Original Research Paper

Volume - 7 | Issue - 7 | July - 2017 | ISSN - 2249-555X | IF : 4.894 | IC Value : 79.96

Orthopaedics

REHABILITATION OF TYPE I CONGENITAL TIBIAL HEMIMELIA WITH ORTHO-PROSTHESIS- A REPORT OF THREE CASES

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ABSTRACT Congenital absence of the tibia is a rare anomaly with an incidence of one per 1,000,000 live births. It is mostly sporadic and can be identified as an isolated disorder or as part of malformation syndromes. The treatment depends on the type of tibial hemimelia. Multiple reconstructive surgeries and the availability of suitable prosthesis have an important role in treatment options. Here, we are reporting three adolescent girls out of which two cases of bilateral congenital Type I, one case of unilateral Type I associated with radial club hand opposite side .Both the bilateral cases were walking over deformed leg with flexed knee, and the unilateral case was using a pair of axillary crutch for her ambulation. There were remote chance for reconstruction in any of these cases. None of them were ready for amputation. Modified above knee prosthesis with specially custome designed socket was prepared accommodating the residual deformed limb. All of them were satisfied with their prosthesis.

KEYWORDS : tibial hemimelia, prosthesis, club hand

Introduction

Congenital absence of tibia is a rare anomaly, It has an estimated incidence of 1 in 1 million live births. Both autosomal dominant and recessive patterns are described[1,2] There may be total or partial, unilateral or bilateral. Total absence is more frequent than partial, unilateral absence occurs more often than bilateral, with right limb more commonly affected than the left. In partial defect, almost always the distal end of the bone is affected, and of the bilateral cases, there may be total absence on both sides, or total on one side and partial on the other. Males are slightly more commonly affected than the females. Though, the family history is usually negative for congenital abnormalities and other diseases, there is a considerable chance of occurrence of congenital defect of the tibia or of other abnormalities, in near or remote relatives Family history of congenital anomaly of tibia, as well as intra-familial phenotypic variations of the defect in twin pregnancy has been reported [3,4]. The anomalies in which the number of skeletal elements increased arise during the first 7 weeks of intrauterine life. A decrease in the number of skeletal parts may arise after, as well as during, this 7-week period [5] The mother of the index case 1 was suffering from pyrexia of unknown origin during the first trimester, and this might have led to the defect by its interaction with some unidentified environmental influences and absent intake of multivitamin supplementation by the mother.[6]

Though the exact etiology is unknown, probable causes are- disruptions during the critical period of embryonic limb development (i.e 4th to 7th week of IUL), vascular dysgenesis, viral infections ,trauma and environmental influences (like smoking) and thalidomide embryopathy etc.

Jone's Classification

1. Type-**1:** a) Total absence of tibia with hypoplastic lower femoral epiphysis
b) Congenital absence of tibia with normal lower femoral epiphysis
2. **Type-2-** Proximal tibia is present.
3. **Type-3**- Distal tibia is present (Rare).
4. **Type-4**- A divergence of distal tibia and fibula with proximal displacement of talus

Reconstructive surgery and a prosthesis adapted to growth together with regular post operative follow up are necessary for optimal functional results.

According to the classification, cases are managed.

1. If the entire tibia is absent-there is often a fixed proximal and lateral positioning of fibula with severe flexion deformity. Here knee disarticulation is generally preferred, although centralisation of fibula (Brown's procedure) combined with Syme's amputation [10] has been described for this situation.

2. In cases of total tibial hemimelia,with poor quadriceps function, long term results of Brown's procedure are not promising.

3. When proximal tibia is present, it can be fused to the fibula with a Syme's amputation and a very reasonable functional limb is achieved.

4. For the distal divergence- Syme's amputation has most often been performed as considerable limb length discrepancy often accompanies this condition.

5. For Type 4 deficiencies- Open reduction of ankle and lengthening is successfully reported, though the treatment is essentially on an individual basis.

Long term result of Browns procedure are not encouraging except type IV deficiency Amputation is usually the preferred treatment option in complete absence of the tibia; however, a conservative management might be implemented in partial forms or in case of amputation refusal because foot ablation or knee disarticulation are not accepted by the parent so easily. It is very difficult to convince parents regarding amputation in early childhood. In long run patients became adopted to their deformity and they use their residual deformed foot and leg for indoor ambulation. To prevent callosities and ulceration they use adoptive devices and alternative mode of transport. This is frequently seen in Bilateral tibial hemimelia

We present here with 3 adolescent girls of tibial hemimelia managed conservatively by the use of specially designed orthosis and prosthesis accommodating their deformed limb. In all the three cases there was no history suggestive of consanguinity among parents. There was no history of any fetotoxic drug intake or radiation exposure. There was no similar congenital malformation in any other family member.

Case report – 1

A 15 years girl reported to us with complaints of bilateral leg shortening and deformity at knee and feet present since birth. On examination both leg gross shortening with flexion deformity of 100 degree. Equino-varus deformity of both the feet with on foot facing perineum and other one completely inverted facing to up.(Fig 1) All the toes were present. Radiographs (AP and lateral views) were taken and diagnosis of bilateral congenital Type I (IA on the left side and IB on the right side) intercalary longitudinal tibial hemimelia was made. She was walking on her flexed knee and leg developing callosities over knee and dorsolateral aspect of ankle. She was persuading her intermediate. She was uncomfortable with her friends and psychologically disturbed of her gross shot stature. She was not agreed for amputation as she was using her deformed leg and foot for ambulation. Custom made above knee prosthesis was designed for both the lower limbs accommodating her deformed leg and foot. Sudden raise of centre of gravity, leads to difficulty in balancing the

prosthesis. With gradual gait training, she achieved the confidence comfortable walking with bilateral modified above knee prosthesis (Fig.2). Looking towards her indoor ambulation a special designed orthosis was prepared with soft ethaplex lining (Fig.3).

