

## Case Report

# MEDICAL MYSTERY: ECTRODACTYLY A CASE REPORT

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## ABSTRACT

Congenital absence of all or part of one or more fingers or toes is defined as Ectrodactyly. It is the word derived from Greek *ektroma* meaning "abortion" and *dactyly* meaning digits. Also called split hand or foot malformation (SHFM), Ectrodactyly is a rare limb malformation with median cleft of hand and foot and aplasia or hypoplasia of the phalanges, metacarpals and metatarsals. Irregular pattern of inheritance exists for ectrodactyly; may be autosomal dominant, autosomal recessive or X linked. In this condition, the median cleft of the hand gives the hand, the appearance of lobster claws- so also called 'Lobster Claw Hand". Ectrodactyly may be very commonly associated with other anomalies of face, eyes and skeletal system. We report a rare case of unilateral ectrodactyly of right hand .The present case is not associated with any other anomaly as described in various textbooks and reference articles. The details of etiology, diagnosis, genetic causes, association with other anomalies and treatment will be discussed.

**KEY WORDS:** Ectrodactyly, Split hand or foot malformations, Median Cleft, Autosomal dominant, Autosomal recessive.

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## INTRODUCTION

**Ectrodactyly**, a more recognisable nick name - "**Lobster Claw Hand**" [1] is a rare limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of hands and feet and aplasia or hypoplasia of the phalanges, metacarpals or metatarsals [1]. The absence of central digital rays gives rise to the median cleft, resulting in the lobster claw appearance of hands or feet. Ectrodactyly has also been known as split hand or foot malformation (SHFM), or Karsch Neugebauer syndrome [2], or EEC (Ectrodactyly- Ectodermal Dysplasia-Clefting) syndrome [3]. First case of

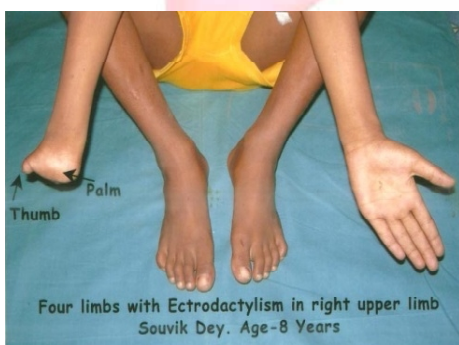
Ectrodactyly was described in 1936. It affects about 1 in 90,000 babies with no sex predilection. Prenatal diagnosis<sup>4</sup> of ectrodactyly was first reported in 1980 and most of the patients lived normal life with very little impairment of limbs.

## CASE REPORT

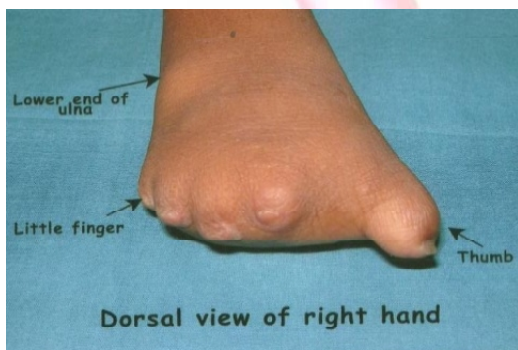
A Hindu boy named Souvik De, aged eight years, resident of Bankura district (village-Khilkalanali), West Bengal, India presented at the Physical Medicine and Rehabilitation OPD of Bankura Sammillani Medical College, with features of ectrodactyly in right hand, in the month of August, 2012. He presented with the following features in the right hand.

1. Rudimentary digits- Five in number, having nails in all digits. Thumb is having the largest well formed nail. In rest of the four digits, rudimentary hard small nails are present.
2. Palm is small, full, and globular in shape, appears like a soft pad. Size of right palm is about half the size of left palm.
3. Thenar and hypothenar eminences can't be demarcated with a small depression in between them.
4. Palmar crease is absent in the right palm.
5. Crease of first metacarpophalangeal joint is present.
6. Girth of right upper limb up to the wrist is normal.
7. Movement of right shoulder joint and elbow joint is normal. But movement of right wrist joint is slightly restricted.

