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Limb Anomalies

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Abstract: *Limb anomalies form 6/10,000 live births.*
Polydactylism: *occurs in 1 per 2,000 births. According to Barsky,*
Syndactylism: *is the most common congenital deformity of the hand. He quoted an estimated occurrence of 1 in 3,000 births and said that it is most frequent between the middle and ring finger. Causative factors are hereditary, teratogens and combination of this with environmental factors. The most susceptible period is 4th – 5th week of intrauterine life. Eight cases of limb anomalies that we have come across have been enumerated along with the x-ray findings wherever possible. The types of anomalies that we came across were polydactyly, synpolydactyly & lobster claw deformity. Though we have not done any genetic study in these cases, the fact that the previous and present generation, siblings and their children are not having these defects rules out the possibility of any genetic defect; some teratogen along with environmental factor may be the causative factor for the defects in these cases.*
Keywords: *Polydactyly, Synpolydactyly, Cleft hand, Cleft foot*.

I. Introduction

Limb anomalies form 6/10,000 live births. 3.4/10,000 involves upperlimb, 2.6 /10,000 involves L.L. Often associated with – cranio facial, cardiac & genitourinary abnormalities. Polydactylism is usually reduplication of the little finger or of the thumb, rather than of any of the intervening fingers and the condition is said to be more common on the radial side of the thumb. It occurs in 1 per 2,000 birth cell death (apoptosis) fails extra digits or toes -lacks proper muscle connection usually bilateral. According to Barsky, Syndactylism is the most common congenital deformity of the hand. He quoted an estimated occurrence of 1 in 3,000 births and said that it is most frequent between the middle and ring finger.

Causative factors

1. **Mainly hereditary:** - Gene mutations

- Mutation in HOXA13 result in hand foot Genital syndrome – presents fusion of the carpal bones and short digits affected female has various forms of duplication of uterus and affected males may have hypospadias
- HOXD13 results in Synpolydactyly
- TBX5 mutation (chromosome 12q 24.1) - Holt-Oram syndrome – upper limb anomalies associated with heart defects.
- COLIAI or COLIA2 gene mutation (dominant) will lead to Osteogenesis Imperfecta.
- FBN1 gene mutation causes Marfan syndrome.

2. **Teratogen**:- induced limb defects are common

- Drug thalidomide causes limb anomalies associated with intestinal atresia & cardiac anomalies

3. **Teratogen & Environment:-**

- Playing a role together as observed in the present cases.
- Amniotic bands may cause ring constrictions and amputations of the limb or digits.

Development

- Most susceptable period is 4th to 5th week of IUL. Around 4th wk of IUL –upper limb bud followed by1 or 2 days later – lower limb bud appears on the ventro lateral body wall. Limb bud mesenchyme is derived from lateral plate mesoderm. Factors secreted by it initiates limb outgrowth. In the 6th week of IUL – terminal part of the bud flattens to form hand & foot plates. First one constriction followed by another constriction occurs in the developing limb. By 6th week hyaline cartilaginous model of the bones occur. In the 7th week of gestation limbs rotate in opposite direction takes place by 12th week ossification centre to shaft appears.

Molecular regulation of limb development

- Positioning of the limbs along the cranio-caudal axis in the flank region is regulated by HOX genes - for upper limb by TBX5 & FGF10; for lower limb by TBX4 & FGF10.
- Limb bud is covered by a layer of cuboidal ectoderm that forms a thickening called apical ectodermal ridge (AER). BMPS (bone morphogenic proteins) expressed in ventral ectoderm induce the formation of AER by signaling through the home box gene MSX2. After the ridge is established it expresses FGF4 and FGF8 which maintain the progress zone and growth occurs in a proximo-distal direction by FGFS.
- A cluster of cells at the posterior border of the limb express Sonic hedgehog (SHH) a secreted factor that regulates the antero-posterior axis, regulated by the zone of polarizing activity (ZPA). These cells produce retinoic acid (Vit A).
- All of the **patterning genes** in the limb have feedback loops. Thus FGF in the AER activate SHH in the ZPA, while WN17a maintains SHH signal. Hence patterning genes maintain limb axis.
- Fingers and toes are formed by cell death in AER. Further formations of digits depend on the AER at the tip. Under the influence of AER, cells distal from the AER differentiate into cartilage & muscle.
- Thus HOX genes expression which results from the combinatorial expression of SHH, FGFS and WN17 a occurs in phases in three places in the limb that correspond to the formation of the proximal, middle and distal parts.
- Hox genes regulate the types & shape of the bones of the limb. HOXA and HOXD clusters are the primary determinants in the limb and variation in their combinatorial patterns of expression may account for differences in forelimb and hind limb structures.

