Original Article

Hereditary ectodermal dysplasia: A retrospective study

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Abstract

Background: Ectodermal dysplasia (ED) is a group of rare, inherited disorders characterized by sparse hair, missing teeth and inability to sweat. **Objective:** To review and analyze cases of ED with an emphasis on clinical manifestations and parent's marriage history. **Methodology:** The present retrospective study was conducted by assessing the clinical records of nineteen cases of ED, available in the archives of the department; for age, gender, family history of consanguineous marriage and clinical manifestations. **Results:** It was 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%); conical shaped teeth(84.21%); high arched palate(68.42%); thin alveolar bone(100.00%); taurodontism(21.05%) and cleft lip & cleft palate(05.26%). The number of teeth present in all the cases ranged from 0 to 19. **Conclusion:** ED patients suffer from social problems and 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.

Key words: Anodontia, consanguineous marriage, ectodermal dysplasia,hydrotic, hypohydrotic, hypotrichosis, taurodontism

INTRODUCTION

Ectodermal dysplasia (ED) is a term used to describe a heterogeneous group of rare, inherited disorders mainly characterized by dysplasia of ectodermal tissues and occasionally of mesodermal tissues of the developing embryo.[1-3] ED is characterized by the triad of signs comprising of sparse hair (atrichosis or hypotrichosis) [Figure 1], abnormal or missing teeth (anodontia or hypodontia) [Figure 2], and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis).[3,4] In addition, nail dystrophy and palmoplantar hyperkeratosis is usually present.[5,6] The sensorineural and adrenal tissues are also affected at various degrees.[1]

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ED was first reported in 1792 by Danz.[7] In 1838, Wedderburn documented ED in a letter to Charles Darwin; describing 10 cases of Hindu male family members.[8] Nicolle and Hallipre in 1895 first described hydrotic ED in a French-Canadian family.[9] In 1913, Christ characterized hypohydrotic ectodermal dysplasia as a congenital ectodermal defect.[7] In 1921, Siemens described the X-linked nature of inheritance. The term ED was coined in 1929 by Weech.[3] In 1936, Touraine described the wide range of features in ED. Thus, hypohydrotic ED is also known as Christ-Siemens-Touraine syndrome.[10] Clouston in 1939 used the term anhydrotic ED.[11] Felsher in 1944 changed