**Fig. 1:** Showing all the four limbs.



**Fig. 2:** Dorsal view of right upper limb only.



**Fig. 3: Radiology-** Skiagram of both hands with wrist joints (AP view) shows:



1. Total absence of bony structures distal to carpal bones on the right side.
2. Seven ossification centres of carpal bones on the left side and six on the right side, suggests the age of the subject is between six to seven years. (According to father, presenting age of the subject was eight years.)
3. All the bones of left upper limb are perfectly developed, at par with age, showing no abnormality.

Though the boy presented the abnormality congenitally, but the affected limb was growing comparatively slowly along with age. No signs of inflammation were present over the affected limb and the boy used his left hand for doing his day to day activities with casual efficiency.

There were no other dysmorphic features and anthropometry was within normal limits. Physical and systemic examination was normal. The child was a student of second standard, average in studies and normal in development for his age. He was product of a nonconsanguinous marriage and full term caesarean delivery with no significant perinatal events. Our case was second in birth order. The first sibling was a female baby delivered by caesarean section (full term) with the same deformity in the left arm. She died at the age of two months due to an acute attack of meningitis. In both siblings caesarean section was done due to the presence of oligohydramnios. Besides ectrodactyly, the parents didn't notice any other abnormal feature in the case mentioned. There was no history of similar clinical profile in any of the relatives of both the parents. Both the parents were not suffering from any chronic illness (like Diabetes, Hypertension or Hypothyroidism), nor they gave any significant history regarding addiction or drug intake. Both were non smokers. Though Bankura is a leprosy prone zone, there was no history of leprosy in the family

## DISCUSSION

Embryogenesis of the upper extremity starts with formation of the upper limb bud, on the lateral wall of the embryo four weeks after fertilization. Eight weeks after fertilization, embryogenesis is complete and all limb structures are present. The majority of congenital anomalies of the

upper extremity occur during this period of rapid limb development [5]. Through research, it was elicited that there is definite role of HOX gene in the development of the limb buds. Nevertheless, it is thought that the T-box genes Tbx5 and Tbx4 [6] might be involved in determining limb identity.

According to the internationally acclaimed classification system of Swanson [7]- Congenital anomalies are divided into seven types:

- a) Failure of formation (transverse or longitudinal)
- b) Failure of differentiation
- c) Duplication
- d) Overgrowth
- e) Undergrowth
- f) Constriction bands
- g) Generalized skeletal abnormalities.

In the present case, probably failure of formation and failure of differentiation were responsible for the anomaly.

Programmed cell death (Apoptosis) is responsible for the tissue break down in the interdigital region and it is probably mediated by Bone morphogenic protein (BMP) signalling molecules of the TGF (Superfamily) [8]. Blocking of the cellular and molecular events may also account for the failure of separation and demarcation of foetal digits.

Ectrodactyly, most properly labelled as split hand or foot malformation (SHFM) can be divided into two broad groups. 1) SHFM with isolated limb involvement-Non Syndromic form 2) SHFM with associated anomalies like tibial aplasia, mental retardation, ectodermal and craniofacial findings like- orofacial clefting and deafness—Syndromic form. Our case belongs to the non syndromic form of SHFM, as there is no associated anomaly.

Five different genetic mutations are known to be associated with SHFM (Syndromic as well as Non syndromic) [9]. Type 1) is the most frequent variety; due to a mutation on chromosome 7 in a region that contains two homeobox genes, DLX5 and DLX6. The anomaly of ectrodactyly develops secondary to one of the chromosomal mutations which results in failure of AER (Apical Epidermal Ridge) to produce molecules that signals nearby cells to differentiate into digital rays. So far the

only mutations known to underline SHFM in humans have been found in TP63 gene [10].

Familial Ectrodactyly are sporadic in nature in family. Familial forms usually are inherited as autosomal dominant forms. Syndromic ectrodactyly has a variable degree of expression. Non syndromic SHFM limited to the hand and feet usually follows the pattern of inheritance of a regular autosomal dominant gene with a high penetrance [11]. However in our case, the probable inheritance pattern is autosomal recessive as only siblings and no other family members are affected.

### TREATMENT

Ectrodactyly can be treated surgically in order to improve function and appearance. Prosthetics may also be used. Parents should be counselled regarding the possibility of recurrence of the disease in the future siblings and antenatal diagnosis by ultrasonography should be offered.

**Conflicts of Interests: None**

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