

## Case Report

# Ectrodactyly: Rare Presentation in Pre-Term Twin Neonates

Dr. Bhavna Gupta

<sup>1</sup>Senior Resident, Department of Pediatrics, DMCH, Ludhiana, Punjab, India

### Article History

Received: 26.03.2022

Accepted: 25.04.2022

Published: 18.05.2021

### Journal homepage:

<https://inlightpublisher.com/journal/hjpah/home>

### Quick Response Code



**Abstract:** Ectrodactyly is congenital malformation of limb-extremities, also known as split hand-foot malformation (SHFM) or lobster-claw deformity, presenting as deep median clefts of hand and/or foot because of central rays deficiency. Condition may present as hypoplasia/aplasia of phalanges, metacarpals or metatarsals and/or syndactyly. It may occur as isolated non-syndromic malformation or may occur as a part of a syndrome. EEC is syndromic form of malformation in the form of combination of ectrodactyly, ectrodermal dysplasia and cleft lip/palate. Here is a case report of non-syndromic form of ectrodactyly in dizygotic twins born prematurely to a primigravida female with placenta previa and antepartum haemorrhage.

**Keywords:** Ectrodactyly, SHFM, hypoplasia.

**Copyright @ 2022:** This is an open-access article distributed under the terms of the Creative Commons Attribution license which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use (Noncommercial, or CC-BY-NC) provided the original author and source are credited.

## INTRODUCTION:

Ectrodactyly is the congenital absence of central rays of limbs<sup>1</sup>. There are defects in median apical ectodermal ridge activity leading to absence of middle digits of hand/foot and/or lobster-claw appearance because of presence of median clefts of hand/foot. Ectrodactyly is one of the rare conditions occurring as 1 in 90,000 births with males and females are equally affected<sup>2</sup>. It may occur as isolated non-syndromic malformation or may occur as a part of a syndrome. Cases can be sporadic or familial but sporadic form is reported to be more common<sup>3</sup>. In familial cases, autosomal dominant inheritance is reported as most common with autosomal recessive or X-linked inheritance is also reported<sup>4</sup>. Non-syndromic form of ectrodactyly may be associated with the defects of long bones such as tibial or fibular aplasia and known as split hand/foot malformation with long bone

deficiency<sup>5</sup>. Syndromic ectrodactyly may have associated defects of craniofacial, genitourinary and ectodermal structures<sup>5</sup>.

## CASE REPORT:

Twin babies born to 30 years old primigravida female at gestation of 33 weeks via emergency caesarian section in view of placenta previa with antepartum haemorrhage and pre-eclampsia with severe features, conceived naturally after non-consanguineous marriage as dizygotic, dichorionic -diamniotic twin pregnancy presented with ectrodactyly, detected at birth by paediatrician attending the delivery.

First twin, female baby with birth weight of 1.5 Kgs had bilateral split feet. Grossly on examination, there was absence of middle digits of both feet as shown in Figure 1.



On radiological assessment, as shown in Figure 1, X-ray bilateral feet suggestive of absence of phalanges (aplasia) of second, third and fourth digits associated with complete absence (aplasia) of bilateral metatarsals and partial absence (hypoplasia) of bilateral tarsals. First twin had bilateral normal hands.



Second twin, male baby with birth weight of 1.4 Kgs had malformation of both hands and feet. As shown in Figure 2, on gross examination, right hand had absence of thumb and index finger with syndactyly of middle and ring fingers.

Grossly, there was absence of middle digits with typical lobster-claw appearance of both feet. On radiological assessment, X-ray suggestive of aplasia of bones of phalanges of thumb and index finger of right hand associated with aplasia of phalanges of thumb of left hand. X-ray bilateral feet showing complete absence (aplasia) of phalanges of bilateral second, third and fourth digits associated with aplasia of bilateral metatarsals and hypoplasia of bilateral tarsals. There was no family history of ectrodactyly or other congenital malformations.

On examination of babies, there were no other features of dysmorphism including no visible craniofacial defects. Genitourinary and cardiac defects ruled out with normal ultrasonic examination and echocardiography respectively.

### **DISCUSSION:**

Ectrodactyly is a rare genetic disorder presenting as congenital malformations of limb-extremities in the form of hypoplasia or aplasia of central digits of hand and/or feet resulting in median clefts and lobster-claw deformity. In this case report, dizygotic twins presented with ectrodactyly with no other syndromic manifestations and no family history of same with less likely possibility of familial autosomal dominant inheritance. Presence of ectrodactyly warrants detailed systemic evaluation for associated anomalies along with orofacial defects including hearing and vision assessment. It results in significant functional impairment of involved limbs with surgical correction considered only if associated with transverse bone or syndactyly<sup>5</sup>. Detailed parental counselling about risk of recurrence of ectrodactyly in future pregnancies should

be considered and should be done in presence of both parents. Parents should be advised for genetic testing in presence of syndromic form along with timely antenatal ultrasounds in all cases.

### **Statement of Ethics:**

Parents of subjects have given their written informed consent to publish their case including publication of images.

**Conflict of Interest Statement:** Nil

**Funding Sources:** None

### **REFERENCES:**

1. Durowaye M, Adeboye M, Yahaya-Kongoila S, Adaje A, Adesiyun O, Ernest SK, et al. Familial ectrodactyly syndrome in a Nigerian child: A case report. *Oman Med J* 2011;26:275-8.
2. Jindal G, Parmar VR, Gupta VK. 2009. Ectrodactyly/split hand feet malformation. *Indian Journal of Human Genetics*, 15(3):140-142. doi:10.4103/0971-6866.60191.
3. Duijf PH, van Bokhoven H, Brunner HG. Pathogenesis of split-hand/ split-foot malformation. *Hum Mol Genet* 2003;12:R51-60.
4. Zlotogora J. 1994. On the inheritance of the split hand/split foot malformation. *Am J Med Genet.*;53:29-32.
5. Muhammed Basheer K T., Sunil Kumar Agarwalla., Pulak Ranjan Mallik and Subhrajit Parida, 2016. "Ectrodactyly: The 'Lobster Claw' Anomaly – Rare Report of an Indian Family Demonstrating Autosomal Dominant Inheritance Pattern", *International Journal of Current Research*, 8, (11), 41192-41194.