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# Split hand/foot malformation with longitudinal deficiency of tibia: novel rehabilitation approach of a rare syndrome

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## ABSTRACT

**Background:** Split hand/foot malformation with longitudinal deficiency of tibia (SHFLD1) is an extremely rare congenital anomaly, even rarer are the descriptions in the literature of suitable techniques of treatment. Most of the literature available are case reports, based on genetic studies. This study highlights on functional management rather than anatomic correction of deformity. A clinician may not come across a patient with such a rare congenital disorder during his entire career.
**Methods:** On retrospective review of hospital record from July 2008 to July 2018, we observed 5 cases of tibial deficiency associated with split hand and or split foot anomalies and diagnosed as SHHLD syndrome. Clinical and radiological analysis was done to plan out surgical and rehabilitation protocol for individual cases.
**Results:** As per Flatt's grouping of split hand, 3 limbs (37.5%) were group-1 type, 3 limbs (37.5%) of group-2 type and group-3 type was seen in 2 limbs (25%) of a patient with bilateral involvement. With respect to Jone's radiological classification system, 4 limbs (57%) were of type Ia, 2 limbs (29%) were having type IV deformity and 1 limb (14%) was with type II deformity. One case agreed for amputation was fitted with above knee prosthesis and another one with custom designed bend knee prosthesis. Eldest case was rehabilitated with tricycle and modified orthosis. Surgical correction was tried in one case.
**Conclusions:** Functional rehabilitation instead of anatomic correction should be tried at late presentation. Cleft closure should be done before development of functional adaptability.

**Keywords:** Ectrodactyly, Gallop-Wolfgang complex, Split hand, Tibial hemimelia, Tibial aplasia, Prosthesis

## INTRODUCTION

Split hand / foot deformity with longitudinal deficiency of tibia (SHFLD1) is an uncommon congenital deformity. The incidence of congenital tibial deficiency (tibial hemimelia) is 1 in 1 million live births. Association of split hand and or foot deformity further lowers the incidence to an extremely uncommon syndrome. The condition is categorised under "Rare disease" by the Office of Rare Disease (ORD) of National Institute of Health and by Ophanet. It is also called as tibial aplasia-ectrodactyly syndrome. Clinically it has wide range of presentation from isolated hypoplastic halluces to classic AHFM with tibial aplasia.[1] There are other phenotypes of SHFLD which include longitudinal deficiency of fibula and ulna. Whenever there is unilateral involvement, the affected extremity may have variable presentation of mild digital changes such as clinodactyly or shortening of individual elements to monodactyly with only the 5$^{th}$ finger remaining. Lower limb involvement ranges from

shortening of the tibia to tibial hypoplasia, tibial aplasia and hypoplasia of fibula. Males are more commonly affected than females. First case of such type of malformation was reported by Sir Ambroise Pare in 1575 which he described it as Gallop-Wolfgang Complex. White and Baker in 1888 reported first time about the first familial inheritance of this disorder.[2]

Reconstruction of limb in tibial hemimelia is a surgical challenge with unpredictable results. Traditional treatment by amputation followed by fitment of prosthesis is not accepted everywhere because of social and cultural issues. Most of these patients need some form of ambulatory aid for their community ambulation which requires fair function of hand. Association of split hand / foot deformity limits the use of ambulatory aid and further complicates the protocol of management.

There is a paucity of literature available on split hand foot limb deficit syndrome. Only sporadic case reports on genetic studies are available. A clinician will hardly get a chance to come across such a case in his clinical career. Looking towards the complexity of presentation of this rare condition, there is no definite guide line existing for its overall treatment. Multiple surgical procedures may be needed with unpredictable results and ultimately requires opinion of a physical medicine and rehabilitation expert. A comprehensive rehabilitation management is helpful for such kind of patients.

