

Incomplete EEC (Ectrodactyly, Ectodermal dysplasia and cleft lip/palate) syndrome with bilateral Entropion: A Case Report

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Abstract—EEC syndrome is a very rare syndrome having ectrodactyly, ectodermal dysplasia and cleft lip/palate as cardinal signs with other variable associated features. Very few cases being reported. It is mostly inherited in an autosomal dominant manner. Such cases need coordinated multidisciplinary approach for treatment. A case of incomplete EEC syndrome having ectrodactyly and cleft palate with absence of the signs of ectodermal dysplasia with no systemic anomalies was reported. Other features noted in this case were syndactyly of fingers and toes with bilateral entropion.

Keywords: Syndrome, Ectrodactyly, Ectodermal dysplasia, Syndactyly, Entropion.

I. INTRODUCTION

EEC syndrome is a genetic developmental disorder characterized by three cardinal signs i.e. ectrodactyly, ectodermal dysplasia and oro-facial clefts (cleft lip/palate).¹ Also known as “split hand – split foot—ectodermal dysplasia –cleft syndrome”. The presence of the cardinal signs together is not mandatory and any of the three can be present with variable expressions.² The exact prevalence it is not known. More than 300 cases have been reported in the literature.³

Every time all features of this syndrome are not present so the term “Incomplete EEC” was coined by Pries et al for a combination in which one or more components are missing.⁴ Kuster and Majanski reported 8 cases in 2 families without ectrodactyly coining the term “Oligosymptomatic EEC”.⁵

The present article is a case report of “Incomplete EEC “having ectrodactyly, cleft palate and entropion with absence of the signs of ectodermal dysplasia with no other systemic anomalies.

II. METHODOLOGY

A rare case of EEC (ectrodactyly, ectodermal dysplasia and cleft lip/palate) was presented in NICU of Mahila Chikitsalaya, a hospital attached to SMS Medical College, Jaipur (Rajasthan) India. Although it was not having all the cardinal signs of this syndrome but having ectrodactyly, cleft palate with other anomalies so called "Incomplete EEC". It is a very rare case so examined and investigated thoroughly to prepare a detailed case report to published.

III. CASE REPORT

A preterm (34-36 weeks) small for gestational age female newborn with a birth weight of 1.7 kg was admitted in the NICU of Mahila Chikitsalaya, SMS Medical College and Hospitals, Jaipur, Rajasthan.

The baby was a product of non-consanguineous marriage, born to a third gravida mother aged 20 years by Caesarian Section at Mahila Chikitsalaya, SMS Medical College and Hospitals, Jaipur, Rajasthan ; the indication being breech presentation. Gravida 1st and 2nd were abortions in the first trimester. In the

present case the antenatal history was uneventful. There was improper antenatal checkup with no antenatal ultrasound done. At birth the baby had an immediate cry with an APGAR score of 7/10 at 1 minute. Baby had mild respiratory distress for which was admitted in NICU. On physical examination, baby had a cleft left hand (absent middle and ring finger, syndactyly of thumb and index finger with a cleft in centre of hand resembling “lobster claw deformity”), bilateral cleft feet (absent 2nd and 3rd toes, syndactyly of 4th and 5th toes with a cleft in the centre of the feet), facial dysmorphism with bilateral microphthalmia and entropion, retrognathia and a partial cleft palate. Genitalia were normal. (Figure 1 & 2)

Figure 1



Lobster claw hand with bilateral entropion

Figure 2



Claw foot with absent middle three toes

Baby passed stool and urine within first few hours of birth. Chest Xray was done which showed mild crowding of the cervical vertebrae otherwise it was normal. Baby was started on Oxygen by hood, intravenous fluids and antibiotics. Blood sugar was normal. Rest of the routine blood investigations and

ultrasound examination could not be done as the attendants took the baby against medical advice within first 24 hours of birth.

IV. DISCUSSION

EEC Syndrome was first described in 1936 by Cockayne EA.⁶ The term EEC Syndrome was first coined by Rudiger et al in 1970.⁷ It is a complex pleiotrophic multiple congenital anomaly / dysplasia syndrome in which any one of the three cardinal signs are present in variable expression.²

The three cardinal signs of the syndrome are ectrodactyly and syndactyly of the hands and feet, cleft lip with or without cleft palate (that can result in speech defects) and abnormalities in several ectodermal structures including skin (i.e. hypopigmented and dry skin, hyperkeratosis, skin atrophy), hair (i.e. fine and sparse hair and eyebrows), teeth (small, absent or dysplastic teeth), nails (nail dystrophy) and exocrine glands (sweat, sebaceous and salivary glands).

A characteristic form of ectrodactyly is the SHFM (Split Hand Foot Malformation) due to the absence of the 3rd digit with clefting into the proximal portion of hand and foot and syndactyly of the remaining digits on each side of the cleft; the hand having a characteristic appearance resembling a “lobster claw”.⁸

Other associated clinical features include abnormalities of genito-urinary system (renal agenesis, urethral atresia, hydronephrosis), conductive or sensorineural hearing loss, choanal atresia, mammary gland / nipple hypoplasia, gland abnormalities like hypoplastic thymus, hypopituitarism, growth hormone deficiency), delayed developmental milestones and malignant lymphoma.

The ophthalmological findings associated with this syndrome include hypertelorism, entropion, trichiasis, punctual scarring, absent meibomian glands, blepharitis, conjunctivitis, corneal scarring, pannus, photophobia, blue iris and albinoid features. Dacryocystitis secondary to congenital duct anomalies was noted by Cockayne in the first published report of the syndrome.⁶

It is a rare syndrome with an estimated incidence of 1.5 per million births. The inheritance is autosomal dominant with incomplete penetrance and variable expression. Exact underlying cause is not known but it is considered to be due to genetic mutation in the codifying gene of a tumour suppressor protein (p63) related to the ectoderm and mesoderm.⁹ Candidate chromosomal regions include 7q21.3 which is a prime locus for ectrodactyly. 7q11 and 9p21 are breakpoints in family with a translocation and EEC syndrome.¹⁰

Diagnosis is based on clinical examination, X-rays of the limbs and according to associated features, kidney ultrasound, ophthalmologic examinations and skin biopsy. Genetic testing may confirm the diagnosis.

Antenatal diagnosis is based on ultrasound examination during the second trimester which may reveal the structural abnormalities. Molecular analysis by chorionic villi sampling or by amniocentesis helps in confirming the diagnosis for families for which the disease causing mutation was identified.

Genetic counselling should be offered to the affected families informing them of the 50% chances an affected person has of transmitting the disease causing mutation.

Management of the clinical features of EEC Syndrome presents a challenge. Early diagnosis, sympathetic and rational attitude and a multidisciplinary treatment approach are necessary for the

physical, psychological and social rehabilitation of such patients. Multidisciplinary approach by orthopedician, plastic and dental surgeons, ophthalmologists, dermatologists and speech therapists.

Prognosis is good with a near to normal life expectancy. Hypohydrosis(reduction / absence of sweat glands) presents the most life-threatening complications, as it can cause seizures, coma and eventually death when not managed correctly.

V. CONCLUSION

This case of EEC calls for a heightened awareness of the possibility of EEC Syndrome in the general population. It would be helpful in early diagnosis and better management of such patients with the earlier diagnosis.

CONFLICT OF INTEREST

None declared till now.

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