Different types of anomalies are:

- **LIMBS** -Partial limb – Meromelia, Complete absence – Amelia, & Some part of the long bones missing - phocomelia (a form of meromelia)
- **DIGITS** - Short digits - brachy dactyly; 2 or more fingers or toes are fused – syndactyly; Extra digits or toes - polydactyly; the combination of the two is the polysyndactyly. Absence of a digit is called ectro dactyly (such as thumb) usually unilateral.
- Cleft hand & foot - Lobster claw deformity - abnormal cleft between 2nd & 4^{th} metacarpal bone

II. Observation

The type of anomalies that we came across was Polydactyly, Synpolydactyly & lobster claw deformity. Eight cases of limb anomalies that we have come across had been enumerated along with the x-ray findings wherever possible.

- **Case-1 -** A 30 year old male had an extra digit in the left foot on the big toe side. The proximal phalanx of the extra digit was also articulating with the 1st metacarpal bone.
- **Case-2 -** A 5 year old boy had extra digit in both the hands.
- **Case-3 -** 55 year old female had extra digit in both the hands.
- **Case -4** - A 27 year old male who had an extra digit in both hands.
- **Case-5-** 48 year old female had extra digit in both the hands and feet.
- **Case-6 -** 30 year old female had extra digit in both the hands and feet.
- **Case-7** - In a boy of 15 years old Synpolydactyly was present on the radial side of the right thumb. Metacarpal and proximal phalanges and skin were fused. Terminal phalanges were separate with intact skin. The present case belongs to Synpolydactyly type ll (where extra digit may have fused, forked showing complete or partial duplications of digit) Incidence is uncertain
- **Case-8 -** 60 years old female reported for SHSF-split hand split deformity or Lobster claw limb deformity.

III. Discussion

- In all these cases no other siblings or parents had these defects. Three of the cases were from a same area in a village close to Eluru, A.P.
- Though we have not done any genetic study in these cases the fact that the previous and present generation, siblings and their children are not having these defects rules out the possibility of any genetic defect; some teratogen along with some environmental factor may be the causative factor for these defects in these cases

Inference

- All these persons must have had mental trauma but however adopted themselves for normal lifestyle and day to day activities.

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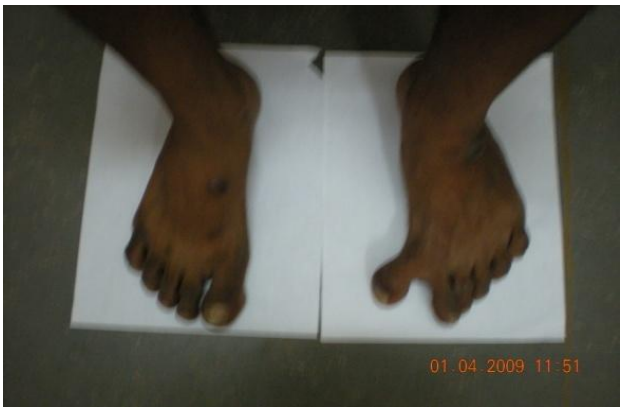
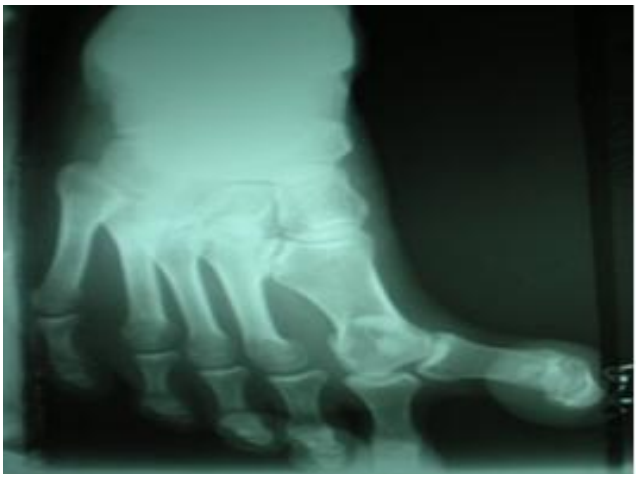
Case - 1