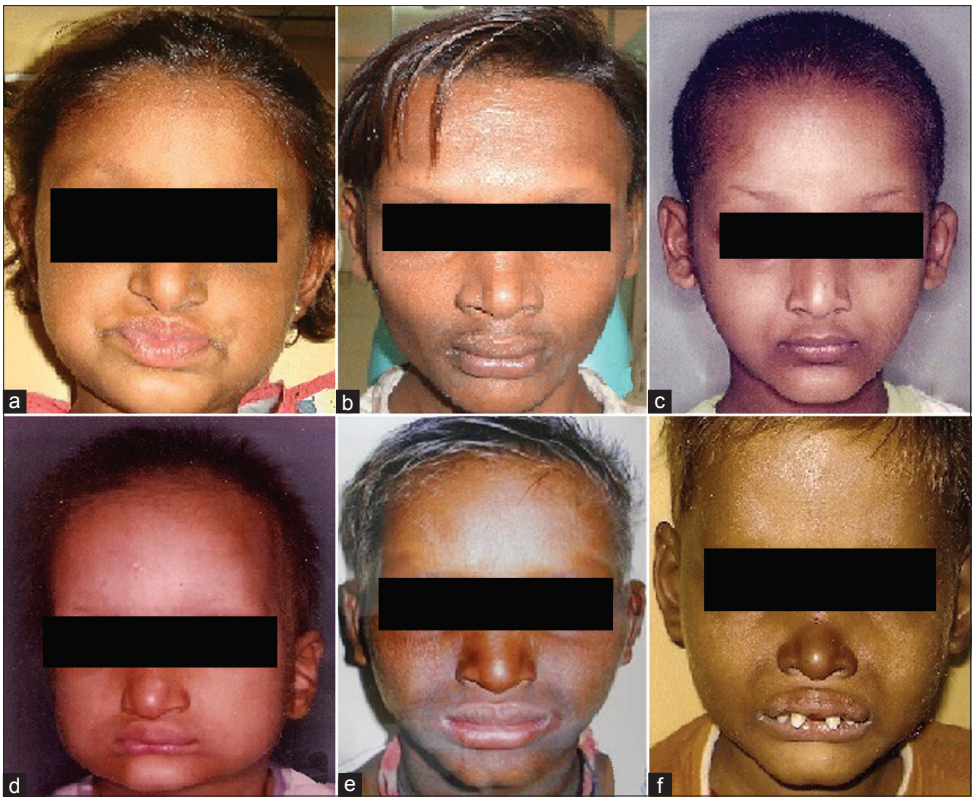


Figure 1: Extra oral photographs of patients with ectodermal dysplasia

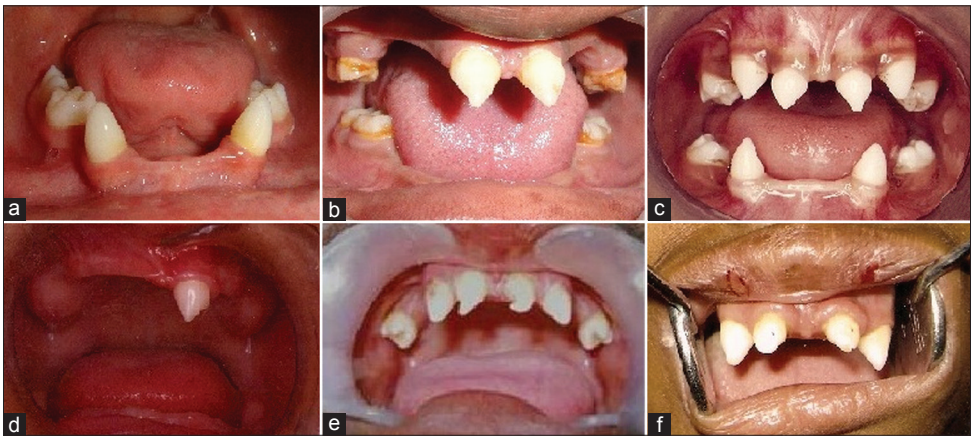


Figure 2: Intra oral photographs of patients with ectodermal dysplasia. Note- partial anodontia and conical-shaped teeth

the adjective anhydrotic to hypohydrotic because the persons termed as anhydrotic were not truly devoid of sweat glands.[12] Clouston in 1939 and Lowry *et al.* in 1966 described ED as a genetic entity.[13] Autosomal dominant disorder. Hydrotic ED is also known as Clouston's syndrome.[11]

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.[1,14,15] They represent a large and complex group of diseases comprising more than 170 different clinical conditions.[2,15,16] There are two major types of ED depending on the number and functionality of the sweat glands; Hypohydrotic and Hydrotic.[15,17] Hypohydrotic type, also called as anhydrotic type, is the most common ED (80%) and is often inherited as an X-linked disorder (XLEDA).[14] 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.[10,15] The hydrotic form usually has normal sweat glands and the condition is inherited as autosomal dominant. This form also affects teeth, nails and hair. But the hereditary patterns and

nail and sweat gland manifestations tend to differ from hypohydrotic type. It is termed as Clouston's syndrome.[10,15]

The purpose of the present retrospective study was to review and analyze 19 cases of ED with emphasis on clinical manifestations and parent's marriage history of these patients.

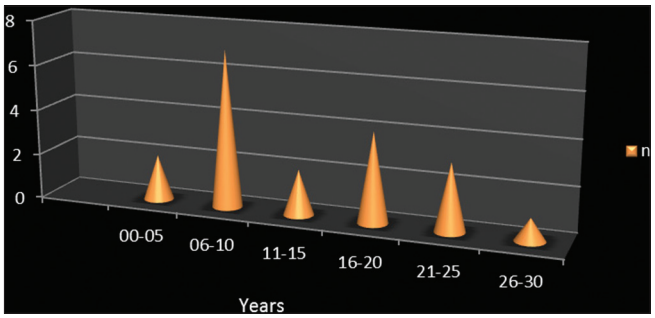
MATERIALS AND METHODS

The present hospital-based retrospective study was conducted by assessing the clinical records of cases of ED from year 2008 to 2012, available in the archives of the department. The permission to undertake this study was obtained from the Institutional ethics committee. The data of total 19 cases of ED were included in the study. The clinical photographs and panoramic radiographs [Figure 3] formed the basis of the present study. The descriptive data of these patients was evaluated and analyzed with respect to the age, gender, family history of consanguineous marriage, clinical and radiographic findings; and compared with previously documented data in the literature.

