Ectrodactyly, ectodermal dysplasia, cleft lip, and palate (EEC syndrome)

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Abstract

The ectodermal dysplasias (EDs) are a large and complex group of diseases. More than 170 different clinical conditions have been recognized and defined as ectodermal dysplasias. Commonly involved ectodermal-derived structures are hair, teeth, nails, and sweat glands. In some conditions, it may be associated with mental retardation. We report a case of 10-year-old male child with ectrodactyly, syndactyly, ED, cleft lip/palate, hearing loss, and mental retardation.

Keywords: Ectodermal dysplasia, ectrodactyly, EEC syndrome

Introduction

Ectodermal dysplasia (ED) syndrome is a large, heterogeneous group of inherited disorders, the manifestations of which could be seen in more than one ectodermal derivative. These tissues primarily are the skin, hair, nails, eccrine glands, and teeth. It is an X-linked recessive disorder.[1]

More than 170 distinctive syndromes have been described with all possible modes of inheritance. The most common syndromes within this group are hypohidrotic ED and hidrotic ED. Several ED syndromes may manifest in association with midfacial defects, mainly cleft lip and palate.[1]

In 1961, Rosselli and Gulienetti[2] reported four patients with hypohidrosis, hypotrichosis, microdontia, dystrophic nails, cleft lip and palate, deformities of the extremities, and malformations of the genitourinary system. Syndactyly was the prominent digital deformity.

Rudiger *et al*.[3] and Freire-Maia[4] in 1970 reported a very similar clinical condition and described it as EEC syndrome that is ectrodactyly—ED—cleft lip/palate syndrome.

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We report a case of 10-year-old male child with EEC syndrome, presented as ectrodactyly, ED, cleft lip and palate, mental retardation, and hearing loss.

Case Report

A 10-year-old male patient reported to the Department of Pedodontics with the chief complaint of multiple decayed teeth since past 2 years. The medical history revealed operated cleft lip of left side, intolerance to heat, and less sweat production.

Family history: There was no history of consanguineous marriage between the parents. In prenatal history, no information worth mentioning was obtained.

On extraoral examination, the patient had dry skin with sparse hair on the body and scalp. Hair present were fine in texture and lighter in color. The skin was warm and dry. There was no hair on the back of the hands. His face was narrowed with a broad nose and a prominent chin. His upper lip showed surgical scar related to cleft lip operation. Lower eyelids of both sides showed sparse eyelashes [Figure 1]. Syndactyly of middle and ring finger was present in the right hand and ectrodactyly of index finger of right hand, third and fourth toes of right feet was also appreciated [Figures 2 and 3]. The parents had seldom noted sweating. There was impairment of hearing but because of lack of cooperation, audiometric analysis could not be performed. The speech defect was attributed to anatomical lip and palate impairment and to his psychomotor retardation. Evaluation of psychomotor development showed moderate retardation. X-ray of skull was also normal.

Intraoral examination showed rotated 21, missing 12 and 22, carious 54, 55, 16, 63, 64, 65, 26, 74, 36, 84, 85, 46 [Figures 4 and 5]. An orthopantomograph was made which revealed congenitally missing maxillary lateral incisors, maxillary left permanent canine, both maxillary second premolars, and mandibular right second premolar. Taurodontism present involving all first and second deciduous molars and first permanent molars [Figure 6].

All carious teeth were restored with Glass ionomer cement and composites. 75 was extracted due to its poor prognosis.

Discussion

The first published report of a patient with ED was in 1848 by Thurman,[5] but the term "ectodermal dysplasia" was coined by Weech[6] in 1929. The condition is thought to occur in approximately 1 in every 100,000 live births.[7]

Weech AA[6] suggested the term "hereditary ectodermal dysplasia" and described three essential features, which are as follows: (a) tissues affected are principally of ectodermal origin, (b) the defects are developmental anomalies, and (c) the hereditary tendencies are strongly developed.

