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Case Report

Polydactyly presentations: A case report

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ABSTRACT

Polydactyly is a duplication of the digits of the hands or feet. It is a congenital anomaly and can manifest as a single disorder or as a component of a syndrome. It is typically an autosomal dominant inherited condition with variable expression and incomplete penetrance. It is genetically heterogenic being caused by mutations in different genes. It manifests in different forms, based on the position of occurrence of the extra digit. It can occur as a single disorder as a component of a syndrome. Its presentation ranges from just a skin tag to a complete digit with the accompanying metatarsal or metacarpal bone. It can result in functional impairment or it may just be a cosmetically unacceptable digit with no functional impairment. Treatment depends on the nature of the extra digit as well as any functional impairment it may be causing. Surgical excision is employed and various reconstruction procedures are also employed in treatment, and must be well planned out to avoid a worsening of function. We present two cases; a 3-month-old baby girl presenting with a hereditary preaxial polydactyly (PPD) (Wassel type V) of both feet and a 29-year-old man with isolated postaxial polydactyly of the right foot with which was symptomatic.

Keywords: Polydactyly, Hereditary, Postaxial, Preaxial, Excision

INTRODUCTION

Polydactyly is a congenital anomaly characterized by multiplication of the digits of the hands and feet. It manifests as; a single disorder, multiple disorder or as a component of a syndrome.[1-3] It is typically inherited as an autosomal dominant disorder with variable expression and incomplete penetrance.[4] The genetic mutations occur in different genes thus it is regarded as being genetically heterogenic.[5-7] It is a disorder that has also been found to occur sporadically without any familial trait or any other congenital abnormality.[8] It has an incidence of 0.3–3.6/1000 live births. Noticed however to be commoner in people of African descent.[4,9] The extra digit can present as a skin tag or as a complete digit with accompanying metacarpal or metatarsal bone.[10] In some cases there is an extra digit present on both hands and feet (Polydactyly 24).[11] Temtamy and McKusick classification is the most widely used classification system employed to characterize polydactyly and it is broadly classified into Preaxial Polydactyly PPD, postaxial polydactyly PAP, and complex polydactyly all with various sub classifications.[4] Patients most times present in the neonatal period or in the 1st few months after birth. Some have been seen to present to the clinician later on in life or even in adulthood. Complaints commonly encountered include cosmetically abnormal appearance, pain, and functional impairment of the hand or foot affected. Some patients may have psychological problems from the cosmetically different appearance of the limbs if not excised early in life.[12] Treatment is usually excision of the extra digit. Care must be taken to clearly delineate the type of polydactyly and the accompanying tendon and ligament

attachments as well as blood supply to avoid complications that may further impair functionality of the hand following surgery.[13] Complications typically occurring include stiffness, angulation, growth arrests, and nail deformities. We present two cases of polydactyly with variable presentation; one in infancy and the other as an adult.

CASE REPORT

Case 1

A mother presented with her 2-month-old baby with complaints of a duplication of the big toes of both feet. These abnormalities were noticed at birth. No other abnormalities were noticed by the mother. The baby, the second child of the parents was the product of a full-term monitored pregnancy. There was no maternal illness during pregnancy and no use of medication during pregnancy except for routine antenatal drugs. No exposure to radiation was identified. Delivery was by spontaneous vaginal delivery. The elder sibling did not have any abnormality. The mother had similar duplication of the toes at birth for which she had excision in infancy. No other family relative was known to have a similar deformity. Examination of the baby revealed complete duplication of both big toes. No other congenital abnormality was identified. A radiograph of both feet revealed preaxial polydactyly with a bifid metatarsal bone in both feet Figure 1. (Wassel V). The parents were counseled on the condition and the baby had an excision of both extra digits Figure 2. The mother was noticed to have small big toes relative to other toes with scars over the metatarsal head medially on both feet Figure 3. Excision was carried out through the metacarpophalangeal joint of the extra digit with the preservation of the lateral soft-tissue structures adjoining the native big toe.

Case 2

A 29-year-old man presented to the clinic with complaints of an extra digit on the right foot (duplication of the small toe) which was painful with an increasing severity of the pain in the foot around the extra digit in the preceding few months and a difficulty in wearing shoes due to pain Figure 4. He did not have any extra digits or any other abnormality. The extra digit was said to have been present since birth but had not been symptomatic initially except for intermittent pain which was treated with analgesics previously. The extra digit was initially small but grew during adolescence. When examined he was noticed to have an extra digit over the small toe of the right foot at the lateral border. No other abnormalities were noticed in the other foot or other body parts. X-ray revealed a widening of the head of the fifth metatarsal bone with a duplication of the proximal phalanx Figure 5. Due to the persistent pain, deformity and difficulty wearing covered shoes, he was worked up for and had excision of the extra digit. Pain subsided and he was able to be well shod after surgery Figure 6.

