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Hand/Peripheral Nerve

# Two Cases of Preaxial Polydactyly of the Foot: Important Implications for Plastic Surgeons

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**Summary:** Although polydactyly is quite common in general, preaxial polydactyly of the foot is rare (0.4 per 10,000 patients) and specifically associated with certain congenital abnormalities and syndromes, which can include craniosynostosis, corpus callosum agenesis, and renal malformations. We present 2 recent cases of preaxial polydactyly of the foot that highlight the importance of maintaining a high level of suspicion for associated abnormalities in these patients. The first patient, who presented with supernumerary preaxial digits on both feet, pre- and postaxial polydactyly of the hands, was also macrocephalic and hyperteloric; this presentation strongly suggested a diagnosis of Greig cephalopolysyndactyly, a *GLI3*-variant syndrome. The second patient, who had 2 preaxial digits on one foot, was found to also have a horseshoe kidney, a malformation that has been associated with limb defects as part of an acrorenal syndrome. These cases emphasize the importance of a thorough clinical approach to patients with preaxial polydactyly of the foot. Although many patients with this anomaly may be well known to geneticists, a child may be referred to a plastic surgeon for reconstruction of what is thought to be an isolated cosmetic or local functional issue. Plastic surgeons should be aware of the complex nature of preaxial polydactyly of the foot and potential syndromic presentation. *(Plast Reconstr Surg Glob Open 2021;9:e3358; doi: 10.1097/GOX.0000000000003358; Published online 17 February 2021.)*

Polydactyly of the foot is a common congenital malformation characterized by supernumerary toes at the medial aspect (preaxial), lateral aspect (postaxial), or middle of the foot (central).[1] Despite the high prevalence of hand and foot polydactyly, preaxial polydactyly of the foot is quite rare, estimated at 0.4 patients per 10,000 in an European population.[2] Supernumerary toes may have functional significance beyond difficulty fitting shoes, with functional tendons and nerves in supernumerary digits. Although specific reconstructive surgery for these anomalies may not be familiar to plastic surgeons, the principles of reconstruction for polydactyly in the hand apply: if functional musculoskeletal or neurovascular elements are absent in the retained digits, those elements when present in the supernumerary digit should be used.

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Because polydactyly may be the only visible difference in a child with associated internal abnormalities, a plastic or orthopedic surgeon may be the first specialist to evaluate the young patient. Preaxial polydactyly of the foot is specifically associated with other congenital abnormalities and syndromes, which can include craniosynostosis, corpus callosum agenesis, and renal malformations.[1,3] Unfortunately, the surgical literature on preaxial polydactyly of the foot is sparse and focuses more on classification and technique for removal of the extra digit(s).[4–6] As a result, there exists little clinical guidance for the surgeon presented with a young patient with these anomalies. In this report, we describe 2 recent cases of preaxial polydactyly of the foot that represent the range of conditions that can be associated with this malformation. The purpose of this article is to demonstrate the value of a multidisciplinary approach to patients with this type of polydactyly and to emphasize the importance of maintaining a high level of suspicion for associated malformations in these patients.

## CASES

### Case 1

A 4-month-old boy of French-Canadian descent was brought to the senior author (DL) by his parents for excision of extra first toes on both feet (Fig. 1A). Notably, he was also born with rudimentary postaxial supernumerary digits

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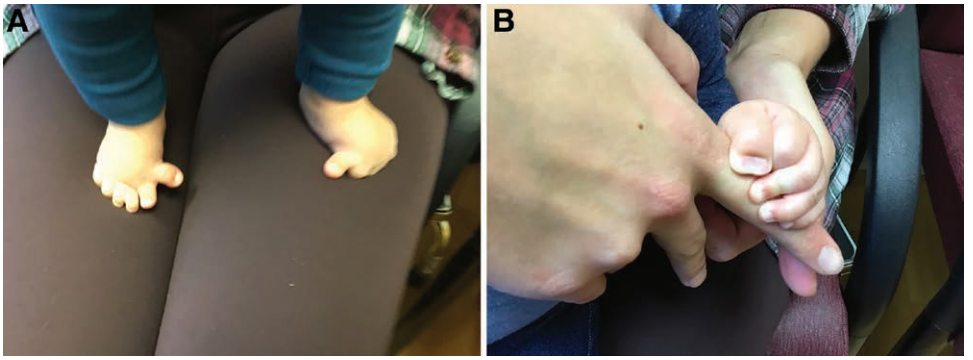


**Fig. 1.** Clinical images. The patient from Case 1 presented with bilateral preaxial polydactyly of the foot (A) and a Wassel type 1 duplicated left thumb (B).