## METHODS

On retrospective review of hospital record from July 2008 to July 2018, a total 25 cases of tibial hemimelia have been reported to our institute. Out of those 25 cases, 5 cases were associated with split hand and or split foot anomalies and diagnosed clinically as SHFLD syndrome. The study was approved by the institutional review board. The inclusion criteria include- cases of tibial hemimelia associated with split hand and or foot deformity, parent had given consent personally or telephonically for participation in the study. Those cases with split hand/foot deformity associated with ulnar or fibular isolated hypoplasia, fatal congenital anomalies and those who had not given consent to participate in the study, were excluded from the study group.

The hospital record of the patients was analyzed for their demographic criteria like age, sex, family history, social background, side of affection and associated split hand and or foot deformity. The radiographs of leg, foot and hand were analysed to determine type of limb deficiencies. The clinical findings of all the patients were recorded with respect to limb length discrepancies, knee flexion deformities; type of hand and foot deformities. Jone's radiological classification was used for classifying the tibial deficiency (Table 1). Split hand deformity was classified as per Flatt's classification (Table 2), split foot deformity by Blauth and Borisch (Table 3).[3,4] Limb length discrepancy was recorded as 'absent'(-), 'mild' (+), 'moderate (++), 'severe' (+++). Standard clinical goniometer was used for measurement of Knee Fixed flexion deformity. None of our patients had the usual associated anomalies of the syndrome like cleft lip, cleft palate, congenital heart disease, imperforate anus, deafness and congenital ptosis.

## RESULTS

Out of 5 patients of SHFLD syndrome reported within 10 years period to our rehabilitation institute, 4 were male and 1 was a female child. Age ranges from 1 year to 40 years. Taking the 40years old as outlier, the mean age was 8.5 years with standard deviation of 7.2. The demographic data and variations of clinical characteristics of the study participants were observed (Table 4). Absence of tibia (tibial hemimelia) was marked in 7 limbs of 5 patients. With respect to Jone's radiological classification system, 4 limbs (57%) were of type Ia, 2 limbs (29%) were having type IV deformity and 1 limb (14%) was with type II deformity (Figure 1). Severe limb length discrepancy with was observed in 60% cases. Almost all the patients had Knee fixed flexion deformity varies from $35^0$ to $120^0$. The mean range of flexion deformity was $84^0$ with standard deviation of 34.5 (Table 4). Of the 10 feet of 5 patients 7 feet were having equinovarus deformity with absence of $1^{st}$ ray in one foot, $1^{st}$ to $4^{th}$ ray in 2 feet, $1^{st}$ to $3^{rd}$ ray in 3 feet. Split foot with absence of $2^{nd}$-$4^{th}$ ray was observed in one foot which was plantigrade. 8 hands of 5 patients had split hand deformity with bilateral involvement in 3 cases. As per Flatt's grouping of split hand, 3 limbs (37.5%) were group-1 type, 3 limbs (37.5%) of group-2 type and group-3 type was seen in 2 limbs (25%) of a patient with bilateral involvement (Figure 2). Except one patient none of our patient had positive family history of limb anomaly.

**Table 1: Jone's classification.**

| | |
|---|---|
| **Type Ia:** | Total absence of tibia with hypoplastic lower femoral epiphysis |
| **Type Ib:** | Congenital absence of tibia with normal lower femoral epiphysis |
| **Type II:** | Proximal tibia is preserved with a short tibial segment. Distal end of tibia absent |
| **Type III:** | Distal tibia is present, but proximal tibia absent |
| **Type IV:** | A divergence of distal tibia and fibula with proximal displacement of talus |

**Table 2: Flatt's classification of split hand.**

| | |
|---|---|
| **Group 0** | All bones present |
| **Group 1** | One ray involved |
| **Group 2** | Two rays involved |
| **Group 3** | Three rays involved |

