Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome: A case report of "Incomplete syndrome"

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Abstract

Ectrodactyly, ectodermal dysplasia, and cleft lip/palate (EEC) is a rare syndrome having ectrodactyly, ectodermal dysplasia, and cleft lip/palate. So far, very few cases have been reported in literature. However, we report a case of incomplete EEC syndrome having ectrodactyly and cleft lip and palate with absence of signs of ectodermal dysplasia with no other systemic anomalies. Other feature noted is the syndactyly of toes which is reported rarely in this syndrome. A multidisciplinary approach for treatment is needed which is co-ordinated by pedodontist or pediatrician.

Keywords: Cleft lip and palate, ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome, ectrodactyly, ectodermal dysplasia, syndactyly

Introduction

The combination of ectrodactyly, ectodermal dysplasia, and cleft lip/palate is known as EEC syndrome.[1,2] The acronym EEC was first used by Rudiger *et al.* with description of a case.[1] This disorder is characterized by deformities of hand or leg fingers (ectrodactyly); anomalies of hair, teeth, nails, nasolacrimal ducts, sweat glands etc. (ectodermal dysplasia); and cleft lip/palate. Other less common findings include microcephaly, mental retardation, deafness or hearing defect, and genitourinary anomalies.[3] These genitourinary anomalies are often present as complications of syndrome.[4] Every time all features of this syndrome are not present, so the term "incomplete EEC syndrome" was coined by Pries *et al.* for a combination in which one or more components are missing.[5] Kuster and Majewski reported eight cases in two families without ectrodactyly coining term "oligosymptomatic EEC syndrome" and suggested that ectrodactyly is not mandatory for syndrome diagnosis.[6]

This is a rare syndrome with estimated incidence of 1.5 per million births.[7] Exact underlying cause of this disorder is not known, but it is considered to be due to genetic mutation. Although majority are sporadic cases, some families with EEC syndrome have been reported showing autosomal dominant inheritance with incomplete penetrance and variable expression.[7] Candidate chromosomal regions include 7q21.3, which is a prime locus for ectrodactyly; 7q11 and 9p2l are breakpoints in family with a translocation and EEC syndrome.[7] Also, a balanced reciprocal translocation between 7q11.21 and 9p12 is localized in three generations of a family with EEC syndrome.[7] Other possible candidate genes for such a syndrome could include transforming growth factors alpha and beta.

Herewith, we report a patient with incomplete EEC syndrome who had reported to our department.

Case Report

A twenty-day-old boy who was second child of healthy unrelated parents was brought to our department. Child was born after 38 weeks of gestation and weighing 2 kg at the time of birth, without any complications during pregnancy and delivery and no anomaly was detected during routine prenatal examinations. The chief complaint was nasal regurgitation of milk in supine position. On oral examination, child was seen to have a complete cleft lip and palate on right side and cleft of hard and soft palate on left side without involvement of alveolus [Figures 1 and 2]. On extraoral examination, ectrodactyly of both hands [Figure 3] and feet [Figure 4] was seen. Both hands showed claw shaped deformity with only two non-fused fingers present. Radiograph showed only two phalanges in each finger with normal development of other bones in hand [Figure 5]. Feet had three toes with absence of 2nd and 3rd toe. There was syndactyly between 4th and 5th toe. Radiograph showed presence of phalanges of all toes, except 2nd toe which was incompletely formed [Figure 6]. Abnormal continuous discharge from eyes of infant was noted which may

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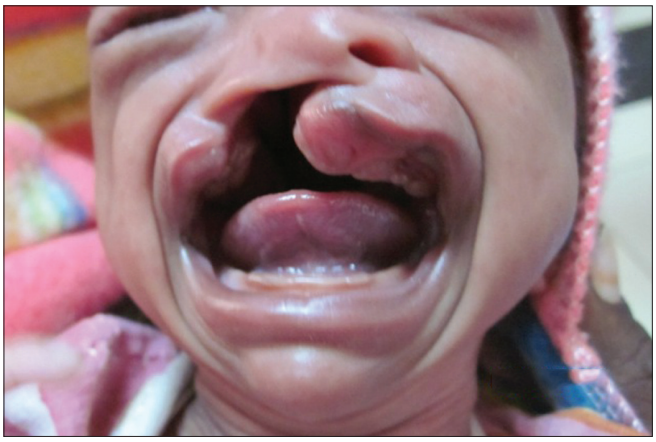

