

Congenital Absence of All Four Limbs A Rehabilitation Challenge

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Abstract

A child with congenital limb deficiency is best managed by an interdisciplinary rehabilitation team with full cooperation of the family members and active participation of the child himself. It is important to understand the changing needs of the growing child. Different aspects of a normal human development, mobility, activities of daily living (ADL), cognitive and psychosocial skills are to be considered while planning the management. We report a case of an eight months old male child with almost absence of all four limbs that was rehabilitated by providing custom made aids.

Key Words: Congenital Limb Deficiency, Phocomelia, Meromelia.

Introduction

Congenital limb deficiencies present a wide range of abnormalities from the absence of a single digit to the complete absence of the limb with an approximate incidence of 0.3 to 1.0 per 1000 live births¹. But complete absence of all four limbs is far more uncommon. WL Wong et al² cited 13 such cases from the literature to which they added 2 more similar cases. A vast majority

of congenital limb deficiencies are sporadic and non-transmissible. A study from the Medical Birth Registry of Norway showed that children born to a mother with a limb deficiency relatively have about 5.6 times the risk³.

Various classification systems have been devised to describe congenital limb deficiencies. The International Standards Organization / International Society of Prosthetists and Orthotists (ISO/ISPO) system have given the accepted standard classification, although its clinical application is inconsistent, and older classification systems are frequently used⁴. Older systems of classification are: Saint-Hilaire (1837), Frantz O'Rahilly (1951) and Frantz O'Rahilly revised (1961)⁵.

Phocomelia, term introduced by Geoffery Saint-Hilaire (1832)⁶, is defined as a type of meromelia characterized by absence of the proximal portion of a limb or limbs, the hands or feet being attached to the trunk of the body by a single small, irregularly shaped bone⁷. In extreme cases there is absence of proximal bones of both the upper and lower limbs so that hands and feet appear attached directly to the body (tetraphocomelia). Thalidomide, when taken during pregnancy, remains the only drug proven to have caused a large number of congenital limb deficiencies⁸. The familial form is transmitted as an autosomal recessive trait where mutation occurs on chromosome no. 8 (8 p 21.1)⁹.

Case Report

An 8 months old male child presented with a rudimentary right upper limb stump (humerus) of about 5cm, complete absence of left upper limb (Amelia), four small toe buds as the right lower limb and a foot with four digits attached to a rudimentary tibia to the pelvic girdle as left lower limb. For ambulation, the child used serpentine movements and log rolling movements. Head, neck and trunk movements were adequate and the child could manipulate toys with his left partial foot and right upper limb stump. The child's cognitive level was age appropriate and he was active and cooperative. Psychosocially, the child was affected in his limited interaction with his environment. There was no significant antenatal history (Fig 1).

The aims, objectives and sequence of the management program were sorted out with group discussions. Utmost part was the counseling of the parents. The management

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plan was directed to achieve independence of the child with mobility aids, self help assistive devices, early training and adaptation to the present condition and to get the maximum possible function.

A multifunctional seat was fabricated with resin (epoxy) and reinforced with glass fiber mat and multiple layers of cotton stockinet, which supported the child on three sides. Anteriorly, the child was secured with Velcro and canvas straps. The seat was mounted on an aluminum frame with four small casters to provide mobility. The right side wall of the seat was extended with an attachment made of the simple door cleat and latch. Over the cleat a tray was fitted (removable) that acted as a table. The tray



Fig 1. Child with tetraphocomelia.



Fig 2. Multifunctional seat with a hinged tray.

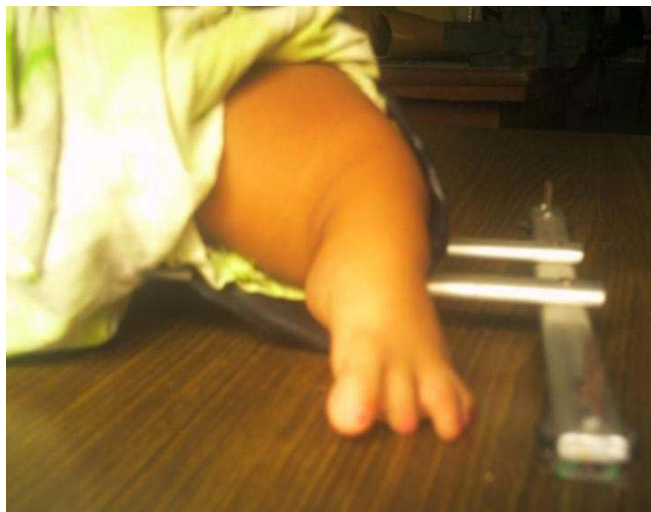


Fig 3. Cart Manipulation by the left foot



Fig 4. Above elbow prosthesis made by parts of a doll.



Fig 5. Training of the child.



Fig 6. Toilet Seat with disposable toilet pan and supports.

could be rotated side to side (Fig. 2). The child used his left foot to manipulate the cart (Fig.3).

Prosthetic fitment was considered for early prosthetic adaptation. The upper limb portion of a doll made of light weight plastic (polyethylene) was fitted to a custom made socket with a single strap suspension system across the axilla of the other side was used. The distal end of the prosthesis was fitted with a modified universal cuff to hold a pen, pencil and spoon etc. (Fig.4,5).

A toilet seat was fabricated similar to the cart seat but with a hole at the bottom and supported with four stable posters. Disposable toilet pan was used (Fig.6).

Discussion

For children with phocomelia, most adaptive devices have been designed on temporary basis and battery powered. Zazula and Foulds described an electric cart designed for a child with multiple limb deficiencies. Three separate double-pole, double-throw switches controlled the device which was operated by the child's partial left foot (to go forward) and his head (for side movements)¹⁰.

Hart designed six adaptive aids to improve classroom activities of a 7 years old boy with severe upper limb deficiencies (a typing aid, a reading aid, a chalk board aid, a dressing stick, a pull over shirt aid and a ball thrower)¹¹.

Weiss-Laubrau et al designed a device to aid for bilateral high level upper limb amputee in independent toileting¹². Nelson et al highlighted a self-directed learning module in making a decision in prosthetic management of pediatric and adult patients⁴.

In the literature, phocomelia described is either of a single limb or both upper limbs. It is very rare to have all four limbs involvement (tetrachomelia). It is important to know the mental status and learning ability of the child. From time to time new aids are required to promote the developing brain. The natural adaptations that this child acquired (serpentine movement) should not be discouraged, as these adaptations are helpful.

While discussing about the assistive and adaptive devices, sophisticated machines like motorized wheel chair as designed by Zazula et al⁹ were considered but due to socio-economic reasons the idea was dropped. It was also difficult but important to counsel the family members who were not well educated and belonged to a rural area.

The motive of reporting this case is to highlight how a totally dependent but otherwise mentally alert child could be made to ambulate to some extent with minimal possible cost using locally available resources.

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