# **Original Research Paper**



## **Anatomy**

#### CASE STUDY IN POLYDACTYLY

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A congenital anomaly characterised by supernumerary digits on the limbs during developmental stage is called ABSTRACT ] polydactyly. The occurrence of polydactyly varies among different populations. It is manifested either as an isolated condition or as an associated congenital condition. We describe two cases where a 30 year old male individual was noted that the left foot comprised of one extra digit on the big toe and another case where a five year old boy had one extra digit on both of his hands on the ulnar side. The pedigree and family history of both the cases did not give any clue towards the heritability of the anomaly.

**KEYWORDS**: polydactyly, extra digits, congenital anomaly.

#### Introduction:

Polydactyly is a congenital abnormality occurring during developmental stage and is characterised by supernumerary digits either on the hand or foot [1]. The extra digits usually comprise of soft tissue or bone without joints. The occurrence of congenital anomalies in neonatal is 2% and 10% of these deformaties are of the upper extremity [2]. Polydactyly belongs to the category of duplication [3] as an association is present between polydactyly and many syndromes. It is appropriate to consult a geneticist if the polydactyly condition is in the family for more than two or three generations [4]. Majority of the congenital anomalies occur during the fourth week of embryonic period of rapid limb development [3]. Studies have shown that polydactyly is associated with 39 genetic mutations [5]. This anomaly can be divided into three major types 1. ulnar or post axial polydactyly where the extra digit is on the ulnar side of the hand( side of the little finger). This type of polydactyly is hereditary. 2. Radial or preaxial polydactyly where the anomaly is on the side of the hand towards the thumb (radial side of the hand). This condition is very often associated with several syndromes. 3. Central polydactyly where the extra digit is on the ring, middle or index finger and it is a rare condition [6].

This congenital developmental anomaly is associated with either mutation in a gene itself or in a cis-regulatory element responsible for the expression of a specific gene. Studies revealed that mutations in HOX A or HOX D clustered lead to polydactyly [7]. Polydactyly can occur by itself or as one feature of a syndrome of congenital anomalies. When it occurs by itself it is associated with autosomal dominant mutations in a single gene [8]. 97 genetic syndromes were found to be associated with different kinds of polydactyly [4]. Classification of the polydactyly condition is performed by using X-ray imaging to see the bone structures[9]. Many of the polydactyly anomalies do not interfere with the functioning of the limbs, but for social instinct they can be treated surgically [4].

The present study was carried out with two cases belonging to West Godavari district of Andhra Pradesh, India. Pedigree analysis and detailed family history was taken from both the cases. Photography and radio diagnosis were the means by which the anomalies were established.

Case I: On phenotypical examination of the foot of a 30 year old male individual, it was noted that both the feet comprised of one extra digit on the lateral side. The foot along with toes were well formed. Radiological examination revealed that the 5th metatarsal bone was found dividing into two and the proximal phalanx of the extra digit was articulating with its metatarsal bone like in other toes. Radiological examination also revealed that the division of 5<sup>th</sup> metatarsal was wider on the right side leading to shorter 6th digit.



Case II: Physical examination in a five year old boy revealed that one extra digit was present on both the hands in the central region. X-ray imaging showed that the 3<sup>rd</sup> metacarpal was bifid and an extra carpal bone was formed on either hand. In both these cases no other abnormalities were found





**Discussion:** Polydactyly condition arises with other abnormalities. The anomaly can be identified by ultrasonography during 14-16 weeks of gestation. It is very important to identify this anomaly in earlier stage so that accurate treatment can be rendered. Excision of the extra digits in the neonates is a safe and simple procedure with good clinical outcome [9]. Ulnar polydactyly is also a part of a syndrome.[10] Comparative studies with different ethnicities revealed that patients with African origin are more often affected with isolated polydactyly condition while in Caucasians it is mostly associated with a syndrome.[3]. In the present cases there was no association of a syndrome. Polydactyly was said to be inherited as a dominant trait. The pedigree and family history of both these cases did not give any clue towards the heritability of the anomaly.

Conclusion: Generally the surgical elimination for preaxial duplication gives successful functional outcome. Surgery should not be delayed much beyond the walking age so that it gives ample time for the bones to remodel. However surgery can be performed at any age with good result. In the initial stage, management of polydactyly of foot appears to be simple, but the multiformity of its configuration deserves careful consideration during surgical correction. The surgery of the polydactyly condition of foot must lead to proper alignment of the toes. Familial transmission of the anomaly must also be kept in mind while the patient is approaching for the treatment.

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