**Table 3: Classification of cleft feet by Blauth and Borisch.**

| | |
|---|---|
| **Grade- I** | Five completely normal metacarpal. Total or partial aplasia of toes 2-5, but usually with the involvement of toes 2 and 3. |
| **Grade- II** | There are 5 metatarsals, but they are partially hypoplastic or synostotic with other metatarsals or phalanges. At least one toe is absent |
| **Grade- III** | There are only four metatarsals. Number 2 or 3 is always missing and the other is hypoplastic. |
| **Grade- IV** | There are only three metatarsals. Number 2 and 3, or 3 and 4 are absent. Toes 2–4 are absent. |
| **Grade- V** | The so-called lobster-claw foot. Complete absence of second, third and fourth rays. |
| **Grade- VI** | Monodactylous cleft foot with only the fifth metatarsal |

The traditional method of treatment for 57% limbs of our series with Jones Type Ia deformity is amputation. However after adequate counselling of the parents about the prognosis of the condition and the need for multiple surgeries and that amputation may be needed at any stage, most parents did not agree for an amputation. Above knee amputation followed by the fitment of prosthesis was done in one patient aged 18 years with type Ia tibial hemimelia (Figure 3A and B). One patient with type II tibial hemimelia had undergone surgery with ankle centralization and in second stage tibialization of distal fibula and proximal fibular growth arrest (Figure 2 A and B). A modified ankle foot orthosis was fitted thereafter for maintenance of correction. Parents of one patient, who was undergoing serial plaster casting of his feet deformity, opted to seek opinion of a higher centre and did not return for treatment at our centre. One patient reported as late as 40 years age (Figure 3) was accustomed to the deformities. He was prescribed with a pair of modified orthotic sandal for indoor ambulation and a tricycle for community ambulation and vocational rehabilitation. One adolescent boy was fitted with bend knee prosthesis with a special custom designed socket for accommodating the deformed limb (Figure 4 A and B). He was managing indoor ambulation using his residual limb, hence was not interested for amputation.

**Table 4: Clinical profile of case series.**

| Sl. No. | Age (years) | Sex | TH type | Split hand (Flatt's group) | Split foot | LLD | KFD µ- 84 SD-34.5 | Management |
|---|---|---|---|---|---|---|---|---|
| **1** | 13 | M | Ia | II (Rt)<br>I (Lt) | Equinovarus<br>Absent 1st toe (Rt) | +++ | $110^0$ | Bend knee prosthesis |
| **2** | 2 | F | II | I (Rt) | Equinovarus (Rt) | ++ | $50^0$ | Surgery |
| **3** | 1 | M | IV (B/L) | II (B/L) | Single toe (B/L) | + | $35^0$ | Plaster |
| **4** | 40 | M | Ia (B/L) | III (B/L) | Medial 3 toes absent | +++ | $105^0$ | Tricycle and modified sandal |
| **5** | 18 | M | Ia | I (Rt) | Split foot (Lt)<br>Med 3 toes absent | +++ | $120^0$ | Amputation &fitment of above knee prosthesis |

TH- Tibial Hemimelia, LLD- Limb Length Discrepancy, + - Mild, ++ - Moderate, +++ - Severe, KFD- Knee Fixed flexion Deformity, µ - Mean, SD- Standard deviation.