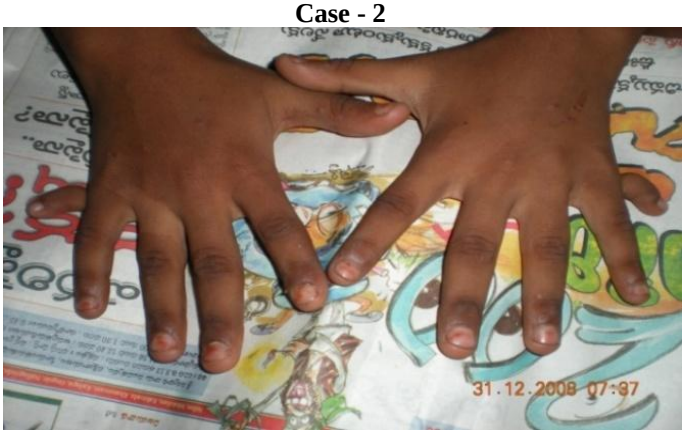


In the 30 year old male extra digit was the big toe side of the



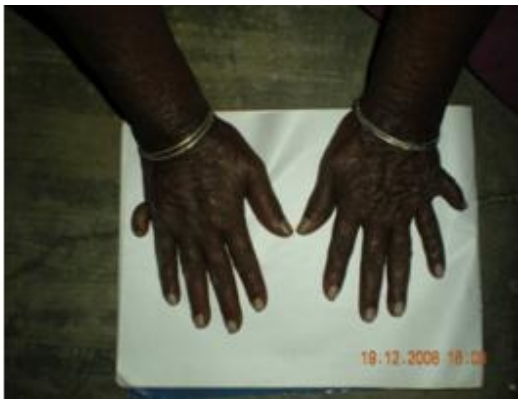
The proximal phalanx of the extra digit was also present on articulating with the 1st metacarpal bone.