Figure - 1

Figure - 2

Figure – 3

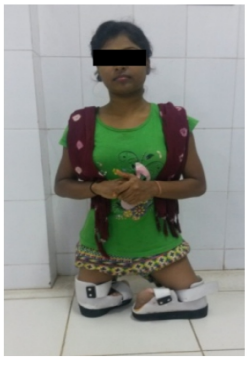

Figure – 4

Case report – 2

16 years girl presented to us with complains of deformity of both leg and feet which are present from birth and difficulty in walking. On examination her both legs were grossly shorten, severe flexion deformity was present at both knees, both feet were in supination and varus, sole of the feet were facing toward up, bilateral absence of great toe.(Fig-5) Quadriceps mechanism was insufficient bilaterally. Callosities were prominent over both knee. Distal end of both fibula was prominent, and single bone was palpable in both legs. Distal neurovascular status was normal. Radiographs (anteroposterior [AP] view) of both knees including hips and ankle were taken, and diagnosis of bilateral congenital Type I A terminal longitudinal tibial hemimelia was made. She use to walk over knee for indoor ambulation and finds difficulty in outdoor ambulation. She was counseled for knee disarticulation for a better prosthetic fitting. She refused for amputation as she is comfortable with walking on knees for indoor ambulation . she was fitted with a pair of stubbies, a special type of short limb prosthesis accommodating her residual deformed limb. She was very much comfortable with outdoor and indoor ambulation using the stubbies. (Fig-6)

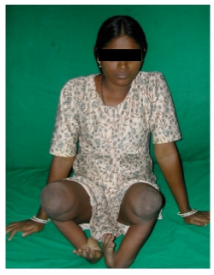

Figure - 5

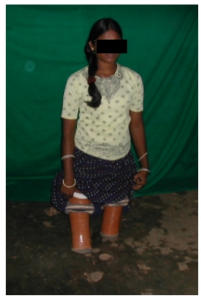

Figure - 6

Case Report – 3

17yrs old girl presented to us with complains of deformity of right leg and foot and deformity of left wrist and fore arm. On examination leg was attached to thigh with a skin web. Foot was facing anteriorly between the thighs with absence of great toe. Left forearm was thin and cylindrical. Wrist deviated to radial side with a non functioning thumb(Fig.7). Radiological evaluation confirmed the diagnosis of tibial hemimelia type 1A and radial club hand with intercalary longitudinal deficiency of radius. As there was gross limb length discrepancy and she can not effectively use axillary crutch because of her club hand, above knee amputation was suggested for her. The patients as well as parent were not agreed for amputation on cultural point of view. She was fitted with a above knee prosthesis with specially designed socket accommodating the residual deformed leg and foot. (Figures.8,9)

Figure – 7

Figure – 8

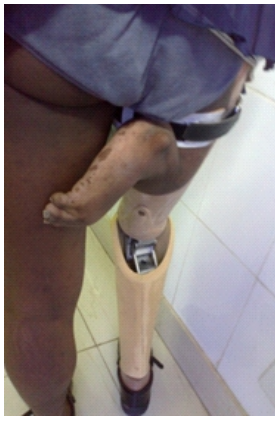

Figure – 9

Discussion:

Treatment options for tibial hemimelia with or without associated anomalies varies according to type of deficiency as well as to culture and acceptance of parents to different treatment modalities especially amputation.

Treatment depend on the quadriceps efficiency, degree of knee flexion contracture, radiological deficiency of tibia, cultural acceptance, parent's acceptance for multiple surgeries or amputation, cost of treatment. Clinical evaluation of the quadriceps extensor mechanism is important because this has significant prognostic value regarding the potential for reconstruction of the knee.

In Type 1A, there is often a fixed proximal and lateral positioning of fibula with severe flexion deformity. knee disarticulation with best prosthetic fitting gives excellent results.[7,8,9] In Type 1B, a functional knee joint exists, and knee disarticulation is not required if the quadriceps mechanism is present and functional. Centralization of fibula (Brown's procedure) combined with Syme's amputation[10] has been described for this situation. In cases of total tibial hemimelia, with poor quadriceps function, long term results of Brown's procedure are not promising.

Study on amputation verses reconstruction[11] using SF-10™ scores, the resultant score were similar in disarticulated and reconstructed patients ($p=0.63$) at the end of 7 year follow up. All scores were significantly higher when disarticulation was performed in cases of knee instability ($p<0.01$). Surgical reconstruction aiming toward possible deformity correction with foot preservation[12] for subsequent use of orthosis or prosthetic compensation improves the overall ambulatory function.

Conclusion:

Classical management protocol for different type of tibial hemimelia may not be applicable when dealing with a adult tibial hemeimelia. By that time they are adopted to their deformities and they use the residual limb for ambulation purpose. The racial and ethnic matter plays a great role against amputation.. There is paucity of literature on prosthetic rehabilitation of tibial hemimelia, despite the fact that even with the best reconstruction procedure the patient need some form of orthosis or prosthetic compensation. Functional evaluation is essential and alternative method of prosthetic management may be looked for before imposing amputation over the patient.

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