CASE REPORT

A rare case of mobile diplopodia mistaken for polydactyly

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How to cite this article: Woodacre T, Dunkerley S, Cox PJ. A rare case of mobile diplopodia mistaken for polydactyly. Arch Clin Cases. 2023; 10(1):15-17. doi: 10.22551/2023.38.1001.10232

ABSTRACT

We present a rare case of a mobile diplopodia in an infant with disorganization syndrome. This was initially mistaken for polydactyly due to the more typical association between these conditions. The resulting corrective surgery was more extensive and complicated than anticipated, with the removal of a partial foot duplication and reconstruction of residual hindfoot structures, rather than the planned digit amputation. We highlight the association of diplopodia with disorganization syndrome, discuss differentiating diplopodia from polydactyly and describe the surgical management of an unusual case.

KEYWORDS: Polydactyly; diplopodia; disorganization syndrome; foot and ankle surgery; pediatric orthopedic surgery

INTRODUCTION

Polydactyly (accessory digits) is the commonest digital abnormality of the forefoot [1]. It can be inherited in isolation as familial polydactyly, or occur as part of a syndrome. It is more common in black males than Caucasian males, with a respective incidence of 13.5/1000 and 2.3/1000, and in males than females [2]. Pre-axial polydactyly, involving the first ray, is much more common than post-axial polydactyly, involving the 5th or 6th ray [3,4]. Significantly, the abnormality is of the forefoot, not of the mid or hind foot. Treatment, if required, is amputation of the accessory digit and reconstruction of the local soft tissues [5].

Diplopodia (duplicate foot) is extremely rare and differentiated from polydactyly by the presence of supernumerary midfoot structures [6]. It is reported in rarity as individual cases, all of which involve other genetic abnormalities [6-8]. Significant pre-operative investigations were undertaken in specific cases to establish muscular, vascular and neurological involvement of the limb to be amputated [9].

We report a rare case of a patient with a fully mobile diplopodia, mistaken initially for polydactyly. We discuss the associated genetic aberrations and manifestations and describe the definitive surgical management.

CASE REPORT

We present a male Caucasian infant born at 40 + 5 weeks gestation by emergency caesarean section after failure to progress. He was initially diagnosed with what was thought to be polydactyly, with an accessory ray arising from the lateral aspect of the left hind-foot, which was itself in equinovarus (Figures 1 and 2). Additional congenital abnormalities included an imperforate anus (for which a colostomy was performed), a duplex kidney with enlarged left ureter, a buttock hamartoma and various soft tissue abnormalities including increased skin folds on the posterior aspect of the left thigh and a left knee contracture. Chromosome testing, ECHO, spinal radiographs and cranial ultrasound were normal. A provisional diagnosis of disorganization syndrome was made.

Examination of the accessory 'ray' revealed it to be sensate, vascularly intact and to have volitional motor control. Imaging was not undertaken due to patient distress and lack of compliance secondary to his young age.

The patient underwent surgical exploration of his polydactyc "ray" at 8 weeks of age. This however revealed a diploid foot, with an intact accessory ray (phalanxes and metatarsal) attached to a rudimentary fused calcaneocuboid cartilage structure complete with its own neurovascular supply, originating from the posterior inferolateral aspect of the calcaneum. The Achilles tendon was attached to this accessory calcaneocuboid structure and not to the normal calcaneum (Figure 3). The neurovascular supply of the diploid foot was ablated and transected. The Achilles tendon was mobilized from the accessory bone. The posterolateral aspect of normal calcaneum was feathered and the Achilles tendon reattached with vicryl sutures. The rudimentary foot was excised *en bloc*. He was managed postoperatively with a soft cast splint for 5 weeks and then pair of Bebax boots. He suffered no acute post-operative complications. His foot gained good active plantar flexion. He continues to reach normal growth and mobility milestones. Long term monitoring of his calcaneal growth is planned due to the uncertainty of the integrity of the physes of his native calcaneum.

Received: November 2022; **Accepted after review:** January 2023; **Published:** January 2023.

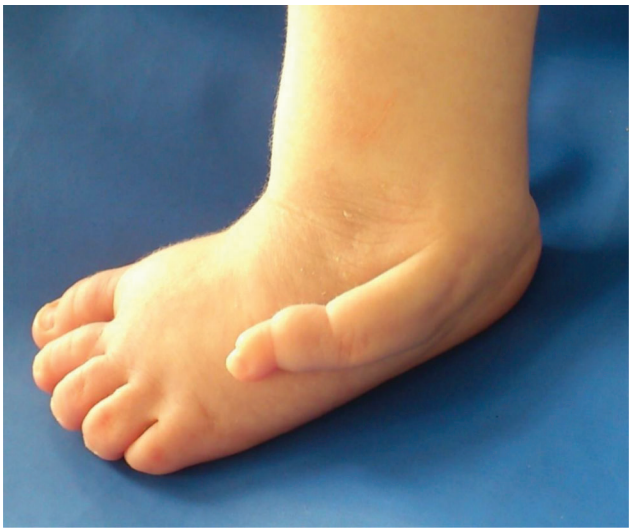

Fig. 1. Diplopodia, accessory ray, lateral view.

