Rare case of tetra polydactyly with different skeletal anatomy in each limb

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Abstract: *Polydactyly is a common congenital physical anomaly affecting the hand and foot where there are more than usual numbers of digits. It may present as a range of defects from minor soft tissue abnormalities to major bony abnormalities. Tetra polydactyly is a rare presentation involving all the four limbs. Here we are presenting a case of a new born baby with tetra polydactyly with different bony configuration in each limb with different functional movements. The overall general health of baby was good and there was no syndromic association. Physical examination and radiographs explain the anatomical abnormalities in detail, which is essential for further planning and management.*

Keywords: Tetra polydactyly, Digits, Congenital, Multifactorial, Genetics

Introduction

Polydactyly is a congenital variation in number of digits of hands and foot. In other words, polydactyl means an extra digit i.e. more than five digits in a limb. When the extra digit is present in all the four limbs, it is known as tetra polydactyly. The extra digits can be a cutaneous soft tissue projection, various bony abnormalities or it can be a fully formed digit. The incidence of tetra polydactyly is very rare [1]. Polydactyly can be present as an isolated skeletal deformity or can be present with other congenital anomalies of body. The various syndromic association of polydactyly are down syndrome, patau syndrome, meckel syndrome, ellis van crevald syndrome and bardet-biedl syndrome etc. [2- 4]. The etiopathogenesis of polydactyl is multifactorial and is generally remains unclear. It can also be inherited as autosomal dominant, autosomal recessive, X-linked, sporadic or familial.

Polydactyly is classified into pre-axial, post-axial and central polydactyly based on origin and location of extra digit [5]. Postaxial polydactyly is the most common polydactyl [6]. Antenatal history, family history with clinical examination and investigations helps to rule out the etiology, other congenital anomalies and the type of polydactyly. Surgery is generally done at around 1 year of age when the skeletal development or ossification has occurred in the affected area and is indicated for cosmetic purpose, pain relief and shoe fitting [7].

Case report

A 36 week late preterm baby girl delivered vaginally with birth weight of 2.2 kg which is appropriate for her gestational age born as a result of nonconsanguineous marriage at tertiary care teaching hospital. The baby was found to have six fingers in all four limbs. The extra digits were well formed except the fifth digit of right foot which was small, at a lower level and seems to be rudimentary (Figure 2). All the digits of right hand had active and passive flexion and extension movement, while there was no active flexion

movement observed at sixth digit of left hand although passive flexion and extension movement were observed (Figure 1). The fifth digit of right foot which is small, at a lower level compared to other digits of same foot and seems to be rudimentary and the range of passive movement were normal but active movements were not so much appreciable. While the sixth digit of left foot which is directed slight laterally had only passive movements and no active movement was appreciable (Figure 2).

Her dermatoglyphics pattern was normal. There was no facial dysmorphism, genital abnormality and other gross congenital abnormality. Her three generation pedigree analysis showed that this was a first born child with no history of abortion, no family history of polydactyly. Antenatal history of mother was uneventful and there was no history of any drug intake.

Her radiograph of limbs showed:

Right hand: 5^{th} metacarpal is short and incompletely formed and proximal phalanx of 5^{th} finger has been shifted proximally but all three phalanges were present suggesting central polydactyly (Figure 3).

Left hand: 6^{th} metacarpal along with its phalanges are present on ulnar side suggesting postaxial polydactyly (Figure 3).

Right foot: 5^{th} metatarsal base is originating from the base of 6^{th} metatarsal in a partial Y pattern. Also ossification centre of middle and distal phalanges of 5^{th} foot finger seems to be absent suggesting of central polydactyly (Figure 4).

Left foot: 6^{th} metatarsal was absent and only the proximal phalanx of 6^{th} foot finger was seen which was originating independently suggesting post axial polydactyly (Figure 4).

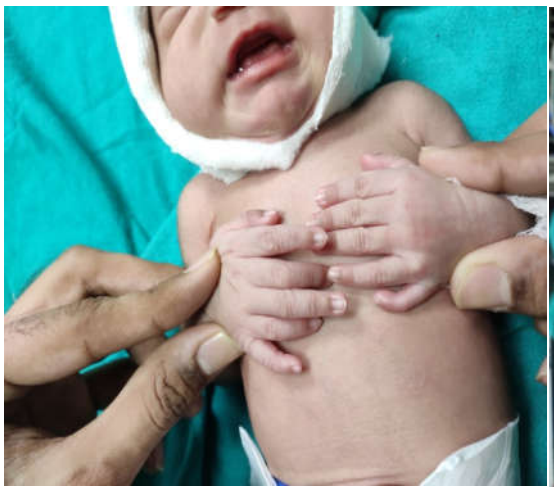

Figure: 1

Figure: 2

Figure 1 and 2 showing polydactyly in bilateral hand and foot.