Figure 1: Infant with cleft lip and palate

Figure 2: Cast showing palatal defect

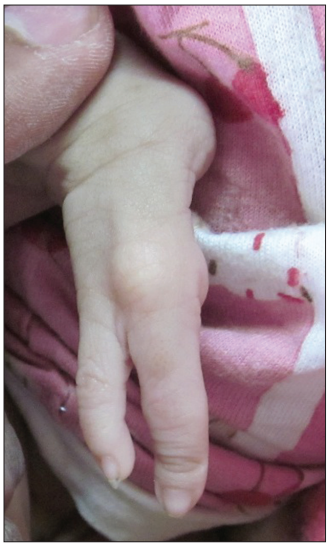

Figure 3: Ectrodactyly of hand

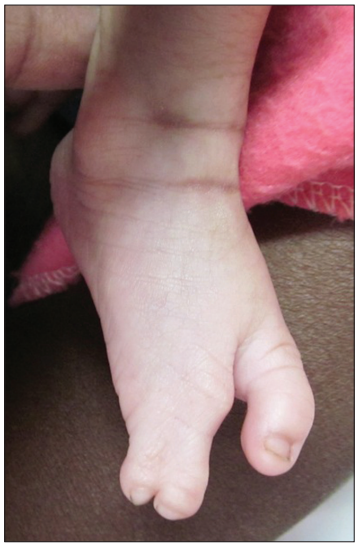

Figure 4: Ectrodactyly of foot with syndactyly of 4th and 5th toe

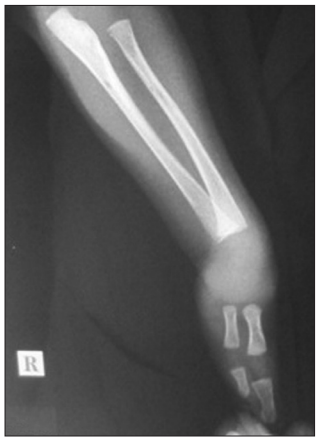

Figure 5: Radiograph of hand

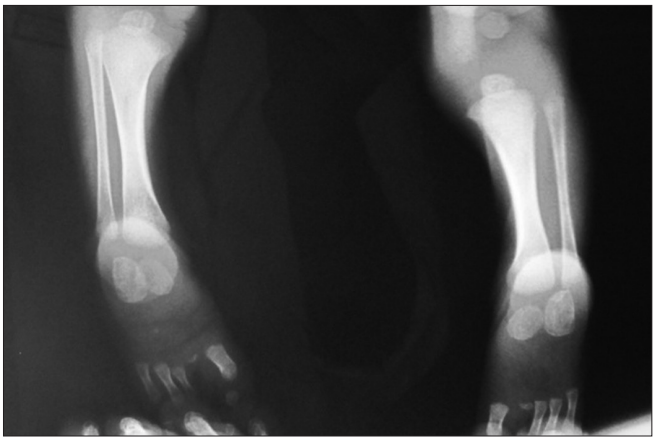

Figure 6: Radiograph of feet

be due to nasolacrimal duct anomaly. But, no other anomaly indicating ectodermal dysplasia was seen in hairs or nails. Complete medical investigations were done which revealed no genitourinary or other systemic abnormality.To treat his chief complaint, an impression of the palatal defect was made using elastomeric impression material and an obturator was constructed using cold cure acrylic material which was later relined using soft tissue liner [Figure 7]. Patient is scheduled for primary cheilioplasty around 4 months and surgical repair of palate around 18 months of age.

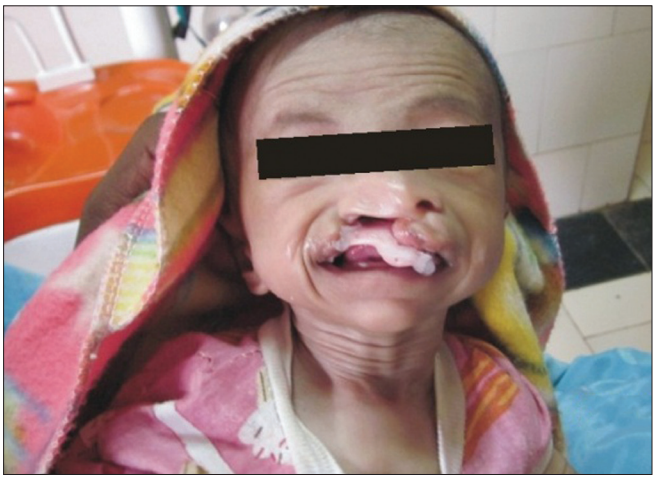

Figure 7: Infant wearing prepared obturator

Discussion

This syndrome results from a developmental abnormality that simultaneously affects the ectodermal and mesodermal tissues. Mostly, each of these defects occurs singly but rarely, a combination of ectrodactyly, ectodermal dysplasia, and cleft lip/palate occur together. At least 180 cases with EEC syndrome have been described in the world, of which only 70 had complete range of symptom.

Management of this syndrome requires multidisciplinary approach.[8] Early diagnosis will allow parents to gain access to accurate counseling. The multidisciplinary treatment team is co-ordinated by a pedodontist/pediatrician and consists of oral and maxillofacial surgeon, plastic surgeon, ophthalmologist, and renal specialist. Managing cases of EEC syndrome is difficult from both practical and psychological viewpoint because patient has a significant facial deformity. Here, pedodontist will play an important role in preserving primary dentition which will encourage maxillary and mandibular growth. Severe ectodermal manifestations can be managed by wigs and cosmetics, but visual complications are difficult to manage. Surgery for duct blockage and corneal grafting can be done.

Prenatal diagnosis has been achieved but high-risk cases should be cautioned because there can be difficulty in identifying affected fetuses, because this condition may have such a variable presentation[9] which was also seen in case of this patient.

In conclusion, the present case is of EEC syndrome which is a rare anomaly and we call for heightened awareness of the possibility of EEC, particularly in pediatric patients presenting with similar clinical picture. This will ultimately help in early diagnosis and better management of the patients with EEC syndrome.

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