Tibial Hemimelia's Deformity Reconstruction Surgery

Subjects: Biology
Contributor: Dror Paley

Tibial hemimelia is a rare congenital deficiency with a wide spectrum of pathology and deformity. This paper aims to give a comprehensive review of tibial hemimelia, with a concise summary of the history, pathology, and clinical findings of tibial hemimelia, while providing treatment recommendations and a review of the current literature. Classifications and surgical treatments are discussed, including amputation, limb reconstruction, and lengthening.

Keywords: tibial hemimelia ; tibial deficiency ; absence of tibia ; tibial aplasia ; Paley classification ; treatment ; patelloplasty ; fibula centralization

1. Introduction

Tibial hemimelia is extremely rare, with a reported incidence of approximately one in a million live births [1][2]. Though the presentation can be variable, tibial hemimelia commonly presents as a shortened leg with knee and ankle deformity. The tibia may be hypoplastic, completely absent, or a non-ossified remnant (anlage) that is invisible on radiographs. Duplication of toes, metatarsals, tarsals, fibulas, and femurs are also characteristic.

2. Evaluation

Initial evaluation should include a full set of radiographs. The presence, absence or partial presence of the tibia will help guide treatment. However, in younger children, non-ossified cartilage or an anlage will not be visible. More information can be obtained from serial radiographs as the child matures, but magnetic resonance imaging (MRI) and ultrasound may be utilized to confirm further detail, especially if no proximal tibia is present on radiographs.

Dissection of specimens with complete tibia aplasia has revealed more deficiencies within the anterior and deep posterior compartments. The posterior tibial bundle is present but shortened, and anomalous tendons may tether the foot in supination. A skin dimple is commonly found over the proximal fibula or over the knee if the patella is missing. Most had toe anomalies, ranging from four to eight digits [3][4].

Tibial hemimelia may be diagnosed with prenatal ultrasound by 16 weeks of gestation [5]. The genetic inheritance of tibial hemimelia varies. Reports have described parent to child transmission [6][7] and families with multiple affected siblings [8][9]. It may also have variable phenotypic manifestations, demonstrated by a report of identical twins with only one twin affected [10].

3. Classification

Beyond the basic classification of congenital deficiencies described by Frantz and O'Rahilly [11], the Jones classification (Figure 1) in 1978 has been commonly used [7]. The Ia group has a distal femoral epiphysis that is hypoplastic, whereas the Ib group has normal ossification that suggests that the proximal tibial epiphysis is still present. Finally, the Type IV deficiencies are marked by distal tibiofibular diastasis along with tibial shortening. Birch proposed adding a type V to include limbs with tibial shortening but with an intact proximal and distal epiphysis [12].

The Weber classification takes into the account the cartilaginous anlage, if present, and has seven types and 12 subtypes, which includes a few rarer forms of tibial hemimelia that did not fit into the Jones classification [13][14]. However, this can become a cumbersome classification to use.

Type 1 is a hypoplastic but nondeficient tibia with relative overgrowth of the proximal fibula. Subtypes include: (A) well-formed distal tibia physis, (B) a delta tibia or bracket epiphysis, and (C) delayed ossification or a cartilaginous anlage, with a missing distal tibial physis. Type 3A often has the talus located between the tibia and fibula due to the lack of tibial plafond. Type 4 is marked by distal tibial aplasia with preservation of the proximal tibial epiphysis.

Prior to the Paley classification, the wide spectrum of pathology of tibial hemimelia could not fit into any classification scheme, as demonstrated by new case reports every year [15][16][17]. The Paley classification allows for inclusion of the entire spectrum of deficiencies and duplications. When comparing the Jones, Weber, and Paley classifications, the latter was the only one that was able to classify all types of tibial hemimelia in a series of 113 cases [18]. It is also the only classification that guides treatment and prognosis.