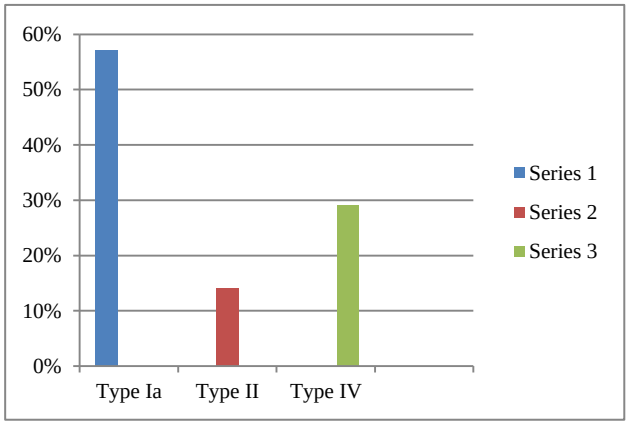


**Figure 1: Jone's type of tibial hemimelia.**

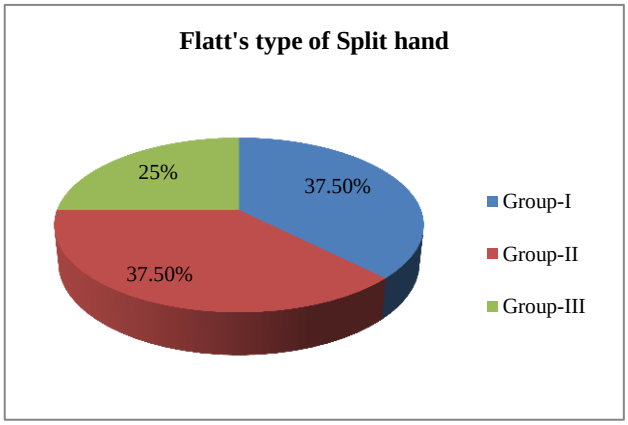


**Figure 2: Flatt's grouping of split hand.**

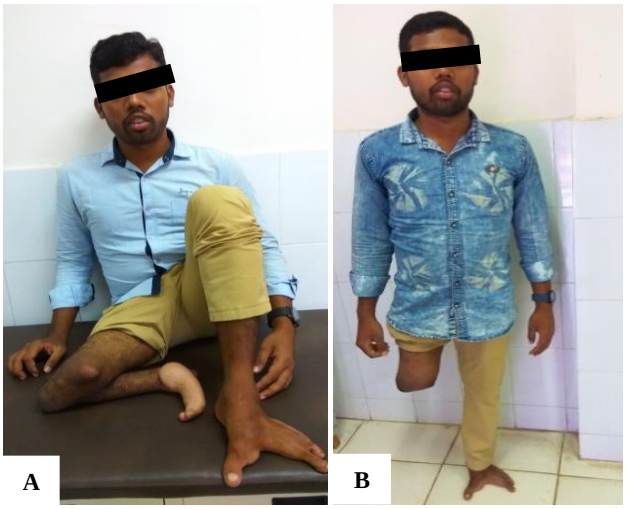


**Figure 1 (A and B): SHFLD with type Ia tibial hemimelia with split hand and foot deformity treated with amputation for prosthetic fitting.**

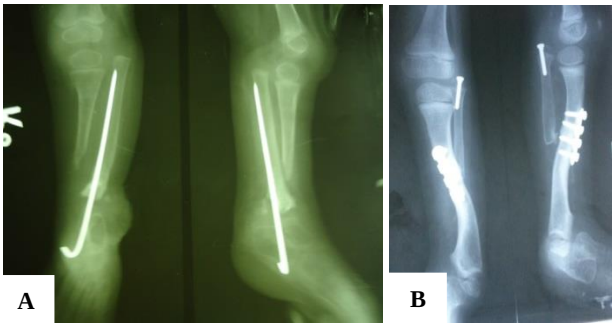


**Figure 2 (A and B): SHFLD with type II tibial deficiency treated with fibular centralization at ankle followed by tibialization of fibula after 3 years.**