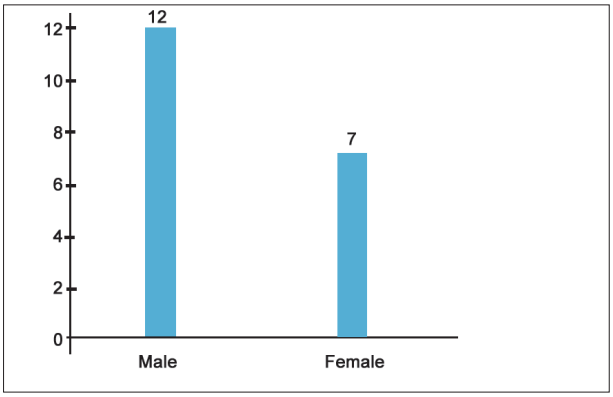
RESULTS AND OBSERVATIONS

In the present study, we observed that total nineteen cases of ED reported to our department were in the age group of 4-30 years with the mean age of 12.89 years. The maximum numbers of patients were in the age group of 6-10 years (36.88%) [Graph 1]. The more number of males (63.16%) were affected then females (36.84%), with a ratio of 1.7:1 [Graph 2]. The hydrotic type of ED was seen in four (21.05%) cases whereas hypohydrotic type was noted in 15 (78.95%) cases [Graph 3]. Related to the marriage history of parents, the total of 12 parents (66.67%) had consanguineous marriage and had 13 (68.42%) offsprings affected with ED; whereas six parents (33.33%) had nonconsanguineous marriage history and had 6 (31.58%) offsprings affected with ED [Table 1]. One sibling was affected in one family to the parents having history of consanguineous marriage [Figure 4].

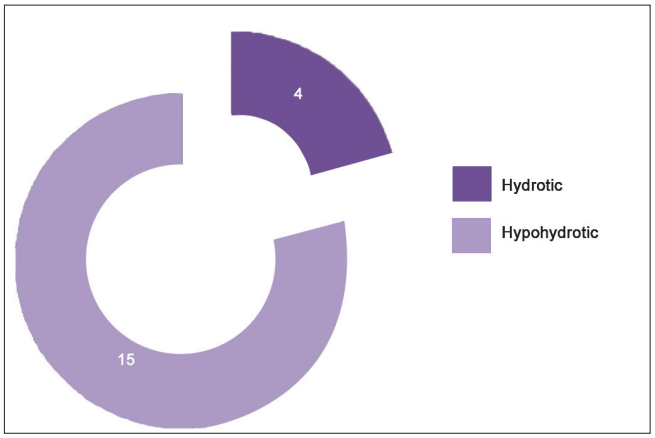
Related to the general manifestations observed in the present study, 18 cases (94.74%) had dry skin; 8 cases (42.11%) had scaly skin; all the 19 cases (100%) had sparse hair on scalp, eyebrows and eyelashes; 12 cases (63.18%) had frontal bossing; 11 cases (57.89%) had saddle nose; 9 cases (47.37%) had hypertelorism; 10 cases (52.63%) had nail abnormality; 4 cases (21.05%) had normal sweat glands; 15 ca ses (78.95%) had abnormal sweat glands; 10 cases (52.63%) had hypoplastic maxilla; 11 cases (57.89%) had protuberant lips; 4 cases (21.05%) had palmoplantar keratosis and 16 cases (84.21%) had



Graph 1: Age-wise distribution



Graph 2: Gender-wise distribution



Graph 3: Type of ectodermal dysplasia

Table 1: Corelation of ectodermal dysplasia cases with parent's marriage history

Parents Marriage history	Total	%	Affected patients	%
Consanguineous	12	66.67	13	68.42
Nonconsanguineous	06	33.33	06	31.58

wrinkled and hyperpigmented facial skin especially surrounding the eyes [Table 2].

