We report a variant of tibial hemimelia in a six-year-old boy that did not comply with recognised classification systems. The femur and knee were normal, but the fibula was displaced proximally and there was severe diastasis of the proximal and distal tibiofibular joints to the extent that a grossly deformed foot articulated with the fibula and there was separate soft-tissue cover for the distal tibia and fibula. Although it would have been preferable to create a one-bone leg, amputate the foot and use the fibula as the stump for a below-knee prosthesis, local circumstances resulted in the choice of a disarticulation through the knee.

This was undertaken without complications, and six months post-operatively the child was walking comfortably with a prosthesis.

Case report

A six-year-old boy was referred with a deformed, short and unusable right leg. His parents were healthy and non-consanguineous and his only sister was normal. His prenatal history was uneventful. A general physical examination, including the chest and abdomen, was unremarkable. He walked with two crutches and there was shortening of the right lower limb measuring 8 cm below the knee. The right femur and knee were normal and the quadriceps had full power. The right foot had migrated proximally and was in a position of extreme varus, with the sole supinated through 180°. The first ray was absent and the foot was significantly smaller than the left. The tibia crossed the fibula in the distal third of the leg and took its position at the lateral side, leaving the talus to form a pseudarthrosis with an enlarged distal fibula, as there was no ankle joint. There was no movement in the subtalar or midfoot joints and passive pronation of the foot was impossible. The dorsalis pedis and tibialis posterior pulses were absent, but sensation in the foot was normal. There was partial active plantar flexion of the toes but no dorsiflexion. There was a separate soft tissue and skin cover for the distal tibia and the separated bones with their individual soft-tissue cover gave a bifurcated appearance to the leg (Fig. 1). Radiographs showed a proximally displaced fibula with diastasis of both the proximal and distal tibiofibular joints. The distal tibial epiphysis...
was absent but the bones of the foot were largely present (Fig. 2). A radiograph of the pelvis and hips was normal.

Various treatment options were discussed with the family, including orthotic treatment alone, limb reconstruction and amputation with a prosthesis. After discussion of the potential risks and complications, it was agreed that treatment should be disarticulation through the knee. This was undertaken without problems, and six months later the child was walking well and was satisfied with his prosthesis.

Discussion

There are a few reports on the gene loci for tibial hemimelia.\(^2,^3\) These genes with different penetrations may cause various phenotypes, for which classification systems can be useful. Because of the separate soft-tissue cover for the distal tibia and fibula, our case does not fulfil the criteria of any recognised classification system. However, it is probably a variant of Jones type 4, in which the foot is usually positioned between the divergent tibia and the fibula.

In our case a Syme’s amputation would not have been possible, and the presence of a normal knee and quadriceps would make a surgeon reluctant to perform a disarticulation through the knee. Instead, one would consider making a one-bone leg by creating a synostosis between the tibia and fibula, amputating the foot and using the fibula as the stump. This procedure should be performed in two stages. As our patient had been referred from another country and had problems with staying in Iran, a knee disarticulation was performed.

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References