



## Challenges involved in developing a personalized, tailor-made rehabilitation program for a child with congenital upper and lower limb deficiency

JANE ELIZABETH SAJAN SAJAN

Department of Physical Medicine & Rehabilitation, CHRISTIAN MEDICAL COLLEGE

**Abstract :** Congenital upper and lower limb deficiency in the same individual is a very rare condition. Here we report a case of bilateral transverse upper limb deficiency and right proximal focal femoral deficiency with fibular hemimelia in a 3 year old child. The child presented several rehabilitation challenges especially related to ambulation and activities of daily living. To aid ambulation, a modified lapboard walker was designed to enable the child to use the trunk to propel forward. The involvement of both upper limbs made ambulation training more challenging. A left universal cuff-based customized assistive device was employed to assist brushing, feeding and grooming. The fact that the child was in the growing phase was a significant factor that was considered during rehabilitation planning. Behavioural issues that interfered with the rehabilitation program were addressed with the help of a child psychiatrist. Parental counselling, re-assurance and adequate training were provided to ensure compliance to the rehabilitation program and to ensure a successful rehabilitation outcome.

**Keyword :** congenital transverse limb deficiency, proximal focal femoral deficiency, rehabilitation, lapboard walker

**Background:**

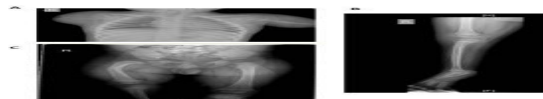
Transverse deficiencies are defined as “the presence of normal development until the point of the deficiency, beyond which the normal anatomy does not exist” (1). Congenital upper limb deficiency has an incidence of approximately 4.1 per 10,000 live births. The most common congenital limb deficiency is the left terminal transverse radial limb deficiency. Longitudinal deficiency of the fibula is the most common long bone absence. Proximal femoral focal deficiency is the second most common congenital deficiency of the lower limb. The term proximal focal femoral deficiency (PFFD) is applied to a spectrum of conditions characterized by partial absence or shortening of the proximal femur and is thought to result from an early disturbance of the growing mesenchyme. Children with limb deficiencies present unique challenges to the rehabilitation team. The rehabilitation strategy has to be multi-disciplinary and tailor-made to the needs of individual patients. Here we present a case report of a 3 year old boy who presented with absent upper limbs as well as deformed right lower limb. A diagnosis of congenital bilateral transverse upper limb deficiency and right proximal focal femoral deficiency with right fibular hemimelia was made. We discuss the rehabilitation challenges that were posed and the manner in which the case was managed.

### Case presentation:

This 3 year old boy was the first child born to non-consanguineous parents. He was delivered at the gestational age of 36 weeks by lower segment caesarian section in view of decreased amniotic fluid. At birth it was noticed that the newborn had bilateral upper limb and right lower limb deficits. The child was brought to the rehabilitation institute at 3 years of age with concerns of difficulty in walking and complete dependence for activities of daily living. There was no significant medical illness during pregnancy and no history of teratogenic drug intake. No other congenital anomaly was reported in other members of the family or first and second degree relatives. On examination, there was deficiency of bilateral upper limbs at subacromial level on right side and transhumeral level on left side. On the left stump, a rudimentary appendage was present (Figure 1).



**Figure 1:** Physical features of the child at the time of admission (3 years old) Measurements done during successive admissions showed that there was growth in his left upper limb stump. Right lower limb proximal femoral deficiency was present with absent fifth toe in right leg. Radiological investigations confirmed clinical findings and also revealed right fibular hemimelia (Figure 2). The left lower limb was normal. At the time of first admission the child had inadequate trunk control.



