

Case Report

Ectrodactyly, ectodermal dysplasia with cleft lip and palate: a case report

Sankha Koley, Sanjiv V Choudhary, Atul Salodkar, Vikrant Saoji

Department of Dermatology, J.N.M.C. Sawangi, Wardha, Maharashtra

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 sporadic case of EEC with all three classical features.

Key words: ectrodactyly, ectodermal dysplasia, cleft lip, cleft palate.

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 2006-7.^{3,4} Neither of them had the combination of all three anomalies. We report a sporadic case with all classical features of EEC.

Case report

A 4-year-old child was referred to our outpatient department for dry, rough skin since birth. On examination the child had unilateral

left sided cleft lip and cleft palate. Scalp hair was sparse, blond and hypopigmented (**Figure 1**). There were splitting of right hand and left foot i.e. ectrodactyly (known as split hand split foot malformation: SHFM). In the right hand the middle and the index fingers were missing. The remaining three fingers were parted; thumb on lateral side and two fingers on medial side (**Figures 2 and 3**). In the left foot the middle digit was missing and the medial two digits were fused (**Figure 4**). Skin of the abdomen and extremities were dry and there was absence of sweating. The lacrimal ducts were normal and there were no auditory and neurological defects. Clinically no other system was involved. No other member was affected in the family. Routine investigations were within normal limits. X rays of the hand, feet and skull showed features corresponding to the clinical picture.

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

Address for correspondence

Dr. Sankha Koley
9 Mandeville Gardens;
Flat No.10C;
Calcutta: 700019, India
E-mail - skoley@gmail.com

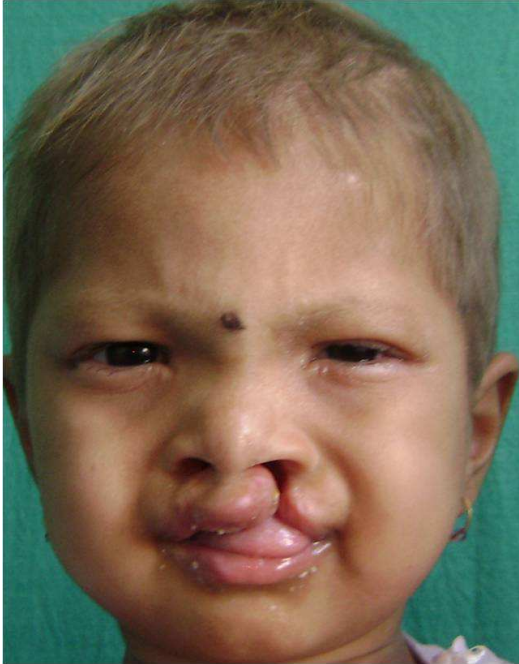


Figure 1 Left-sided cleft lip and cleft palate with hypopigmented scalp hair.



Figure 2 Lobster claw deformity of right hand

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, the



Figure 3 Lobster claw hand.



Figure 4 Lobster claw deformity of left foot.

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 Marfatia³ 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 sporadic cases of EEC syndrome has prompted us to report this case. Our patient had the triad of ectrodactyly of a hand and a foot; ectodermal dysplasia in form of dry rough skin with sparse, hypopigmented hair; and unilateral left sided cleft lip and

palate. All the three characteristics of EEC were noted.

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Authors Declaration

Authors are requested to send a letter of undertaking signed by all authors along with the submitted manuscript that:

The material or similar material has not been and will not be submitted to or published in any other publication before its appearance in the *Journal of Pakistan Association of Dermatologists*.