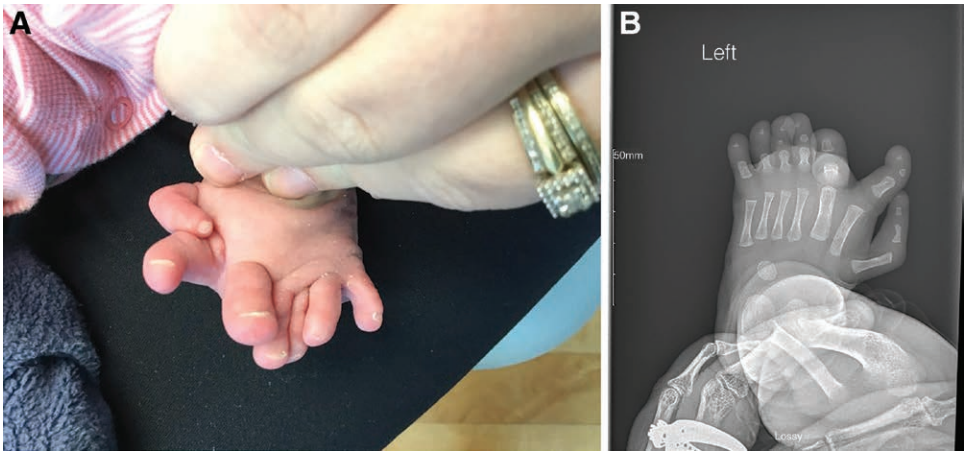


**Fig. 2.** Clinical images. The patient from Case 2 presented with 2 supernumerary preaxial toes on the left foot (A). X-rays revealed proximal, middle, and distal phalangeal bones in each supernumerary digit and a single supernumerary metatarsal (B).

on both hands and Wassel type 1 duplicated left thumb (Fig. 1B); the rudimentary postaxial digits were removed by ligation at the age of 2 months in a pediatric orthopedics clinic. In contrast, the supernumerary digits on both feet contained obvious bony phalangeal elements. The patient also displayed hypertelorism and a head circumference greater than the 99$^{th}$ percentile for his age at 2 months.

The patient was referred to a pediatric geneticist who obtained a pedigree revealing a 5-generation family history of polysyndactyly and macrocephaly following an autosomal dominant pattern. No genetic testing had been performed on any of the affected individuals, but the geneticist strongly suspected a *GLI3*-variant-associated syndrome, such as Greig cephalopolysyndactyly syndrome. The family declined testing for a *GLI3* variant. The patient underwent surgery at 10 months of age for excision of the extra digit on each foot. The supernumerary digits were nonfunctional, and reconstruction was accomplished with simple excision.

**Case 2**

A 5-week-old girl was referred to the senior author for removal of two supernumerary preaxial toes on the left foot (Fig. 2A). X-rays revealed that both extra digits contained proximal, middle, and distal phalangeal bones and a supernumerary metatarsal (Fig. 2B).

The patient was otherwise normal-appearing, but prenatal ultrasound at 20 weeks gestation had revealed a horseshoe kidney. At the age of 3 months, she was seen by a pediatric nephrologist, who performed a renal ultrasound and advised close monitoring for vesicoureteral reflux and a low threshold for further imaging to include a voiding cystourethrogram. Family history revealed that one of the patient's paternal cousins also had a horseshoe kidney, and another paternal cousin had been diagnosed with an unspecified structural kidney problem. The patient's family declined further genetic work-up. The patient underwent surgery at 8 months of age for excision of the extra toes of her left foot. During surgery it was found that the abductor hallucis, the flexor hallucis longus, and an anomalous tendon slip of the extensor hallucis longus inserted on a supernumerary digit. The abductor hallucis and flexor hallucis longus were sutured to the collateral ligaments of the retained great toe digit for reconstruction. Postoperative appearance was satisfactory.

## DISCUSSION

These cases demonstrate the importance of maintaining a comprehensive clinical approach in treating patients with preaxial polydactyly of the foot. Nearly half of all patients with preaxial polydactyly of the foot have multiple congenital anomalies, which can range from benign differences like syndactyly or macrocephaly to severe malformations, such as craniosynostosis, corpus callosum agenesis, or renal agenesis.[2,3,7] Burger and colleagues identified 21 distinct disease entities associated with preaxial polydactyly of the foot and proposed 3 main groups based on phenotype.[7]

Patients in the first group have isolated preaxial polydactyly and do not merit genetic testing due to the high frequency of sporadic cases.[7,8] The second group, which includes our patient from Case 1, consists of patients with preaxial polydactyly plus additional limb malformations. These patients are often found to have mutations in the *GLI3* gene, which codes for a downstream mediator of the Sonic hedgehog (Shh) cell signaling pathway. Shh plays an essential role in developmental tissue patterning of the limb and digits, as well as cranial suture morphogenesis and calvarial bone development.[9] When preaxial foot polydactyly co-occurs with macrocephaly and hypertelorism, the phenotype is classified as Greig cephalopolysyndactyly syndrome.[10]

Patients in the third group—those with preaxial polydactyly and an anomaly in a different organ system—should be followed closely and referred to a clinical geneticist for evaluation.[7] These patients are more likely to have a syndromic cause for their malformations, such as craniofrontonasal dysplasia, Apert syndrome, Carpenter syndrome, or Pfeiffer syndrome.[7] The third group also includes some patients affected by an acrorenal syndrome, a collection of disorders defined by the co-occurrence of congenital limb and urinary tract defects. Limb anomalies in acrorenal syndrome can include oligodactyly, syndactyly, or polydactyly, and common renal defects include renal agenesis, ureteric hypoplasia, hydroureteronephrosis, and horseshoe kidney.[3] Our patient from Case 2, with preaxial polydactyly of the foot and horseshoe kidney, belongs in this third group and had a comprehensive urologic work-up in addition to excision of her extra toes.

The cases presented here emphasize the importance of a thorough clinical approach to patients with preaxial polydactyly of the foot. Both of our patients were found to have additional abnormalities that have been previously linked to preaxial polydactyly of the foot.

## CONCLUSIONS

Young patients with preaxial polydactyly of the foot may be referred to a plastic surgeon for modification of what is thought to be an isolated cosmetic or local functional issue. In these cases, it is critical for the surgeon to be aware of the complex nature of this form of polydactyly and the potential implications for the child's overall health.

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