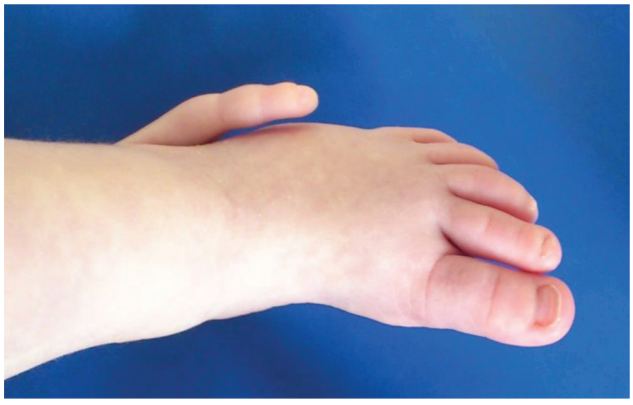

Fig. 2. Diplopodia, accessory ray, dorsal view.

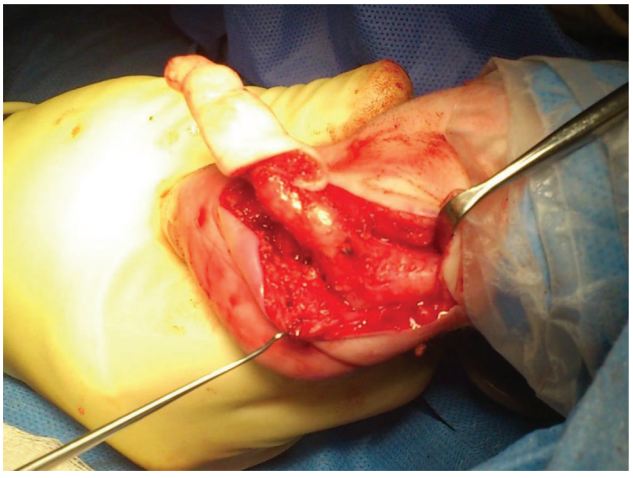

Fig. 3. Diplopodia. Surgical exploration, attachment of the Achilles tendon.

DISCUSSION

Disorganization syndrome is defined by multiple congenital abnormalities which fall outside of other recognized syndromes. It was first observed in mice by Hummel in 1959 [10] and correlated to human conditions by Winter et al. in 1989 [11]. Predisposition is thought to occur following a spontaneous genetic mutation that affects structures derived from all germ layers and expression is subsequently thought to occur following exposure to an external influencing event (potentially explaining the incomplete penetrance seen in individuals with such mutations) [12]. This latter influence is thought to occur during the first 4 weeks of development when an embryo is susceptible to global morphogenetic influences which can affect lateralization, midline structure formation and limb development, all characteristics associated with disorganization disorder [13]. Hallmark features include internal organ, skeletal and extremity duplications (commonly polydactyly), defects attributable to amniotic band sequence (for example circumferential rings, digit reduction and facial clefts), renal agenesis, hamartomas and an imperforate anus [14,15]. The patient described in this case is thought to suffer from disorganization syndrome due to the shared features described above, including extremity duplication, in this case diplopodia. Two other case reports exist in published English literature that describe diplopodia in association with an imperforate anus [8, 9]. This specific unusual combination may represent a subset of human disorganization syndrome however as the exact causative genetic anomaly of this syndrome is unknown, this cannot currently be investigated.

VACTERL association is a further group of congenital disorders linked with extremity duplication. It includes Vertebral abnormalities, Anal atresia, Cardiovascular abnormalities, tracheo-eosaphageal fistula, Renal abnormalities and Limb defects. It was initially linked to the case in question but deemed unlikely due to the patient's normal spinal and cardiovascular systems which are experienced in 80% and 75% of cases respectively. It is also more commonly associated with upper limb extremity duplication.

The presence of polydactyly or diplopodia should alert the clinician to the possibility of further congenital syndromes if not already identified. Amputation of accessory digits in polydactyly is typically relatively minor surgery. Diploid limbs often require more careful surgical planning due to vascular, neurological and soft tissue abnormalities [9]. Surgical outcome in the case presented was not affected by the intra-operative identification of a diploid limb, however pre-operative identification would typically be desired (in this case the absence of imaging pre-operatively limited the ability for this diagnosis). In this instance this resulted in an unplanned Achilles tendon transfer in an infant. However there appears to have been no negative sequalae with a neurovascularly intact, fully mobile post-operative foot. Long term follow-up will be required to ensure that normal physes continue to develop in the retained calcaneum and that they were not excised in the diploid limb or affected by surgery.

CONCLUSION

Despite the association of polydactyly with disorganization syndrome, for future similar cases, the lower rate of polydactyly in Caucasians, the low incidence of lateral,

post-axial polydactyly and any involvement of the hind-foot should provide clinical suspicion of the alternative diagnosis of diplopodia.

We hope this case adds to the evidence body for human disorganization syndrome and improves awareness of diplopodia as a surgical diagnosis.

Competing interests

The authors declare that they have no competing interests.

Consent for publication

Written informed consent from the patient has been taken and is available for review by Editor in chief of the journal.

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