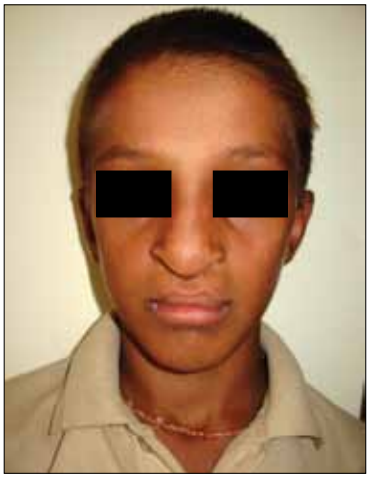

Figure 1: Facial view

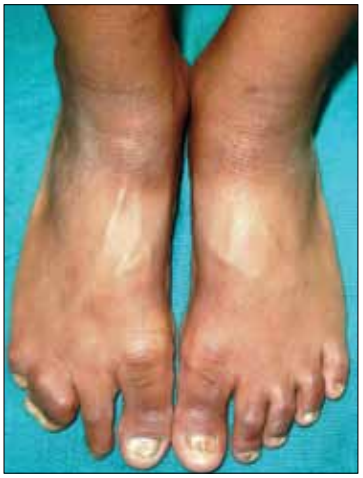

Figure 3: Ectrodactyly of third, fourth toes of right feet

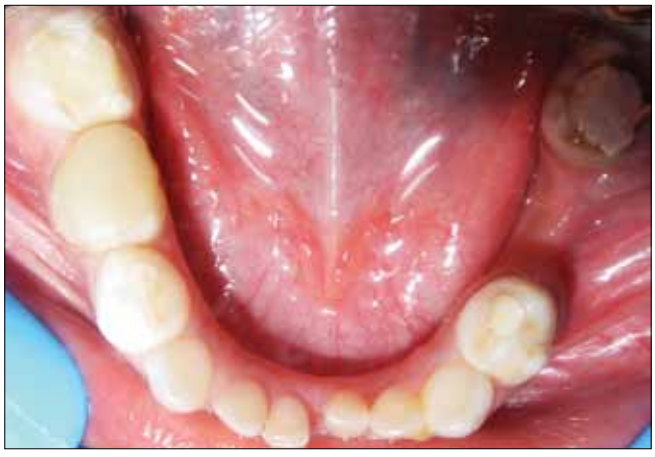

Figure 5: Mandibular arch

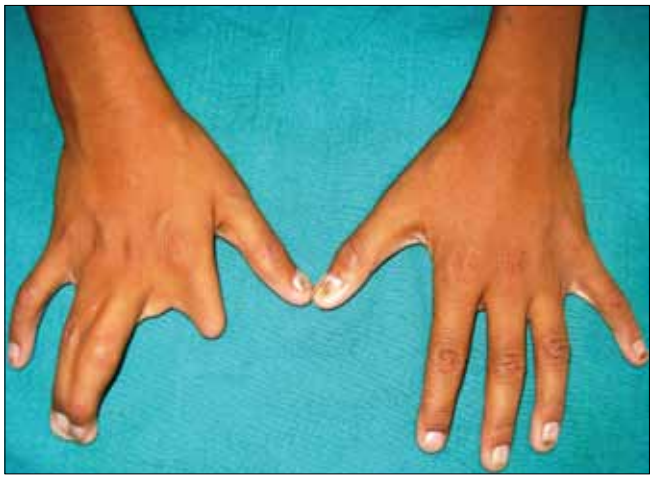

Figure 2: Ectrodactyly of index finger, syndactyly middle and ring finger of right-hand and dystrophic nails