Case - 2



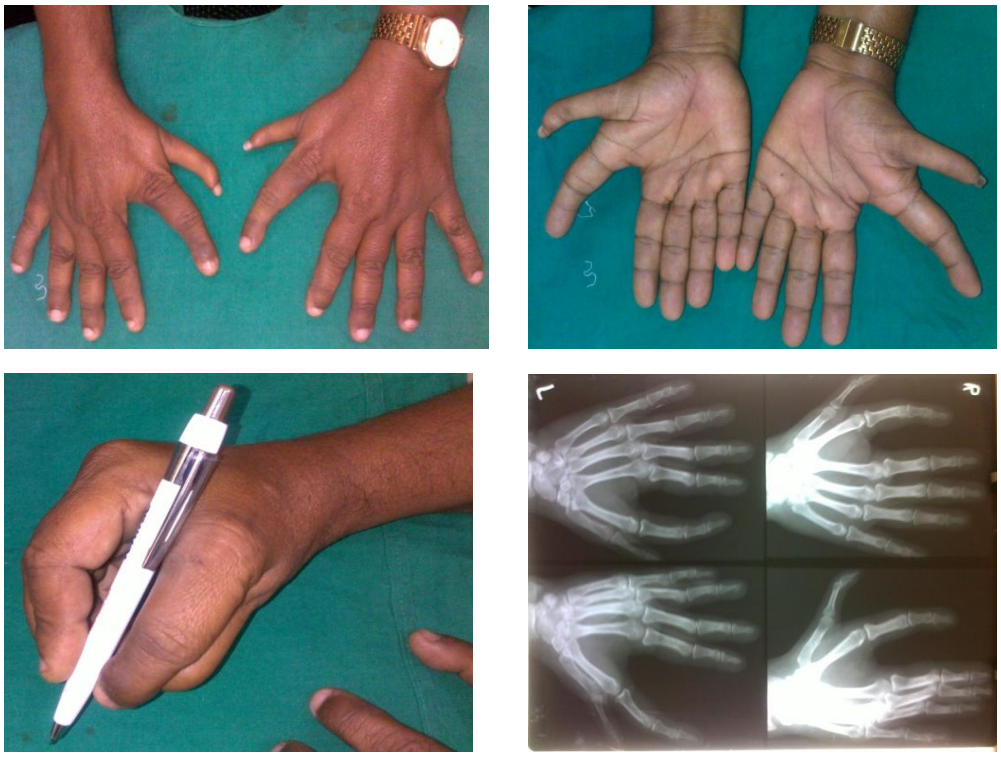
A 5 year old boy had extra digit in both the hands

Case – 3



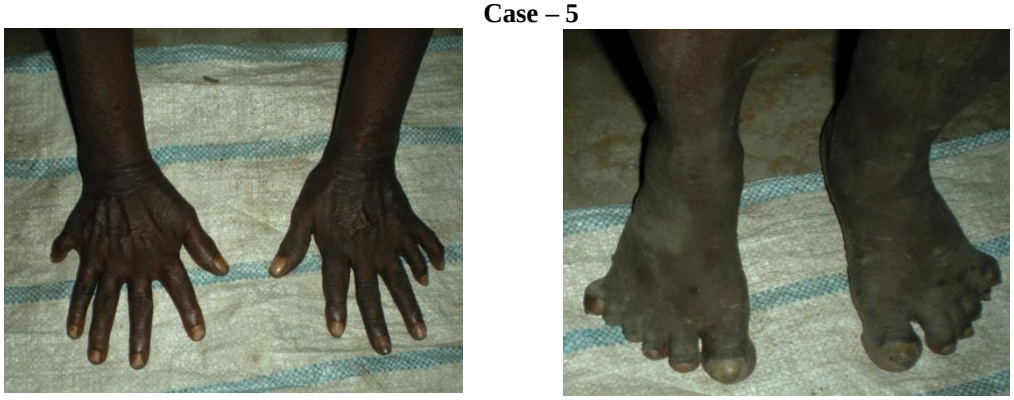
A 55 year old lady who had an extra digit in both hands