**Figure 2:** Radiological imaging showing bilateral transverse upper limb deficiency (A), right fibular hemimelia (B) and right proximal focal femoral deficiency (Aikter's class A)©

### Rehabilitative measures undertaken:

Rehabilitation was undertaken over three separate hospital admissions. At the time of first admission, the child's indoor mobility was by bottom shuffling and rolling on the floor. He was completely dependent for outdoor mobility. The initial rehabilitation challenge was to improve mobility. Different

options for mobility enhancement were tried since mobility training was complicated by the absence of upper limbs. The child was trained to sit independently and maneuver a tricycle for indoor mobility. Suspension walking was initiated with a modified right shoe with height correction. The shoe had a wide base and the middle part of the sole was scooped out to reduce the weight of the footwear. Gait training was continued with the help of a novel modified lapboard walker (Figure 3).



**Figure 3:** Child using the customized lapboard walker and modified right shoe with height correction

The lapboard walker allowed the child to use his trunk to propel the walker forward in order to ambulate for short distances and, by the end of the hospital stay, he was able to walk up to 50m with the lapboard walker and minimal assistance. Trunk control was crucial for mobility using the lapboard walker. Therefore, therapy to improve his trunk control was also an important consideration. This included spinal extension exercises, kneelwalking and kneel-standing. He was encouraged to use his left upper limb stump as well as left foot for coloring, scribbling and activities of daily living. A left universal cuff-based customized assistive device was employed to assist brushing, feeding and grooming (Figure 4).

**Figure**



**Figure 4:** Child holding a spoon using the left universal cuff-based customized assistive device

During the second admission 4 months later, further height correction and other modifications were done for his right footwear in addition to therapy to improve trunk control and mobility using the lapboard walker that was initiated during the previous admission. There was improvement in his posture. He was encouraged to wear the universal cuff-based assistive device for longer duration. Use of prosthetic arm was deferred in order to encourage use of the left upper limb stump. Use of the left upper limb stump and left foot was encouraged for colouring, scribbling, feeding and other activities of daily living. At discharge, he was able to walk with the help of the lapboard walker for 300m. During the third admission 6 months later, multidisciplinary team meetings were conducted to plan the prosthetic prescription. Further height correction and modification of the footwear by including a medial arch support was done. There were several challenges faced during rehabilitation of the child. There were behavioral issues (temper tantrums) that interfered with the rehabilitative process. He was assessed by the Child Psychiatrist to address his behavioral issues. ABC (antecedent behavior consequence) analysis was suggested. Rewarding for good behavior and ignoring unacceptable behaviour (time out) were suggested to address his temper tantrums. At the time of discharge after the third admission there were improvements in behavior and co-operation in therapy was better. The International Classification of Functioning Disability and Health (ICF) score at first admission and last discharge are given in Table 1. The scores are given on a scale of 0 to 4, where 0 indicates complete independence and 4 indicates complete dependence.

**Table 1:** ICF scores at admission and at the time of discharge from hospital

	At admission	At discharge
Feeding	4	2
• Rice	4	1
• Biscuits	4	4
Grooming	4	3
Bathing	4	4
Toileting	4	4
Dressing	4	3
• Upper half of body	4	2
• Lower half of body	4	2

## Discussion

Congenital limb deficiency is a rare condition. In India, a hospital-based report of 200 cases published by Jain et al (1994) (2) documents a prevalence of approximately 37 per 1000 amputees visiting their center. Among transverse deficiencies, upper limb deficiencies were relatively more common than lower limb deficiencies. The most common transverse upper limb deficiency was forearm partial deficiency (~50%). Upper arm deficiency was relatively uncommon (9 out of 200 cases (4.5%)). In addition, unilateral involvement was seen in all cases of upper arm deficiency. The present case report describes a child with bilateral upper arm deficiency, which is very rare, with an associated right PFFD, which makes rehabilitation of this child very challenging. In transverse deficiencies, all skeletal elements beyond a certain level are absent, and the limb resembles an amputation stump (1). Amniotic bands are the most common cause; the degree of deficiency varies based on the location of the band, and typically, there are no other defects or anomalies. The remaining cases are mostly due to underlying genetic syndromes such as Adams-Oliver syndrome or chromosomal abnormalities (3,4).