Related to the oral manifestations observed in the present study, 18 cases (94.74%) had partial anodontia and 1 case (05.26%) had complete anodontia; 16 cases (84.21%) had conical shaped teeth; 13 cases (68.42%) had high arched palate; 19 cases (100.00%) had thin alveolar bone; 4 cases (21.05%) had taurodontism and in 1 case (05.26%) cleft lip and cleft palate [Table 3]. The number of teeth present in all the cases ranged from 0 to 19 [Graph 4].

DISCUSSION

ED is a rare congenital hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo. ED is usually transmitted as an X-linked recessive trait, in which gene is carried by the females and manifested in males.[1,18] Hence men are more frequently and severely affected.[16] The heterozygote usually show minor defects. In the present study, more number of males were affected then females with a ratio of 1.7:1.Our findings did not match with the findings of Yavuz Izzet *et al*.,[1] wherein the males and females were equally affected. We also noted that, hypohydrotic type was more common then hydrotic type. This finding was similar to the findings of Tauseef Kamran *et al*.[16] and Giansanti *et al*.[19]

The Latin word consanguineous means 'of common blood'. A consanguineous marriage is one in which two individuals related by blood, get married and have children. Consanguineous persons have at least one common ancestor in the preceding few generations. The risk of birth defects is higher in consanguineous marriages because there is a greater chance of two related individuals sharing a common harmful gene and both of them passing it on to the child. The risk and type of birth defects in consanguineous marriages varies according to how closely the couple is related and thus the chance of inheriting a recessive disorder is increased. Most consanguineous couples can have normal healthy children.[20] It is significant to note that the marriage history of parents of all the cases revealed that history of consanguineous marriages were more than nonconsanguineous marriages. This finding when correlated with the number of offspring affected with ED, it was distinctly observed that a consanguineous marriage was directly proportional to ED. Various database search could not reveal studies on this variable. The result of our analysis directs towards the need of establishing the prevalence and occurrence of ED in children born to consanguineous parents.

Table 2: General manifestations observed in cases of ectodermal dysplasia

Skin		Frontal bossing	Saddle nose	Sparse hair		Hypertelorism	Nail abnormality	Sweat gland function		Hypo-plastic maxilla	Protuberant lips	Wrinkled and hyper-pigmented facial skin	Palmo-plantar keratosis
Dry	Scaly			Scalp	Eyebrows, eyelashes			Normal	Abnormal				
√	√	√	√	√	√	√	√	√	√	√	√	√	√

√: Denotes present

Table 3: Oral manifestations observed in cases of ectodermal dysplasia

Case no.	Total number of teeth present	Anodontia	Conical shaped teeth	High arch palate	Thin alveolar bone	Taurodontism	Cleft lip and Palate
1	08	Partial	√	√	√		
2	11	Partial	√	√	√		
3	10	Partial	√	√	√		
4	01	Partial	√		√		
5	16	Partial		√	√	√	
6	04	Partial	√	√	√		
7	13	Partial	√		√		
8	05	Partial	√		√		
9	08	Partial	√	√	√	√	√
10	16	Partial			√		
11	Nil	Complete			√		
12	06	Partial	√		√		
13	15	Partial	√	√	√		
14	14	Partial	√	√	√	√	
15	14	Partial	√	√	√		
16	19	Partial	√	√	√		
17	04	Partial	√	√	√	√	
18	07	Partial	√	√	√		
19	10	Partial	√	√	√		

√: Denotes present

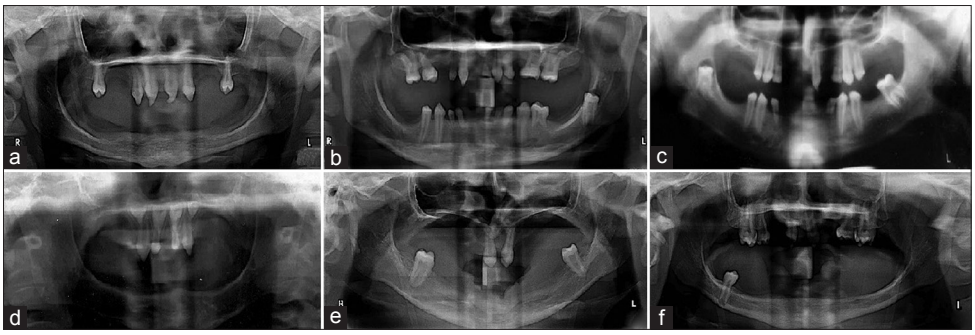


Figure 3: Panoramic radiographs showing partial anodontia, conical-shaped teeth and thin alveolar bone