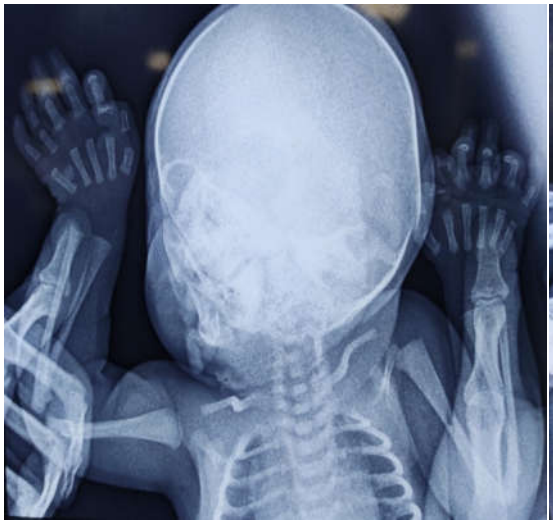

Figure: 3

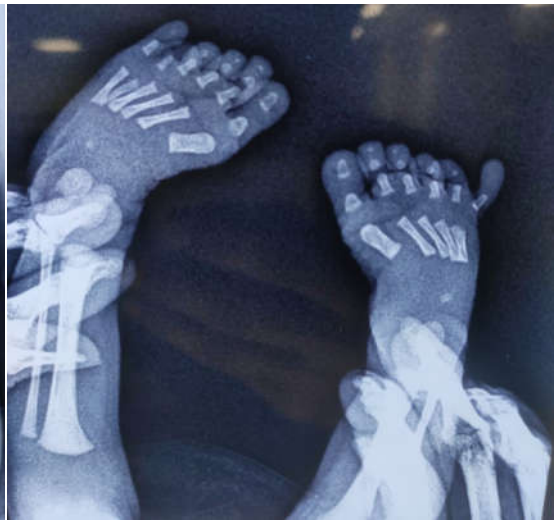

Figure: 4

Figure 3 and 4 showing radiographs of bilateral hand and foot.

Her echocardiography and ultrasonography of abdomen, skull and spine did not reveal any abnormality. Her biochemical tests including hormonal studies were within normal limits. Genetic testing could not be tested due to affordability issue. As it was new born baby, so attendants were advised regular follow up to assess for any functional limitation and further plan of management.

Discussion

Polydactyly is a congenital variation in number of digits of hands and foot. When the extra digit is present in all the four limbs, it is known as tetra polydactyly. Polydactyly can be present as an isolated skeletal deformity or can be present with other congenital anomalies of body the incidence of polydactyl is 1 in approximately 500 live births. Whereas tetra polydactyly has a very rare incidence[1]. The etiopathogenesis of polydactyl is multifactorial. Previously it was thought as a failure of programmed cell death involving fetal limb development [8]. But the current theories have a genetic basis involving specific locus mutation like 7p15-q11.23, 19p 13.1-13.2 which have a high penetrance [9,10]. The inheritance pattern of polydactyly can be autosomal dominant (in African black and African American), autosomal recessive (in white population), X-linked or sporadic familial [11]. In our patient, there was no specific pattern of inheritance as analysed by three generation pedigree analysis. In various previous

reports, polydactyly had been found to be associated with down syndrome, patau syndrome, meckel syndrome, ellis van crevald syndrome and bardet-biedl syndrome etc. [2- 4].

Polydactyly is classified into pre-axial, post-axial and central polydactyly based on origin and location of extra digit. In preaxial polydactyly, there is duplication of the first digit or radial side involvement is there. In post axial polydactyl, there is duplication of fifth digit or ulnar side involvement is there whereas in central polydactyly the duplication can be at second, third or fourth digit. Similarly, tibial, fibular and central polydactyly is used to describe lower extremity polydactyly. In crossed polydactyly, there is preaxial polydactyly in one limb and postaxial polydactyly in opposite limb [5]. Postaxial polydactyly is the most common polydactyly [6]. In our case, there was postaxial polydactyly in one limb and central polydactyly in opposite limb, the similar pattern was seen in upper as well as lower limbs which is very rare presentation for this type of polydactyly.

Antenatal history, family history with clinical examination and investigation helps to rule out the etiology, other congenital anomalies association and the type of polydactyly. Radiographs of the involved limbs are essential for planning of surgery. Polydactyly does not always need surgery, however surgical correction is indicated for conditions like cosmetic purpose, pain relief and shoe fitting. Surgery is generally done at around 1 year of age when the skeletal development or ossification has occurred in the affected area so that accurate anatomic assessment is possible before the surgery [7]. However, the skeletal anatomy is important is deciding the surgical strategy. A case like this, where each limb has a different surgical anatomy, will needs a different management plan for each limb, and hence surgical correction might be challenging. In our patient surgery was defered for a later age as the accurate anatomical and functional assessment couldn't be performed at this stage and the surgical correction may be challenging.

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Conflict of interests: Authors declares that there is no conflict of interest

Patient consent: Informed written consent was taken from father of neonate.

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