Case - 4



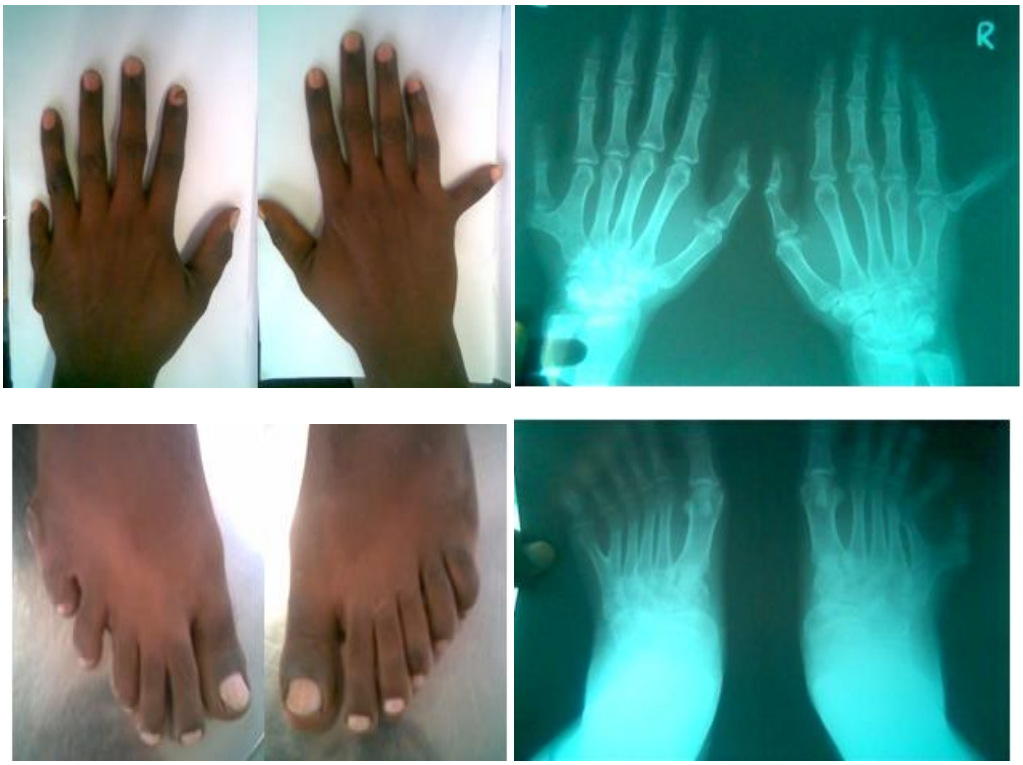
A 27 year old male who had an extra digit in both hands

Case – 5



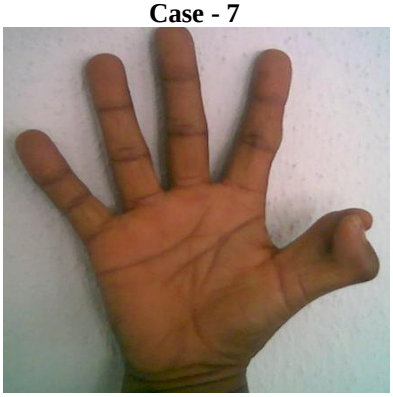
A 48 year old lady who had an extra digit in both hands & feet

Case – 6



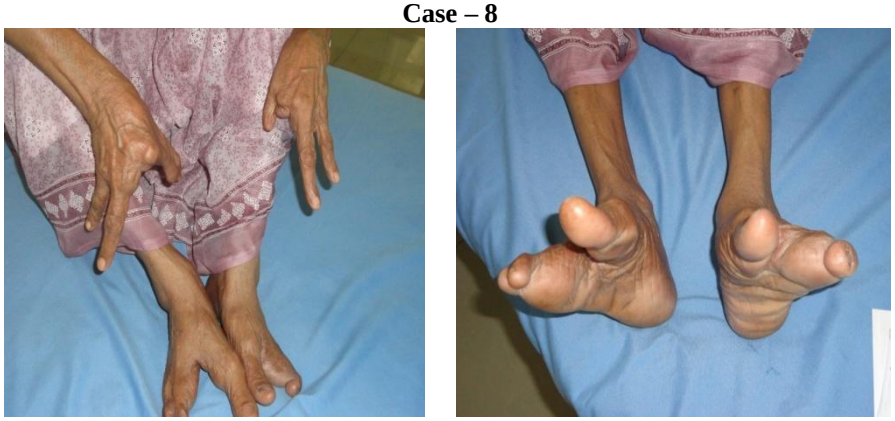
30 year old female had extra digit in both the hands and feet

Case - 7



In a boy of 15 years old Synpolydactyly was present on the radial side of the right thumb. Metacarpal and proximal phalanges and skin were fused. Terminal phalanges were separate with intact skin

Case – 8



60years Mrs. Muthammal reported for SHSF- split hand split deformity or Lobster claw limb deformity