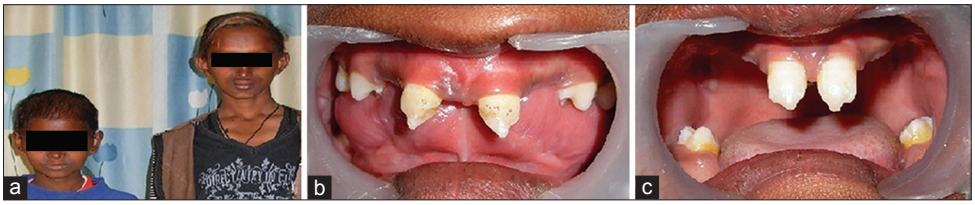


Figure 4: Siblings affected with ectodermal dysplasia

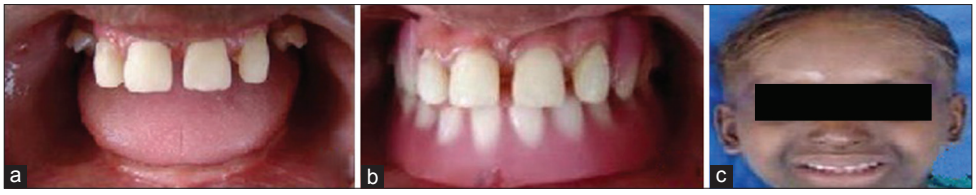
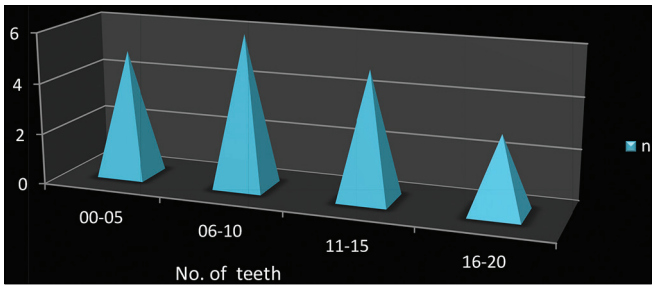


Figure 5: Oral rehabilitation in ectodermal dysplasia

ED manifests with numerous clinical features, both general and oral. The characteristic signs of ED include sparse hair, anodontia or hypodontia and inability to sweat due to lack of sweat glands.[4] All the cases in the present study were diagnosed on the basis of these features. We noticed that partial anodontia was seen in majority of the patients and complete anodontia in one case. Multiple missing teeth were seen more in the mandible and both the deciduous and permanent dentition were involved. These finding was analogous to the studies of Nunn *et al.*[21] and Ruhin *et al.*[22] Anodontia was the most important problem faced by most of our patients and were inclined towards the replacement



Graph 4: Number of teeth present

of the same. The anterior teeth were found to be smaller in size and had conical peg-like shape. In addition, other features like dry skin, scaly skin, frontal bossing, saddle nose, hypertelorism, nail dystrophy, hypoplastic maxilla, protuberant lips, palmo-plantar keratosis, wrinkled and hyperpigmented facial skin, high arched palate, thin alveolar bone, taurodontism and cleft lip and cleft palate, were observed in most of the cases. The studies conducted by Yavuz Izzet *et al.*[1] and Ruhin *et al.*[22] also had similar observations.

CONCLUSIONS

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. A multidisciplinary team comprising of dermatologist, psychiatrist, stomatologist, orthodontist, prosthodontist and pedodontist have responsibility to rehabilitate these patients [Figure 5]. Significantly in our study, 68% of the cases had positive family history of consanguineous marriage among their parents. Related to this, we did not come across any such studies who have established a possible association between ED and consanguineous marriages. The results of our study will promote the need of establishing the prevalence of ED in children born to parents with consanguineous marriages and also highlights the importance of taking relevant family history.

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