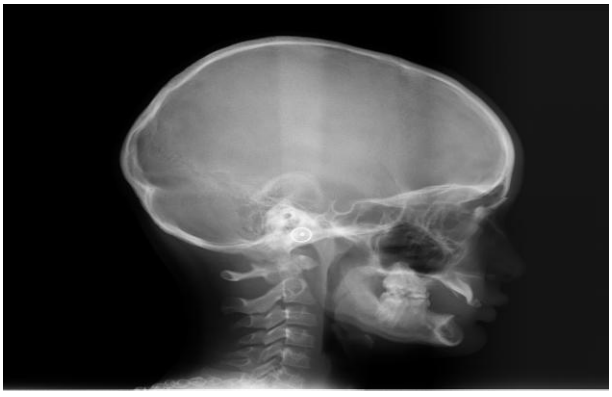

Figure:- 6 Lateral Cephalogram reveals presence of deficient maxilla, and mandible with missing teeth

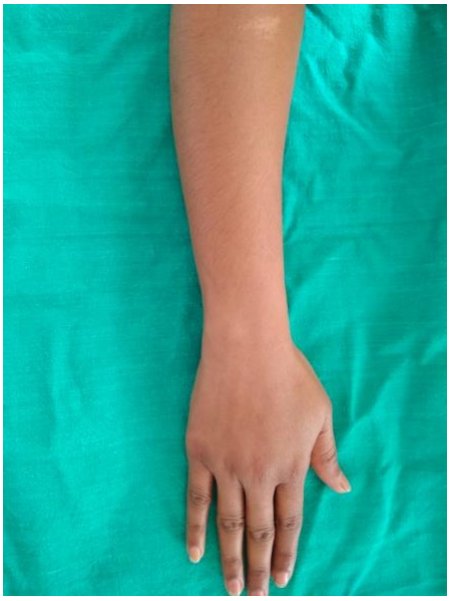

Figure:- 7 Presence of few, thin and sparse hairs

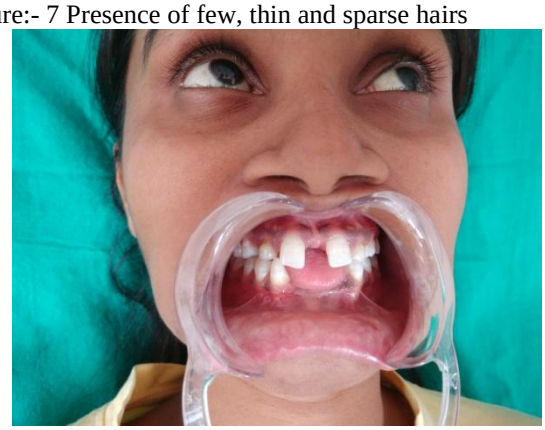

Figure:- 8 Missing mandibular anterior teeth with midline diastema

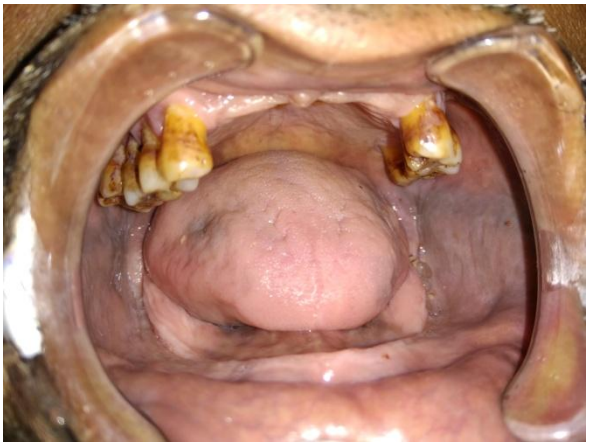

Figure:- 9 Missing upper and lower teeth

Figure:- 10 Presence of thin and sparse scalp hairs

DISCUSSION

In the majority of cases of hereditary hypohidrotic (anhidrotic) ectodermal dysplasia it is an X- linked recessive mendelian character. However in some forms the abnormality can also be transmitted as an autosomal dominant or recessive characteristic. It is the most common type of various ectodermal dysplasia which, as a group, may present abnormalities of the skin, hair, nails, eyes, teeth, facies, sensorineurinal apparatus & adrenal glandular structures in various combinations & of varying severity.[4] Ectodermal dysplasia syndrome results from aberrant development of ectodermal derivatives in early embryonic life. Genes responsible for the varied syndromes are located on different chromosomes and may be mutated or deleted. The disease may also be inherited by autosomal dominant, autosomal-recessive or X-linked genetic transmission. X-linked genetic transmission is the most common among these.[5]

X-linked hypohidrotic ED has been mapped in the proximal area of the long arm of band Xq-12-q13.1.[6] Decreased expression of the epidermal growth factor receptor has been proposed as playing a causal role in this condition's phenotype. The gene ED1 responsible for the disorder has been identified. Males are affected much more frequently then females. Soft, smooth, thin, dry skin, partial or complete absence of sweat glands, such persons cannot perspire and patient suffer from hyperpyrexia and inability to endure warm temperature. In our 3 cases, 2 were males.

The sebaceous glands & hair follicles are often defective or absent. The hair of the scalp & eyebrows tend to be fine, scanty & blond. Symptoms of a reduction in hair follicles vary from sparse scalp hair to a complete absence of hair. Hair bulbs may be distorted, bifid, and small. This is similar as reported in our cases. Moustache & beard are usually normal in appearance.[7]

