A Complex Congenital Lower Limb Deformity: Uncommon Presentation of tibial hemimelia or Popliteal Pterygium Syndrome

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Congenital lower limb deformities with complex presentations are difficult to classify and manage. We present a 9-year-old girl with deformity of right limb falling in between popliteal pterygium syndrome and tibial hemimelia. The management options include reconstruction and disarticulation or amputation. The determining factor is the earlier age at presentation and stable joints. Here, as the child was poor candidate for reconstruction, she had patella preserving disarticulation at the knee. Post-operative functional recovery was satisfactory.

Keywords: amputation, pterygia syndrome, tibial hemimelia.

The most common cause of morbidity and amputation in children in early to adolescent age group is complex congenital lower limb deformities. Although congenital entities like clubfoot and dysplasia of hip occur in 1 in 1000 population, treatment is highly successful unlike the conditions like tibial hemimelia or popliteal pterygium syndrome which are very hard to treat because of their rarity (1 in 1 million population) and need multiple reconstructive procedures and patient and family compliance.[1,2]

Case Report

A 9-year-old girl presented with complex deformity of the right lower limb since birth. The girl was of average height per her age and had 25 cm shortening of right lower limb as compared to left side **(Figure 1)**. As the child was regularly putting pressure

Figure 1: There is 90 degree flexion at the knee both on rest and standing posture. Middle third and distal third tibia shows varus deformity as well ankle going into varus.

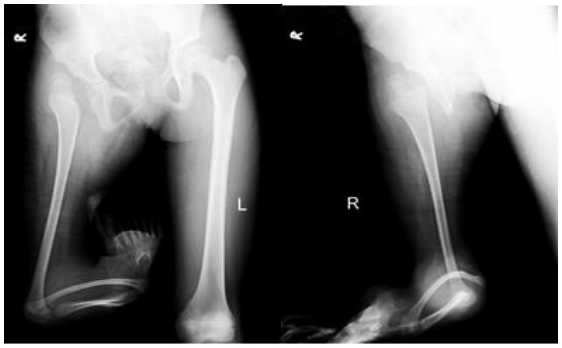

Figure 2: Anteroposterior and Lateral X-ray views showing Dislocated Right Hip. Shortened and thin bones on Right side compared to Left. Knee is subluxated with fibula migrated upward and abnormally bowed. We also see supernumerary toes and unstable ankle with missing tarsal bones.

over the knee, there was callosities present. There was a web of skin at the popliteal region connecting thigh and leg**.** The child couldn't actively flex or extend the knee. The patella was hypoplastic. The right limb had varus-medial bowing at the middle third and distal third of the lower leg. The deformity was also present on the foot with bifid great toe and supernumerary toes. She could actively dorsiflex the ankle upto neutral and do partial movement of the toes. The hip also had a positive telescopic test suggestive of dislocated hip **(Figure 2).**

She had normal sensation and no other associated anomalies seen in the body and orofacial, genito-urinary, integumentary areas and spine. As the child age was approaching maturity and had significant limb length discrepancy with hypoplasia, reconstruction was not possible and child underwent patella preserving knee disarticulation. Intra-operative findings included a very hypoplastic tibia and fibula which mimicked the scapula and clavicle bones; and foot had many tarsal bones missing.

Figure 3: A) At 6 months post-op, child has active full range of motion at the hip. B) Child on prosthesis and on full weight bearing. The hip is asymptomatic.

At 6 months follow up, the patient can carry out daily activities easily with the help of a prosthesis **(Figure 3)**.

Discussion

Popliteal pterygium syndrome and tibial hemimelias are both rare congenital malformation entities. Unlike the classically described Popliteal pterygium syndrome, our child didn't have cleft lip or palate with lower-lip pits; neither she had genital anomalies. However, she had a unilateral typical pterygia at the knee.[3,4] Similarly tibial hemimelia also comes with similar picture with associated deformities of deafness, cleft palate, pseudo-hermaphroditisim, cryptorchidism and hypospadias which were again absent in our child.[5] Syndactyly and ectrodactyly may be present in both. A weak quadriceps or deficient quadriceps mechanism with flexion contracture of the knee are also often present.[6,5,7] Our atypical case couldn't be accommodated either in Jones classification nor in Paley's new classification. However, it falls in between a popliteal pterygium syndrome and a tibial hemimelia.[8,9] The latter being a more prognostic classification helps in management.

The management and prognosis is to be thoroughly discussed with the care-givers. What determines the outcome is not merely the deformity or shortening but rather the concomitant joints stability at the knee and the ankle. The goal is an ambulatory child with stable joints. In 1965, Brown devised a technique of reconstruction of knee joint even in total absence of tibia.[2] Similarly, ankle stabilizing and limb lengthening procedures have been pioneered by Dror Paley.[8]

If the child presents earlier and caregivers promise to have strict compliance to reconstructive procedures, then the choice would be reconstruction of the knee and ankle followed by lengthening provided functioning muscles are present.[8,10] If the child is brought late with severe muscle atrophy and flail joints, then the option would be amputation or disarticulation.[7,11,12]

Conclusion

The presented case represents an extremely rare and challenging congenital anomaly,

where options include reconstruction versus amputation. In the absence of stable joints and extreme limb length inequality, the latter was chosen.

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