International Journal of Integrative Medical Sciences,
Int J Intg Med Sci 2015, Vol 2(3):87-89. ISSN 2394 - 4137
DOI: 10.16965/ijims

Case Report

EEC (Ectrodactyly-Ectodermal dysplasia Clefting) Syndrome in a Newly Born Baby

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ABSTRACT

EEC syndrome is characterized by deformities of the hands and feet (ectrodactyly), abnormalities of the skin, hair, and nails (ectodermal dysplasia), and cleft lip and/or cleft palate (clefting). Other signs and symptoms include dental, eye, skin, and kidney abnormalities. The present case describes the clinical presentation in a premature neonate born with this rare genetic syndrome.

KEY WORDS: Ectrodactyly, Ectodermal Dysplasia, Cleft Palate.

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Online Access and Article Informtaion

Quick Response code

DOI: 10.16965/ijims

International Journal of Integrative Medical Sciences
www.imedsciences.com

Received: 01-03-2015 **Accepted:** 20-03-2015
Reviewed: 01-03-2015 **Published:** 31-03-2015

Source of Funding: Self **Conflicts of interest:** None

INTRODUCTION

EEC syndrome is an autosomal dominant disorder associated mainly with a triad of cardinal signs ectrodactyly, ectodermal dysplasia and cleft lip/palate. Ectrodactyly, also called split hand/foot malformation, is a central reduction defect of the hands and feet that is often found with syndactyly [1, 2]. Ectodermal dysplasia can present as dry skin, sparse hair, dystrophic nails, or hypoplastic teeth, and lacrimal duct obstruction is often present as well [3]. Clefting may affect the lip and/or palate [3]. Other less common findings include microcephaly, mental retardation, deafness or hearing defect, and genitourinary anomalies [1]. Ophthalmological abnormalities include entropion, hypertelorism, absence of lacrimal puncta, hypotrichiasis, blepharitis, photophobia, corneal opacification, and dacryocystitis [4]. The exact prevalence of this syndrome is unknown.

CASE REPORT

A 26 year old primigravida, with a history of non-cosanguinous marriage went into preterm labour at 34 completed weeks. Antenatal history was significant for history of polyhydraminos. A preterm male baby was born by normal vaginal delivery. At birth, the baby had immediate cry. He was transferred to the neonatal intensive unit for prematurity with multiple congenital anomalies.

The weight of the baby was 1675gm; length 42cm and head circumference 31cm. Skin was dry and scaly. There was a minor facial dysmor-

phism with retrognathia and antimongloid slant of eyes. He also had a cleft palate without cleft lips. Skeletal anomalies were bilateral claw foot. Ophthalmological examination revealed bilateral corneal opacity and absent lacrimal punctum.

There were no aural, urogenital, or neurological anomalies on clinical examination. There was no family history of EEC syndrome.

X-rays of the hands and feet showed radiological findings corresponding to the clinical features. There was no urogenital anomaly on ultrasonography.Screening 2D ECHO was norma.l

Fig. 1: Lower limb showing absent 2nd, 3rd and 4th toes.

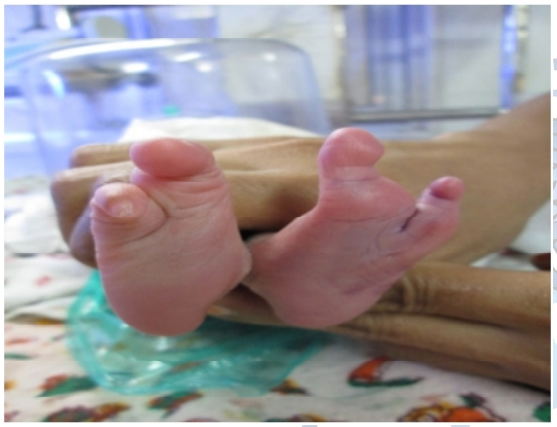


Fig. 2: X ray of lower limb showing ectodactyly.

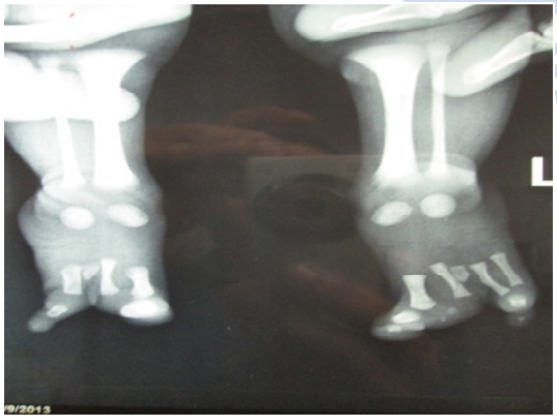


Fig. 3: Shows skin is dry and scaly, minor facial dysmorphism.

