Ectrodactyly, Ectodermal Dysplasia with Cleft Palate: A Case Report

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Abstract- Ectrodactyly-ectodermal dysplasia-cleft lip or palate syndrome (EEC syndrome) is characterized by the triad of ectrodactyly, ectodermal dysplasia and facial clefting (lip/palate). However, manifestation of all the three anomalies is very uncommon. The incomplete forms, with absence of one or more of the cardinal signs, are often noted. Neither of the two cases, recently reported from India, had all the three anomalies. We report a case of EEC with all three classical features

Index Terms- Ectrodactyly, Ectodermal dysplasia, cleftpalate

I. INTRODUCTION

Ectrodactyly-ectodermal dysplasia-cleft lip or palate syndrome (EEC syndrome) is characterized by the triad of ectrodactyly, ectodermal dysplasia and facial clefting (lip/palate). It was first described by Cockayne in 1936.[1] The simultaneous presence of these three anomalies is extremely rare.[2] Apart from these cardinal features, lacrimal abnormalities, urogenital abnormalities, mental retardation and conductive deafness may be present.

EEC is inherited as autosomal dominant trait of low penetrance and variable expressivity. Sporadic cases have also been reported. There were two interesting (one familial and one sporadic) case reports of EEC from India in 2006-[3,4] Neither of them had the combination of all three anomalies. We report an autosomal dominant familial case in a neonate with all classical features of EEC.

II. CASE REPORT

A 14 days old term male neonate, birth weight of 2.9 kg, 2nd born, out of non-consanguinous marriage was admitted to SRM Medical college hospital nicu unit, for severe pallor (Hb-5.3gm) with history of preceding jaundice, secondary to Rh incompatibility (mother A-ve, baby A+ve) .

On examination, multiple congenital anomalies were noted. Systemic examination revealed no abnormality clinically. Rh incompatibility and anemia were treated accordingly. Baby was investigated further for other possible associated anomalies.

Figure 1 shows midline cleft palate.

Figure 2 shows ectrodactyly of both foot, left foot shows big toe, fusion of 3rd and 4th toe and missing of 2nd and 5th toe. Left foot shows big toe and prominence of 3rd and 4th toe, absence of 2nd and 5th toe.

Figure 3 shows ectodermal dysplasia in form dystrophic nails in 2nd and 3rd finger.

Figure 4 shows ear anomaly (prominent anti-helix).

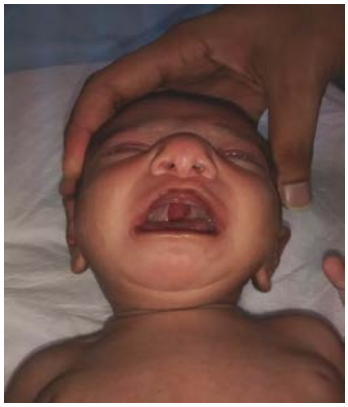

Figure 1

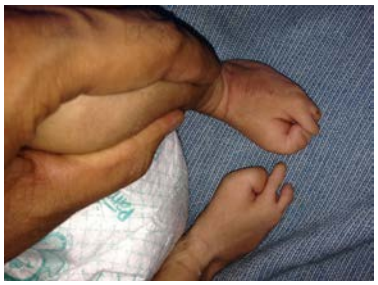

Figure 2

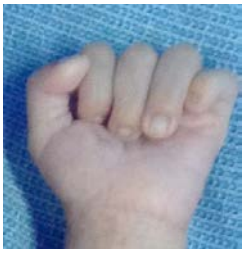

Figure 3

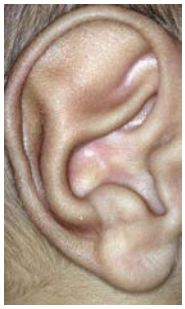

Figure 4

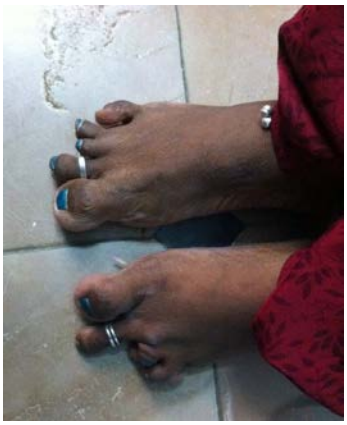

Figure 5

Figure 5, showing feet deformity of his Mother.

III. DISCUSSION

Ectrodactyly refers to deficiency or absence of one or more of central digits of hands and feet. Ectodermal dysplasia involves organs derived from embryonic ectoderm. These abnormalities involve both the superficial ectodermal layer as well as the deeper mesoectodermal layer, formed from the neural crest. Other ectodermal anomalies include mild hypohidrosis; coarse, dry hair with hypotrichosis; xerostomia; dystrophic nails; dental enamel hypoplasia with microdontia. Associated anomalies include blepharophimosis, lacrimal duct anomalies, deafness, choanal atresia and abnormalities of genitourinary tract.

The EEC syndrome results from simultaneous ectodermal and mesodermal developmental defects. Although any of the three cardinal signs can present with variable expression and can occur as a separate entity each, combination of all three anomalies appears to be a rare occurrence. [2] There is lot of doubt whether the incomplete forms reflect a reduced expression of the gene or one or more separate clinical entities. Some authors claim that clefting in EEC always involves the lip with or without the palate and use this marker as means to distinguish from other syndromes. However Buss *et al*. suggested that the diagnostic criteria of EEC should include ectodermal dysplasia other than two of the following additional features: ectrodactyly, cleft lip/palate and lacrimal duct abnormalities. [5]

Thakkar and Marfatia[3] reported a family with ectrodactyly and ectodermal dysplasia without cleft lip/palate. According to Wallis *et al*., who reported a similar presentation with ectrodactyly and ectodermal dysplasia (hypotrichosis and abnormal dentition) without cleft lip/palate, this represents a distinct clinical entity. [6]

There was another report of a sporadic case by Cyriac and Lashpa. Besides ectrodactyly and cleft palate, the patient had maxillary hypoplasia and low-set ears. But he did not have ectodermal dysplasia. [4]

Paucity of reports of EEC cases with all manifestations has prompted us to report this case. Our patient had the triad of ectrodactyly involving feet; ectodermal dysplasia in form of dry rough skin with sparse, hypopigmented hair; and midline cleft palate, associated with dystrophy of finger nails, deformed ear, and broad nasal bridge. USG abdomen and 2D Echocardiogram were normal. All the characteristic features of EEC were noted. Mother had the minor deformity of feet (Shown in Figure 5) suggesting that it is autosomal dominant inheritance which is the commonest form of familial variety.

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