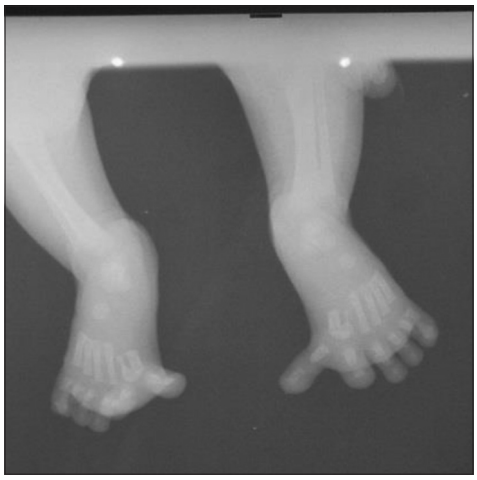

Figure 1: X rays of both feet of baby showing a bifid first metatarsal in both feet (Wassel V).

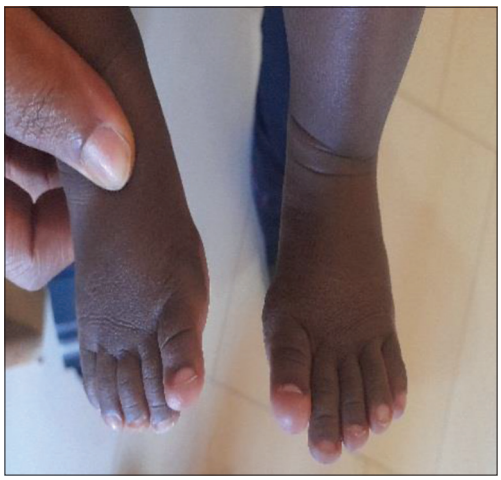

Figure 2: Post operative Clinical Picture of baby's feet.

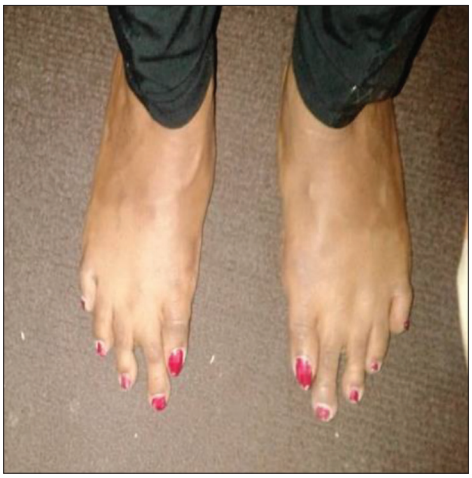

Figure 3: Clinical Picture of Mother's feet with small big toes, following excision of polydacyty in infancy.

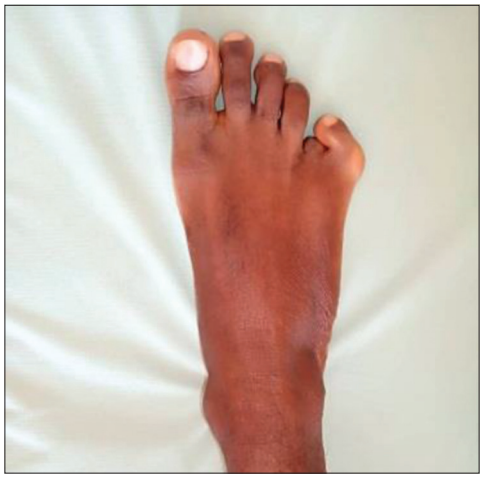

Figure 4: Clinical picture of 29year old man's foot with polydactyly.

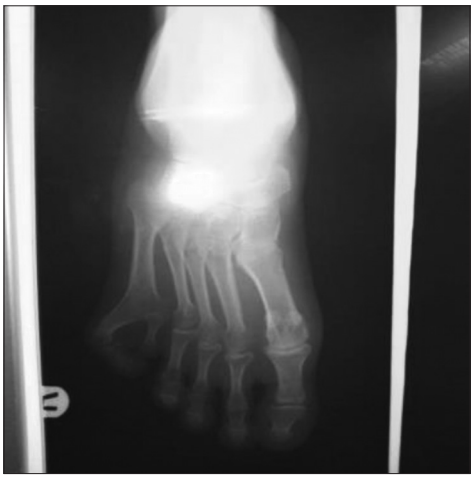

Figure 5: X ray of the foot of the 29year old man.

DISCUSSION

Polydactyly can present in multiple ways. It is can be a familial condition, a component of a syndrome or as an isolated disorder.[3,9] It is a congenital condition and is usually seen at birth with most patients presenting shortly after birth. It is typically inherited as an autosomal dominant disorder with varying degrees of penetrance and also can present as an isolated disorder without familial occurrences and no other abnormalities.[4,8] The cases presented are non syndromic polydactylies manifesting as familial and isolated disorders, respectively. The first case presented shortly after birth. She had a familial variety as her mother had also been born with the same bilateral preaxial polydactyly for which she had excision. No other congenital anomaly was noted ruling out its existence in a syndrome. She had a bifid first metatarsal with a complete duplication of all phalanges on both feet. She had an excision of the extra digits through the metatarsophalangeal joint, with sparing of the soft-tissue