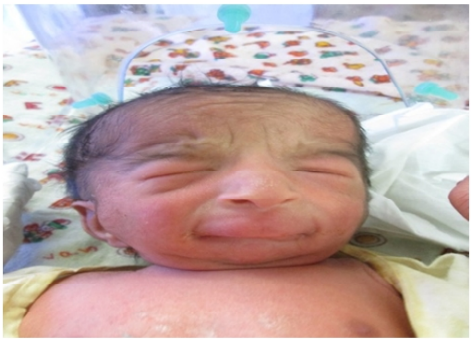
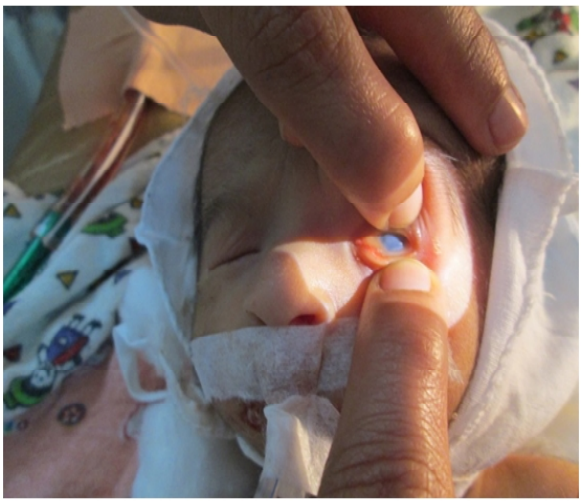


Fig. 4: Shows corneal opacity and absence of lacrimal punctum opacity.



DISCUSSION

The term EEC was first used by Rudiger et al. with description of a case [5]. EEC syndrome is known to be a disorder of variable expressivity and reduced penetrance [6]. The fact that EEC syndrome may or may not show all the three cardinal signs as well as associated symptoms may vary in each case makes the correct and timely diagnosis a challenging task. The diagnosis of EEC is based on clinical examination, X-rays of the limbs and jaw, and, according to the associated features, ultrasound KUB region, ophthalmologic examinations, and skin biopsy. Genetic testing may confirm the diagnosis.

The EEC syndrome should be differentiated from the other syndromes that have ectodermal dysplasia and facial clefts such as: Rapp-Hodgkin syndrome, the Hay-Wells syndrome, ankyloblepharon ectodermal and harelip facial cleft and the split-hand and foot malformation syndrome (SHFM).

The abnormalities found in this syndrome can involve not only the ectodermal cells, but also disruption to development in the mesoectodermal layer constituted by the neural crest cells during embryonic development [7,8].The karyotype is usually normal. This disorder has been attributed to mutations in a gene encoding the p63. p63 is a transcription factor that regulates the activity of the tumor suppressor gene TP53. Based on linkage studies and the analysis of chromosomal abnormalities, three loci have been identified in humans [7].

A review of 230 cases from English, German, French, Italian, and Dutch publications found EEC syndrome to include ectrodactyly (84%), ectodermal dysplasia (77%), cleft lip and/or palate (68%), lacrimal tract abnormalities (59%), urogenital abnormalities (23%), and conductive hearing loss (14%)[9]. EEC may or may not present with all the three cardinal signs.

Fryns et al, reported variable manifestations of EEC in 2 families, confirming that no symptom is obligatory for the diagnosis [10]. The present case presented with all the three cardinal features i.e. ectrodactyly, ectodermal dysplasia and cleft palate, also the ocular symptoms (corneal opacity, absence of lacrimal puctum) were present.

Management of this syndrome requires multidisciplinary approach [11]. Plastic surgeon, pediatrician, ophthalmologist and if renal anomalies are associated nephrologist, for dental defects oromaxillary surgeon i.e. depending on the clinical presentation, the different specialties will be required to coordinate. Early and correct diagnosis is important so that parents can be counseled appropriately and timely.

CONCLUSION

The present case highlights the clinical presentation of EEC syndrome in a newborn; this will be helpful for newborn physicians in diagnosing cases with similar clinical presentation. It emphasizes on the importance of knowledge of the classical cardinal features of EEC Syndrome for the clinicians, as well as the fact that EEC syndrome presents high variability in clinical presentation.

Conflicts of Interests: **None**

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How to cite this article: Niraj kumar Dipak, Samir Sheikh, Anita Srinivasan. EEC (Ectrodactyly-Ectodermal dysplasia Clefting) Syndrome in a Newly Born Baby. Int J Intg Med Sci 2015;2(3):87-89. **DOI:**10.16965/ijims