CASE REPORT

Tibial hypoplasia with a bifid tibia: an unclassified tibial hemimelia

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SUMMARY

Tibial hemimelia is a rare congenital limb deficiency which is characterised by a hypoplastic/aplastic tibia. It actually represents a spectrum of anomalies, ranging from mild hypoplasia of the tibia to total absence of the tibia. Several classifications based on radiological description exist in the literature. The tibial hemimelia is usually described with preaxial mirror polydactyly, split hand/foot syndrome—ectrodactyly, polydactyly—triphangeal thumb syndrome (Werner syndrome) and micromelia—trigonal brachycephaly syndrome. We describe a child with unclassified tibial hemimelia. The child had right incomplete tibial hemimelia with bifid tibia, left complete tibial hemimelia, bilateral split hands and left split foot. This is the first report of the bifid tibia in the literature.

BACKGROUND

Tibial hemimelia is a rare congenital anomaly which is characterised by the hypoplasia or aplasia of the tibia. Several classifications exist in the literature. Each classification is based on the radiological appearance of leg and foot.^{1–3} Bifid femur, split hand, split foot and duplication/absence of formed elements of foot are associated anomalies with tibial hemimelia.^{4–6} However, to the best of our knowledge split tibia or bifid tibia has never been described in the literature. We describe a rare case of bifid tibia with tibial hypoplasia on one side with complete tibial hemimelia on the opposite side.

CASE PRESENTATION

A 1½-year-old boy was presented with congenital anomalies of all four limbs. He was the first-born

Figure 1 Clinical photograph of the left leg and foot showing flexion deformity of the knee with equinovarus deformity and absence of the great toe with ectrodactyly.

Figure 2 Clinical photograph of the right leg and foot showing equinovarus deformity with short hyperextended great toe with the presence of all lesser toes.

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Figure 3 Plain radiographs of the left side leg showing complete absence of the left side tibia with normal fibula and four ossified tarsal bones.

Figure 4 Plain radiographs of right side leg showing small tibia with bifid distal end of tibia with absence of distal tibia ossification with normal fibula.

Figure 5 Clinical and radiological images of hands showing split hand with the absence of third ray on right side and absence of three metacarpals on left side.

child to a non-consanguineous couple at full-term gestation. No known teratogen exposure was noted during pregnancy. His birth weight was 3 kg. He has no other siblings. Both parents and grandparents were normal. His developmental milestones were normal except delayed walking. His speech was monosyllabic. Left testicle was retractile. The echocardiogram was normal.

His weight was 8.8 kg. His head circumference was 44 cm. There was no facial dysmorphism. No apparent spinal deformity was present. He had congenital malformations of the lower limbs and hands. Bilateral split hands and left split foot were noted. On the left lower limb, he had fixed flexion deformity of the knee with ectrodactyly. The first ray was absent (figure 1). On the right lower limb, no fixed knee deformity with equinovarus deformity was noted (figure 2). Poor quadriceps function was noted on the left, while quadriceps function was excellent on right. Radiographs showed left complete tibial hemimelia (figure 3) and right partial tibial hemimelia with a bifid distal end of the tibia (figure 4). The hands showed split hand with an intermetacarpal split with absent third ray on the right and absence of middle three rays on the left (figure 5). Pelvis with hip radiographs ruled out developmental dysplasia of the hip and proximal femur focal deficiency (figure 6).

TREATMENT

The parents were explained for the limb ablative surgery (knee disarticulation) on the left and the reconstruction of the right lower limb. We plan to do limb ablative surgery on the left and advanced imaging with MRI on the right to know the bone and cartilage characteristics of the distal end of tibia and soft tissue release. At last follow-up, the parents are not willing for limb ablative surgery.

DISCUSSION

Several classifications exist in the literature.^{1–3} All classifications are based on the radiological appearance of tibia and fibula. Jones *et al* proposed a detailed classification which has gained widespread acceptance. Four types of deformity were recognised based on the initial radiographs in this classification. Type 1, (a and b) the tibia is completely absent; type 2, partial absence of the distal tibia; type 3, partial absence of proximal tibia; type 4, short tibia with distal tibiofibular diastasis. Kalamchi and Dawe classified tibial hemimelia into three types corresponding to Jones' radiological types 1, 2 and 4, respectively. Weber introduced a more comprehensive classification.³

It is very difficult to make comprehensive classification of tibial hemimelia due to the rarity of tibial hemimelia. However, it is necessary to report all subtle variations encountered in the

Figure 6 Plain radiograph of the pelvis showing normal development of hips.

practice to make future classification more comprehensive and meaningful.

A myriad of associated skeletal and visceral congenital abnormalities have been described in children with tibial hemimelia including diplopodia, polydactyly, syndactyly, triphalangism, clubfoot, bifid femur, proximal femur focal deficiency, developmental dysplasia of the hip, imperforate anus and caudal regression syndrome.^{4–6}

To the best of our knowledge, the association of a bifid tibia with tibial hypoplasia has not been reported and this variant has not been included in any of the current classification system. Apart from the rarity of a bifid tibia, there are treatment implications. If limb salvage and reconstruction is being planned, one needs to decide which side of the bifid tibia is to be preserved while reconstructing the ankle and foot.

Learning points

- ▶ We describe a new variant of tibial hemimelia with bifid distal tibia.
- ▶ Bifid distal tibia should be included in the existing classification system.
- ▶ If reconstruction of the limb is planned in such a rare limb deficiency, the abnormality of distal end of the tibia has to be addressed to achieve a stable ankle.

Contributors KS was involved in drafting the manuscript. HS was involved in writing the manuscript and management of the patient.

Competing interests None declared.

Patient consent Obtained.

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REFERENCES

- 1 Jones D, Barnes J, Lloyd-Roberts GC. Congenital aplasia and dysplasia of the tibia with intact fibula. Classification and management. *J Bone Joint Surg Br* 1978;60:31–9.
- 2 Kalamchi A, Dawe RV. Congenital deficiency of the tibia. *J Bone Joint Surg Br* 1985;67:581–4.
- 3 Weber M. New classification and score for tibial hemimelia. *J Child Orthop* 2008;2:169–75.
- 4 Verghese R, Shah H, Rebello G, *et al.* Pre-axial mirror polydactyly associated with tibial deficiency: a study of the patterns of skeletal anomalies of the foot and leg. *J Child Orthop* 2007;1:49–54.
- 5 Klopocki E, Lohan S, Doelken SC, *et al.* Duplications of BHLHA9 are associated with ectrodactyly and tibia hemimelia inherited in non-Mendelian fashion. *J Med Genet* 2012;49:119–25.
- 6 Wada A, Nakamura T, Fujii T, *et al.* Limb salvage treatment for Gollop-Wolfgang complex (femoral bifurcation, complete tibial hemimelia, and hand ectrodactyly). *J Pediatr Orthop B* 2013;22:457–63.

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