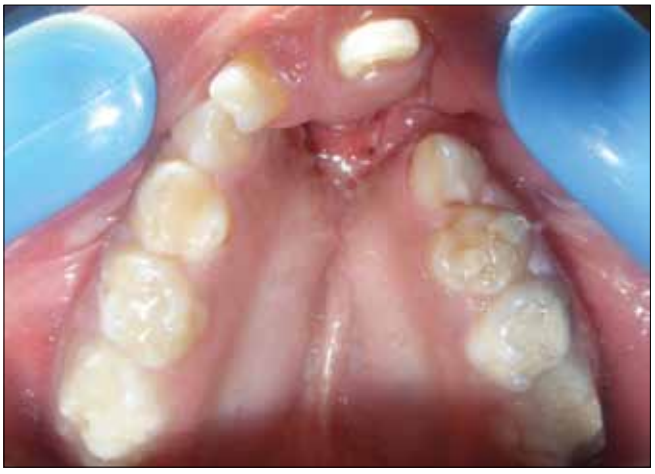

Figure 4: Maxillary arch

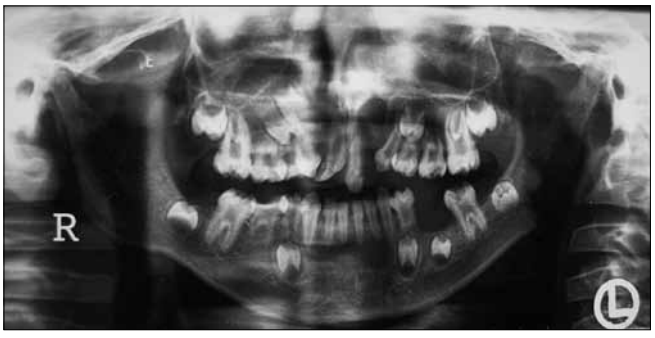

Figure 6: Orthopantomograph

The first classification system for EDs was proposed by Pinheiro and Freire-Maia in 1982.[8] The patients are classified into subgroups based on the presence or absence of the following: (1) trichodysplasia, (2) dental abnormalities, (3) onychodysplasia, and (4) dyshidrosis.

Overall, EDs were classified into either group A disorders, which were manifested by defects in at least two of four classic ectodermal structures as defined above (1 to 4), with or without other defects, and group B disorders were manifested by a defect in one classic ectodermal structure, in combination with a defect in one other ectodermal structure, i.e., ears, lips, and dermatoglyphics.

A condition characterized by only ectodermal signs is called a pure ectodermal dysplasia; if it combines ectodermal signs and malformations, it is termed as ectodermal dysplasia malformation syndrome or an ED syndrome.[9] Our case belongs to ED syndrome category.

EEC syndrome (ectrodactyly, ED, and cleft lip/palate) is a relatively common syndrome which includes most of the manifestations of the syndactyly, ED, and cleft lip/palate syndrome.

The term EEC syndrome was coined by Rudiger *et al*. in 1970.[3] It is a complex pleiotrophic multiple congenital anomaly/dysplasia syndrome in which any one of the cardinal signs is present in variable expression.

Ectrodactyly means absence of all or part of one or more digits. Ectrodactyly appears to be the only constant component of this syndrome. EEC syndrome may exist in two forms: one with cleft lip with or without cleft palate and with cleft palate alone. Both forms are inherited as autosomal dominant traits with variable expressivity, involving partial penetrance in the former and complete penetrance in the latter.[10]

The individual frequency of ectrodactyly is reported to be 1.5 per 100,000 live births and 1 per 100,000 live births for cleft palate with or without cleft lip.[10] The occurrence of all three disorders in one, i.e., ectrodactyly, ED, and cleft lip/palate is reported to be approximately 1.5 per 100 million.[3] In our case, ectrodactyly was present with respect to right-hand index finger, along with the syndactyly of right-hand middle and ring finger and third, fourth toes of right foot. The syndactyly involved right-hand fingers and toes, but the function of extremities was not profoundly affected though cases reported by most of the other authors[3,4,10] have had more severe limb defects.

It has been suggested that the EEC syndrome with complete and incomplete forms and other similar conditions form a "community" or "family" of different although very similar syndromes, as suggested by Pinheiro and Freire-Maia.[8]

Our case showed similar problems as that of Bowen and Armstrong[11] showing syndactyly, ED with abnormalities of scalp hair, and teeth and hidrotic problems, but with unilateral cleft lip and palate instead of bilateral cleft lip and palate. In addition, he was mentally retarded and hearing loss was also present.

We suggest that our case represents either a variant of EEC syndrome or a new syndrome with autosomal recessive inheritance.

Sparse and kinky hair with small, carious teeth, and broad nose and a prominent chin gave a typical facial appearance to our patient, similar to cases of Brill *et al*.[12] In addition, our case had congenitally missing both upper and lower right second premolars, missing upper permanent lateral incisors, missing upper left permanent canine, and taurodontism in all deciduous molars and first permanent molars. In our case, there was congenital absence of teeth unrelated to alveolar ridge clefting and the same was reported by Bowen and Armstrong 1976.[11] His skin was dry and thickened and sweating was absent according to the parents.

Mental retardation has been reported in the EEC syndrome (Rudiger *et al*. 1970[3] and Brill *et al*.[12]) but appears to be infrequent and the same was present in our case.

Conclusion

Ectrodactyly—ED—cleft lip/palate (EEC syndrome) is a rare form of ED, the symptoms of which can vary from mild to severe. The most common symptoms found in patients with EEC syndrome are as follows: missing or irregular fingers and/or toes (ectrodactyly), abnormalities of hair and glands, cleft lip and/or palate, or unusual facial features, as well as abnormalities of eyes and urinary tract. For accurate diagnosis of patients with this rare syndrome, a careful and thorough examination should be carried out.

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