**Figure 3: Type Ia tibial hemimelia with group III split hand deformity.**

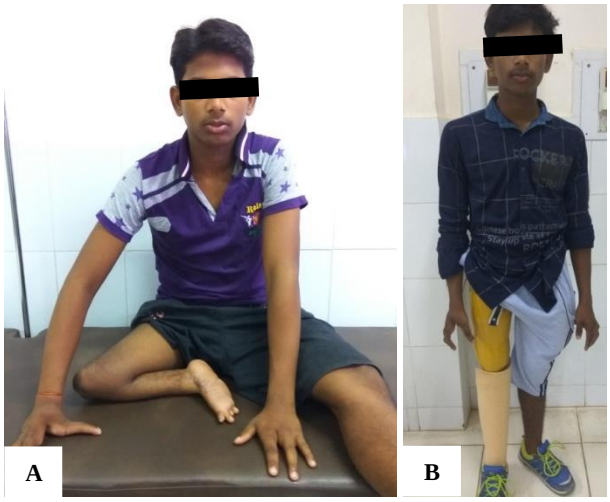


**Figure 4 (A and B): SHFLD with type Ia tibial aplasia and split hand deformity managed with bend knee prosthesis.**

All the cases except one that has lost to follow up after serial plaster casting were from poor socioeconomic group and under privileged area. All were very much accustomed with their hand deformity and hardly found any difficulty in their activities of daily living. As ambulation was the chief concern of the patient and parent, none of them agreed for reconstruction of split hand deformity. A uniform hand function assessment was not possible as two of the cases were of within 2 years age. All the patients had undergone thorough assessment of their hand function by occupation therapist with respect to activities of daily leaving. Alternative trick movements were explained for the patients not interested for hand surgery.

## DISCUSSION

The upper and lower limb buds appear at 4$^{th}$ weeks of conception. The limbs form rapidly during the subsequent 3 weeks in a proximo-distal sequence, hence the upper arm and thigh appears before fore arm and leg and in turn before hand and foot. By 7$^{th}$ week of post conception, the embryonic skeleton is well formed. Any insult to embryo that have acted between 3$^{rd}$ and 7$^{th}$ intrauterine week might affect the development of limbs. In ectrodactyly or split hand deformity there is congenital absence of one or more rays of hand.[5] Walker and Coldius reported that there is centripetal suppression of the developing hand plate.[6] Progressively more severe forms affects first the middle and then the index ray, followed by successive suppression of ulnar ray, which tends to fuse together to give a classic lobster claw appearance. The oldest morphological classification of split hand is by Barsky into two types.[7] One is typical cleft hand with a deep palmar cleft, which separates the two central metacarpals. It is V shaped and often bilateral and may involve the feet.

In the atypical cleft hand, the central rays are absent, and only short radial and ulnar digits remain with a shallow cleft giving a U-shaped deformity. Nutt and Flatt had a large series of 80 hands.[3] They have further classified the deformity clinico- radiologically from group 0 to group 3 with respect to number of rays absent.[3] Only 58% of their large series had undergone surgery with a conclusion that function of the hand cannot be predicted from its anatomical state as their patient did not consider their central deficit as a social catastrophe. In our series too, none of our patient were ready for the surgical reconstruction of their hand.

Cleft foot has been classified radiologically by Blauth and Borisch in to six grades.[4] In our series 1 foot had grade V and 2 feet had grade VI split foot deformity.

Absence of tibia combined with ectrodactyly otherwise called Gollop-Wolfgang complex is a rare malformation with highly variable manifestations.[8] The full-blown syndrome consists of aplasia of tibia and split-hand/split-foot deformity. Distal hypoplasia or bifurcation of femora, hypo- or aplasia of ulna, and minor anomalies such as patellar aplasia, hypoplastic big toes may be additional malformations. It is an autosomal dominant skeletal disorder characterized by variable expressivity and incomplete penetrance. Autosomal recessive inheritance has also been proposed in some families. There are number of literature published on genetic locus for SHFLD.[9] Lezirovitz et al identified the pathogenic chromosomal region of 17p13.1–17p13.3 and found to be associated with SHFLD syndrome.[10] The most common aetiology of SHFLD is considered to be17p13.3 duplication. Genomic duplications encompassing BHLHA9 are associated with SHFLD and non-Mendelian inheritance characterized by a high degree of non-penetrance with sex bias.[1,11] Petit et al. identified a BHLHA9 duplication in 13 unrelated families.[12] One case of our series had positive family history where his son was a case of SHFM syndrome without involvement of any long bone.

## CONCLUSION

Ambulation being the first priority in the process of rehabilitation, management of lower limb disability seeks more attention by parents as well as treating physician. Most cases of ectrodactyly do not require surgical intervention and live with modest functional impairment of the limbs. However parents should be motivated for cleft closure at early child hood for cosmetic correction before the patient became adopted to deformity. To the best of our knowledge, our study with 5 cases of SHFLD syndrome seen over 10 years will be one of the largest of this kind of study from a single center in our country.

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