4. Conclusions

Both the rarity and spectrum of the presentation of tibial hemimelia make it a complex and difficult deformity to treat. Many early attempts at reconstruction have failed and converted to amputation, but surgical techniques have improved over time and can provide excellent outcomes in experienced hands. It is important to classify the type of tibial hemimelia in order to determine prognosis and develop a reconstructive plan. Reconstructive surgery for the treatment of tibial hemimelia has improved over the past decade and will continue to evolve, but long-term outcomes have not been reported for the complex reconstructions of complete tibial agenesis (Paley type 5).

References

1. Brown, F.W. The Brown Operation for Total Hemimelia Tibia. In Selected Lower-Limb Anomalies: Surgical and Prostheti cs Management: A Symposium Held in Washington; Aitken, G.T., Ed.; The National Academies Press: Washington, DC, USA, 1971.
2. Weber, M.; Schroder, S.; Berdel, P.; Niethard, F.U. Nation-wide registration of limb deficiencies in Germany. Z. Orthop. I hre. Grenzgeb. 2005, 143, 534–538.
3. Turker, R.; Mendelson, S.; Ackman, J.; Lucicky, J.P. Anatomic considerations of the foot and leg in tibial hemimelia. J. P ediatric Orthop. 1996, 16, 445–449.
4. Granite, G.; Herzenberg, J.E.; Wade, R. Rare case of tibial hemimelia, preaxial polydactyly, and club foot. World J. Clin. Cases 2016, 4, 401.
5. Ramirez, M.; Hecht, J.T.; Taylor, S.; Wilkins, I. Tibial hemimelia syndrome: Prenatal diagnosis by real-time ultrasound. P renat. Diagn. 1994, 14, 167–171.
6. Nutt, J.; Smith, E. Total congenital absence of the tibia. Am. J. Roentgen 1941, 46, 841.
7. Jones, D.; Barnes, J.; Lloyd-Roberts, G. Congenital aplasia and dysplasia of the tibia with intact fibula. Classification an d management. J. Bone Jt. Surg. Br. Vol. 1978, 60, 31–39.
8. Aitken, G.T. Tibial Hemimelia, in Selected Lower-Limb Anomalies: Surgical and Prosthetics Management: A Symposium Held in Washington; The National Academies Press: Washington, DC, USA, 1971; pp. 1–19.
9. Emami-Ahari, Z. Bilateral absence of the tibias in three sibs. Birth Defects Orig. Art. Ser. 1974, 1974 10, 197–200.
10. Leite, J.A.D.; Lima, L.C.; Sampaio, M.L.B. Tibial hemimelia in one of the identical twins. J. Pediatric Orthop. 2010, 30, 7 42–745.
11. Frantz, C.H.; O'Rahilly, R. Congenital skeletal limb deficiencies. J. Bone Jt. Surg. Am. 1961, 43, 1202–1224.
12. Clinton, R.; Birch, J.G. Congenital tibial deficiency: A 37-year experience at 1 institution. J. Pediatric Orthop. 2015, 35, 385–390.
13. Weber, M. New classification and score for tibial hemimelia. J. Child. Orthop. 2008, 2, 169–175.
14. Devitt, A.T.; O'Donnell, T.; Fogarty, E.E.; Dowling, F.E.; Moore, D.P. Tibial hemimelia of a different class. J. Pediatric Ort hop. 2000, 20, 616–622.
15. Shrivastava, S.; Nawghare, S.; Dulani, R.; Singh, P.; Jain, S. A rare variant of tibial hemimelia and its treatment. J. Pedi atric Orthop. B 2009, 18, 220–224.
16. Shah, K.; Shah, H. Tibial hypoplasia with a bifid tibia: An unclassified tibial hemimelia. Case Rep. 2016, 2016, bcr2016 216622.
17. Senthil, V.; Kottamttavide, I.V.; Shah, H. Unclassified tibial hemimelia. Case Rep. 2016, 2016, bcr2016215305.
18. Paley, D. Tibial hemimelia: New classification and reconstructive options. J. Child. Orthop. 2016, 10, 529–555.