PFFD is a congenital deficiency that can present itself in a large variety of ways, primarily as a clinical presentation of hip flexion, abduction, and external rotation. Several methods of classifications for PFFD have been described. Aitken's method is the most broadly accepted classification (5). Aitken classified PFFD levels according to acetabular and femoral involvement as A, B, C or D, with class A being the least involved and class D being the most severely involved. In the present case, the child was classified as Aitken Class A. There are guidelines for rehabilitation of children with transverse limb deficiency published by the British Society of Rehabilitation Medicine (BSRM) in 2003 (6). These guidelines recommend that: children with congenital limb deficiency should be seen by a consultant in rehabilitation medicine as soon as possible, ideally within one month of birth, and certainly by the age of 6 months; when diagnosis has been made during gestation, the parents should be referred to the service as soon as the deficiency is recognized; early specialist occupational therapist (OT) input is obtained; the opportunity of availing of skilled counselling and support services such as clinical psychology should be offered; as parents may have concerns regarding future pregnancies, the opportunity to meet with a geneticist should be offered; the role of voluntary organizations in supporting children with limb deficiency and their families is promoted, and should be highlighted to parents. In India and many other developing countries, such guidelines do not exist. In this case, the child was first seen by a consultant in rehabilitation medicine only at the age of 3 years, instead of the recommended 6 months. Although antenatal ultrasound investigation had suggested a defect in the right lower limb, transverse upper limb deficiency was missed. Specialist occupational therapist input was delayed, as was counseling for the parents. Rehabilitative measures in transverse upper limb deficiency places heavy reliance on the intact lower limbs for ambulation and other activities of daily living (7).

However, the involvement of the right lower limb (PFFD) in this child rendered the rehabilitation process more complicated. At the time of first admission the child's mobility was restricted to bottom shuffling and trunk control was moderate. In order to take advantage of the trunk to propel movement, a lapboard walker was designed for indoor and outdoor mobility. The lapboard walker allowed the child to propel forward using his trunk. At the time of discharge, the child had acquired the ability to ambulate independently using the lapboard walker.

In cases of PFFD, Van Ness Rotationplasty is usually done to augment ambulation training using a below-knee prosthesis (8). However, it was decided against this in this child after consultation with pediatric orthopedics for the following reasons:

Absence of both upper limbs would render ambulation training with a below knee prosthesis difficult. In view of the fact that the affected right lower limb supported the normal left lower limb in activities of daily living, it was considered too risky to perform an irreversible operative procedure on the limb that would potentially jeopardize the functional outcome. The fact that the child was only 3 year old and growing rapidly meant that any prosthesis that was prescribed would have to be revised at regular intervals. In order to encourage use of the rudimentary left upper limb and the lower limbs for activities of daily living, the use of prosthesis was deferred till maximum use of the appendage could be attained. However, it is known that children use a prosthesis most successfully when it is fitted early and becomes an integral part of their body and body image during the developmental years (9). Therefore, customization of an upper limb prosthesis that could serve as many needs as possible was planned. Since the child had significant improvement in his ambulation and had achieved partial independence in ADL with customized lapboard walker and assistive device, his parents opted to initiate schooling and chose to defer definitive upper limb prosthesis to a later date during the school holidays. The child was advised to review at a later date for further rehabilitative measures including prosthesis fitting.

Behavioral issues are often associated with children with disability. This may arise due to inadequate adjustment to the environment. In addition, the child's disability may promote the parent's lack of inclination to train the child and develop an indulgent attitude which may further complicate the task of rehabilitation. These issues have to be addressed in order to ensure effective rehabilitation of the child. In this case the child psychiatrist's input contributed towards enhancing the child's co-operation with the rehabilitation efforts to achieve a satisfactory functional outcome. Concurrently, the psychological support and training of care-givers is required in most cases. Parents often require counselling, re-assurance and adequate training to address the physical and psychological burden for the on-going care of a child with congenital disability to ensure successful rehabilitation outcome (10,11).

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