CASE REPORT

Unclassified tibial hemimelia

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SUMMARY

Tibial hemimelia (congenital longitudinal deficiency of the tibia) is rare (1 in 1 000 000). There are several classifications in the literature. We report an unclassified case of tibial hemimelia. A 6-year-old girl presented with shortening of the right lower limb, with a small rudimentary foot (presence of all toes) and hyper lax ankle. Quadriceps function was excellent. Radiograph showed a partial tibia and fibula in synostosis. The Jones and Kalamachi type 2 classifications both mention similar tibial anomalies; however, the fibula is normal in both varieties. The present variety can be considered as a variant of type 2 tibial hemimelia.

BACKGROUND

Tibial hemimelia (congenital longitudinal deficiency) is a very rare anomaly, with incidence of 1 in 1 000 000.¹ The tibial defect may be partial, total, unilateral or bilateral with intact fibula. It occurs as an isolated anomaly or as a syndromic manifestation with skeletal and extraskeletal malformations. Tibial hemimelia commonly presents with flexion contracture of the knee, with skin dimple at the proximal tibia region, foot in varus and supination facing the perineal region with first metatarsal shortening and other ray defects. Many classifications exist in the literature. In 1978, Jones classified tibial hemimelia into five radiological types.² In 1985, Kalamachi and Dawe³ classified it into three groups. In 2008, Weber⁴ classified tibial hemimelia depending on the radiological variations of the hip, femur, patella, tibia, fibula and foot. The first two classifications mentioned above have been widely used and accepted.

We describe a rare case with proximal tibiofibular synostosis and an absent distal part. Jones and Kalamachi have both, in their classifications, mentioned similar radiological anomalies in type 2, however, the fibula is intact in both classifications.⁵

CASE PRESENTATION

A 7-year-old girl presented with deformity and shortening of the right lower limb since birth. Her birth history was normal. All developmental milestones were within normal limits except delay in walking, requiring support (1.5 years). General and systemic examination was normal. No other congenital anomaly was present, confirmed with USG and X-ray scanogram. The child had a small rudimentary planovalgus foot with presence of all toes, a hyper lax ankle and the absence of tarsal bones (figure 1). Quadriceps function was excellent. She had genu varum deformity of the knee, with no instability. Radiograph showed proximal tibiofibular synostosis with an absent distal segment (figure 2).

Figure 1 Clinical photograph: (A) bilateral lower limbs with right tibial hemimelia; (B) right tibial hemimelia with genu varum, hyper lax ankle and planovalgus foot.

TREATMENT

The child's parents were not willing to have her undergo amputation, hence a custom made below knee prosthesis in keeping with the dorsiflexed ankle was fitted.

FOLLOW-UP

At final follow-up (6 months from presentation), the child was walking without support, using the prosthesis.

Figure 2 Plain radiograph: (A) anteroposterior view showing synostosis of the proximal tibia and fibula, with absent distal segment; (B) lateral view with absent distal tibia, fibula and talar bones.

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DISCUSSION

Tibial hemimelia is a rare congenital anomaly characterised by deficiency of the tibia with intact fibula. Many classifications exist in the literature, the most common being those of Jones and Kalamachi. The Jones classification is divided into four types. Type 1 (a, b) consists of an absent tibia with or without normal femoral epiphysis. Type 2 consists of an intact fibula with presence of a proximal tibia and an absent distal tibial segment. Type 3 is a rare variety with an intact fibula and absent proximal tibia, with distal tibial present. Type 4 is distal tibiofibular diastasis. The Kalamachi classification is grouped into three types. In type 1, the tibia is absent. Type 2 has the presence of a proximal tibia and type 3 is with distal tibiofibular diastasis. The fibula is not deformed and is intact in both author classifications.

Our patient presented with a partial tibia (proximal segment), presence of the fibula in synostosis with the tibia and an absent distal tibiofibular segment. Jones and Kalamachi type 2 classifications both mention similar radiological tibial anomalies; however, in their classification, the fibula is normal.

Complete absence of the distal tibia and fibula is not mentioned in any classification. The variety described in this case can be considered as a variant of both type 2 Jones and Kalamachi tibial hemimelia classifications.

Management of type 2 has many options, depending on the radiological type and quadriceps function. Parents are counselled before surgical intervention and options have ranged from amputation of the rudimentary foot by Syme's or modified Boyd's procedure to Brown's procedure, in which the intact fibula at the level of the tibial anlage is osteotomised and fused side to side with the fibula, using a plate and screws. The former operation is usually carried out around 10–12 months of age and the latter performed around 3–7 years of age.^{6 7}

In our case, since the proximal portion of the fibula was in synostosis with the tibia, in order to salvage the foot, a custom

made below knee prosthesis was fitted to allow the child to ambulate.

Learning points

- We describe a new radiological variant with intercalary deficiency of the distal tibia and fibula, with a rudimentary foot.
- Awareness of this radiological variant among orthopaedic surgeons is needed for diagnosis and management.
- The intercalary deficiency of the distal tibia and fibula can be managed effectively using a custom made prosthesis.

Competing interests None declared.

Patient consent Obtained.

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