ECTRODACTYLY-ECTODERMAL DYSPLASIA CLEFTING SYNDROME A RARE CASE REPORT

Jayakar Thomas¹*, R.G.Sharada², R.Sathya Narayanan³, Manoharan D⁴

Professor & Head¹, Junior Resident², Assistant Professor³, Professor⁴, Department of Dermatology, Sree Balaji Medical College & Bharath University, Chennai 600044, Tamilnadu, India.

Corresponding Author: - Dr. Jayakar Thomas
E-mail: jayakarthomas@gmail.com

**Article Info**

<table>
<thead>
<tr>
<th>Received 15/11/2015</th>
<th>Revised 27/11/2015</th>
<th>Accepted 15/12/2015</th>
</tr>
</thead>
</table>

**Key words:** Lobster claw deformity, EEC syndrome, Ectodermal dysplasia

**ABSTRACT**

Ectrodactyly-ectodermal dysplasia clefting syndrome (EEC syndrome) is a very rare form of ectodermal dysplasia transmitted as an autosomal dominant inheritance disorder characterized by ectodermal dysplasia, ectrodactyly, cleft lip/palate, lacrimal duct anomalies and abnormalities of the epidermal appendages including hypotrichosis, hypodontia, dystrophic nails and occasional hypohidrosis. We report a case of Ectrodactyly-ectodermal dysplasia clefting syndrome in 8 years old boy who presented with complaints of deformed hands and feet and sparse hair since birth.

**INTRODUCTION**

Ectrodactyly-ectodermal dysplasia clefting syndrome (synonym: split hand-split foot-ectodermal dysplasia-clefting syndrome) was first described by Cockayne (Cockayne, 1936) in 1936 and later Rudiger in1970 described as EEC syndrome (Rudiger et al., 1970) It is a rare genetic disorder transmitted as autosomal dominant inheritance but can be transmitted sporadically with an incidence of around 1 in 90,000 in general population (Iorio et al., 2012). Three clinical morphotypes, EEC-1, EEC-2, and EEC-3 exist. EEC-1 is transmitted as autosomal dominant inheritance and occurs due to mutation of the p63 gene (a homolog of tumor suppressor p53), located on chromosome 7q11.2–q21.3 (Fukushima et al., 1993; Qumsiyeh, 1992). p63 is responsible for genesis of the limbs and craniofacial regions in fetal life. Gene locations for EEC-2 and EEC-3 have been identified on chromosome 19 and 3q27 respectively. The main features of the EEC syndrome are ectrodactyly (split hand or foot deformity), cleft lip/palate and tear duct anomalies.

**CASE REPORT**

An 8 years old boy presented to our skin OPD with complaints of deformed hands and feet and sparse hair since birth. History from the mother revealed that he was born out of non-consanguineous marriage. Antenatal and postnatal period was uneventful. He was delivered by normal vaginal delivery and birth weight was 2.65kg. There was no developmental delay. He had cleft palate at birth, which was repaired at the age of 8 months. There was no history of difficulty in walking, hoarseness of voice, seizures, hearing or visual disturbances. Bowel and bladder habits were normal. There was no history of similar complaints in the family members.

Dermatological examination revealed ectrodactyly of hands [Figure 1] and feet with post inflammatory hypopigmentation present over the left foot [Figure 3]. Scalp hair was thin, sparse and brittle [Figure 2]. Eyebrows and eyelashes were thin and sparse. Teeth, nail, mucosa, palms and soles were normal. Sweating was normal. Systemic examination was normal. Slit-lamp examination and Schirmer test for lacrimal function were normal. Complete blood counts done were normal. X-rays findings of the hands and feet were corresponding with the clinical features.
DISCUSSION AND CONCLUSION

Ectrodactyly (lobster claw deformity) is a major feature of this disorder and occurs in over 90% of affected individuals. Tetramelic involvement is the most common (75%) and the third and fourth digits are most commonly involved. Cleft palate with or without cleft lip are present in 70%–100% of the cases. The typical facial appearance includes hypoplastic maxillae, short philtrum and broad nasal tip. Choanal atresia, oligodontia, anodontia, premature loss of secondary teeth and peg-shaped teeth may be present. The nails are hypoplastic and dystrophic. The scalp hair is fine and sparse, light-colored and may be wiry in texture. Eyebrows and eyelashes are short, thin and sparse. Axillary, pubic and body hair may also be affected. Atresia or hypoplasia of the lacrimal duct is seen in over 90% of affected individuals. Photophobia, corneal ulcers and corneal scarring and perforation, may occur as a result of lacrimal gland hypoplasia. Secretions from the lacrimal gland may be diminished. Dry, scaly skin and scalp dermatitis are common. Sweating is usually normal. Many other abnormalities may be associated. Some of these include conductive deafness, genitourinary malformations (e.g. glandular hypospadias, urethral reflux and hydronephrosis) (Roelink, 1988), hypothalamic-pituitary dysfunction (Gershoni-Baruch, 1977), mental retardation and hamartoma of the tongue (Hanna, 1994).

Differential diagnosis of EEC are odontotrichomic syndrome may be differentiated by severe tetramelic reductions and autosomal recessive mode of inheritance and aplasia cutis congenita with limb defects which does not have clefting or ectodermal defects other than absence of skin Others include Hay-Wells syndrome, Rapp-Hodgkin syndrome, ADULT syndrome, LADD syndrome, limb-mammary syndrome, Rosselli–Gulienetti syndrome, Martinez syndrome and Zlotogora–Ogur syndrome.

Diagnosis is by radiography of deformed hands and feet showing absence or hypoplasia of metacarpals and metatarsals. Treatment involves surgical correction of the limb defects, cleft lip/palate, lacrimal duct and genitourinary abnormalities. DNA-based prenatal diagnosis is available for selected families where the gene defect is known. This case is reported because of its rarity.
ACKNOWLEDGEMENT: None

CONFLICT OF INTEREST:
The authors declare that they have no conflict of interest.

STATEMENT OF HUMAN AND ANIMAL RIGHTS
All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

REFERENCES