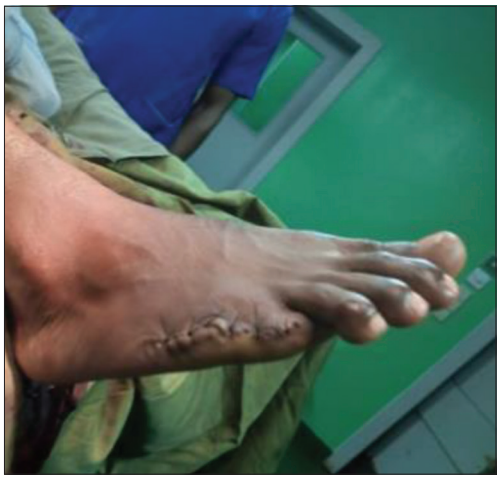

Figure 6: Post OP clinical picture of the foot of the 29year old man

structures on the lateral side to prevent functional impairment to the native hallux. Her immediate postoperative foot had a cosmetically acceptable appearance. Complications post-surgery can be in the form of stiffness, angulation, shortening or nail deformity.[13] The patient's mother who had excision of her extra digits during infancy was noticed to have hypoplastic big toes which can occur when vascular supply to the residual toe is compromised during surgery. Care must be taken during excision to avoid damage to supplying blood vessels as well as ligaments and tendons of the residual digits to limit post-operative complications.

The second case was a 29-year-old man. He presented with and isolated postaxial polydactyly. This was quite unusual due to the age of this presentation as most cases are seen in the neonatal period or in infancy. His complaints were largely pain and difficulty in wearing covered shoes. He noticed increasing pain in the foot during his teenage years, typically with the growth spurt that occurs during this period. There was also an increased growth of the extra digit. The pain and deformity worsened and was very discomforting. Apparently the presence of the extra digit alone had not been as worrisome as the difficulty in wearing shoes, which had an adverse effect on his job. The resultant pain from attempting to force his feet into shoes was the indication for his seeking treatment for the deformity.

CONCLUSION

Polydactyly presents in different ways with different reasons for the patients seeking care. Although a congenital condition, some patients may be seen for the 1st time in adulthood. Excision must be carefully carried out to prevent complications.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms.

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Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Goldfarb CA, Wall LB, Bohn DC, Moen P, van Heest AE. Epidemiology of congenital upper limb anomalies in a midwest United States population: An assessment using the Oberg, Manske, and Tonkin classification. J Hand Surg 2015;40:127-32.e1-2.
2. Mbuyi-Musanzayi S, Lumaka A, Asani BY, Kasole TL, Tshilobo PL, Muenze PK, *et al*. Preaxial polydactyly of the foot: Variable expression of trisomy 13 in a case from Central Africa. Case Rep Genet 2014;2014:365031.
3. Chen CP, Wang LK, Chern SR, Wu PS, Chen SW, Wu FT, *et al*. Wolf-Hirschhorn syndrome: Prenatal diagnosis and molecular cytogenetic characterization of a *de novo* distal deletion of 4p (4p16.1→pter) in a fetus with facial cleft and preaxial polydactyly. Taiwan J Obstet Gynecol 2020;59:425-31.
4. Umair M, Ahmad F, Bilal M, Ahmad W, Alfadhel M. Clinical genetics of polydactyly: An updated review. Front Genet 2018;9:447.
5. Barone C, Novelli A, Capalbo A, Del Grano AC, Giuffrida MG, Indaco L, *et al*. An additional clinical sign of 17q21.31 microdeletion syndrome: Preaxial polydactyly of hands with broad thumbs. Am J Med Genet A 2015;167:1671-3.
6. Volodarsky M, Langer Y, Birk OS. A novel GLI3 mutation affecting the zinc finger domain leads to preaxial-postaxial polydactyly-syndactyly complex. BMC Med Genet 2014;15:110.
7. Ni F, Han G, Guo R, Cui H, Wang B, Li Q. A novel frameshift mutation of GLI3 causes isolated postaxial polydactyly. Ann Plast Surg 2019;82:570-3.
8. Materna-Kiryluk A, Jamsheer A, Wisniewska K, Wieckowska B, Limon J, Borszewska-Kornacka M, *et al*. Epidemiology of isolated preaxial polydactyly Type I: Data from the polish registry of congenital malformations (PRCM). BMC Pediatr 2013;13:26.
9. Goldfarb CA, Shaw N, Steffen JA, Wall LB. The prevalence of congenital hand and upper extremity anomalies based upon the New York congenital malformations registry. J Pediatr Orthop 2017;37:144-8.
10. Dijkman RR, van Nieuwenhoven CA, Selles RW, Habenicht R, Hovius SE. A multicenter comparative study of two classification systems for radial polydactyly. Plast Reconstr Surg 2014;134:991-1001.
11. Atanda OO, Owonikoko KM, Adeyemi AS, Bajowa O. Polydactyly 24 in a female neonate. Case Rep Obstet Gynecol 2013;2013:798138.
12. Perez NM, Raciti MC. Polydactyly in a young boy: A cause of teasing and local tribal suspicion. JAAPA 2011;24:31-3.
13. de Almeida CE. Analysis of surgical results and of residual postoperative deformities in preaxial polydactyly of the hand. J Plast Reconstr Aesthet Surg 2017;70:1420-32.

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