The bridge of nose is depressed. As seen in our first cases. Supraorbital ridges & frontal bosses are pronounced. Lips are protuberant. Facial appearance of these individuals is quite characteristic & that they resemble each other enough to be mistaken for siblings. The mouth may be dry from hypoplasia of the salivary glands, lacrimal glands also may be deficient.Teeth show abnormal morphogenesis or are absent. Nails are often brittle and thin or show abnormal ridging but they may be grossly deformed.[8] Differential diagnosis includes sporadic oligodontia, radiation therapy in childhood, chondroectodermal dysplasia, Down's syndrome, Ehlers Danlos syndrome and Focal dermal hypoplasia syndrome (Goltz syndrome).[9]

Koszewski & hubbard[10] reported a 25% incidence of refractory chronic anemia in patients with hereditary ectodermal dysplasia. Other Signs and Symptoms are lack of breast development, deficient hearing or vision, cleft lip and/or palate and missing fingers or toes are also seen.

Chandramani et al[11] observed that ED was more prevalent in males, with a ratio of 1.7:1. The hypohydrotic type was more common (78.95%) than hydrotic type (21.05%). The marriage history of parents revealed that 66.67% had consanguineous marriage and had 68.42% offspring's affected with ED; whereas 33.33% had history of non-consanguineous marriage and had 31.58% offspring's affected with ED. The clinical manifestations observed were- dry skin(94.74%); scaly skin(42.11%); sparse hair on scalp, eyebrows and eyelashes(100%); frontal bossing(63.18%); saddle nose (57.89%); hypertelorism (47.37%); nail abnormality(52.63%); normal sweat glands(21.05%); abnormal sweat glands(78.95%); hypoplastic maxilla(52.63%); protuberant lips (57.89%); palmo-plantar keratosis(21.05%); wrinkled & hyper pigmented facial skin(84.21%); partial anodontia(94.74%).

Management of hypohidrotic ectodermal requires genetic counseling for the parents and the patient. The dental problems are best managed by prosthetic replacement of the dentition with complete overdentures, or fixed appliances, depending on the number and location of the remaining teeth.[12] Malformed can be built up with composite or prosthetic crown.

CONCLUSION

ED is a rare inherited disorder affecting hairs, skin, salivary glands and nails. ED patients undergo severe social problems and suffer from poor psychological and physiological development as a result of unacceptable esthetics and abnormal function of orofacial structures. Oral rehabilitation thus becomes mandatory, although it is often difficult; particularly in pediatric patients.

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Source of support: Nil **Conflict of interest:** None declared