DEMOGRAPHIC CHARACTERISTICS OF CHILDREN WITH CONGENITAL LIMB DEFICIENCY: A RETROSPECTIVE STUDY

Abdel-Fattah A. AL-Worikat MD, Ali S. AL-Ghuweri MD**

ABSTRACT

Objective: To determine the demographic characteristics, types, levels of congenital limb loss, etiological factors, prosthetic fitting in children with congenital limb deficiencies and to compare findings with other studies.

Methods: Patients with congenital limb deficiencies attending the prosthetic rehabilitation clinic at the Royal Rehabilitation Centre, King Hussein Medical Centre, Royal Medical Services in Jordan between January 1988 and January 2006 were reviewed and their demographic characteristics were analyzed.

Results: One hundred and forty three children with limb deficiency were reviewed. Sixty-two (43.4%) were males and 81 (56.6%) were females, with males to females ratio 1:1.3. The mean age at the first visit to the clinic was 7.15 years. The total number of limb deficiencies involved was 170 limbs of which 95 were upper limb and 75 were lower limb deficiencies. Of these 91 limbs were right sided and 79 limbs were left sided deficiency. The transverse limb deficiency was more than longitudinal limb deficiency, 112 and 58 respectively. The most common deficiency was the transverse forearm partial deficiency (below elbow) followed by transverse forearm total deficiency (elbow disarticulation) in the upper limbs. The longitudinal femoral partial deficiency followed by transverse leg partial deficiency (below knee) was the commonest in the lower limb. One hundred and seven (75%) children were fitted with prostheses. No definitive cause for the limb deficiency in children was found.

Conclusion: This is the first study in Jordan concerned with congenital limb deficiency and forms a baseline for further new studies. The findings can be helpful in future management and planning of materials, facilities, budget needs for children with limb deficiency.

Key words: Children, Congenital, Limb deficiency

JRMS August 2008; 15(2): 28-33

Introduction

Limb deficiencies in children may be due to congenital disorders or acquired amputations. Congenital limb deficiency means partial or total absence of one or more skeletal elements of the limb at birth. The congenital limb anomalies are classified as being either transverse, in which all the skeletal elements distal to the level of loss are absent as in acquired amputation or longitudinal in which some distal skeletal elements remain.[1-6]

*From the Department of Physical Medicine and Rehabilitation, Royal Rehabilitation Centre, King Hussein Medical Centre, Amman-Jordan
Correspondence should be addressed to Dr. AF AL-Worikat, P. O. Box 541591 Abu-Nsair 11937 Jordan,
E-mail: abdfalwr@hotmail.com
Manuscript received October 10, 2006. Accepted February 15, 2007

Table I. Characteristics of children with congenital limb deficiencies

Type of deficiency	Number of patients	Male	Female	Upper limb	Lower limb	Right side	Left side	Transverse deficiency	Longitudinal deficiency	No. of limb deficiencies
Unilateral limb deficiency	124	52	72	67	57	69	55	70	54	124
Multiple limb deficiency	19	10	9	28	18	22	24	42	4	46
Total	143	62	81	95	75	91	79	112	58	170

The etiology of most children with congenital limb deficiencies is unknown.[2,5,7-11] The incidence of congenital limb deficiencies in children has been variously reported.[2,3,11-13] Some reported that about all children with congenital limb deficiencies were fitted with prostheses, but not all use their prostheses in activity of daily living (ADL).[3,4,6]

The aim of this study is to determine the demographic characteristics, types, levels of congenital limb loss, etiological factors, prosthetic fitting in children with limb deficiencies, and to compare it with other studies.

Methods

The medical records of all patients who suffered of upper and lower limb amputations due to different causes and attended the prosthetic rehabilitation clinic at Royal Rehabilitation Center (RRC), King Hussein Medical Centre (KHMC), Royal Medical Services (RMS) in Jordan, between January 1988 and January 2006 were reviewed.

The medical records of patients with congenital limb deficiency were analyzed and the following data were obtained. Demographic data such as age of the child at the first visit of the clinic and gender, type of the defect of the upper and lower limb, in accordance with the classification of the international standard, ISO 8548-1:1989,[1] the predicted etiology of the limb loss, and if the child with limb deficiency was fitted with prosthesis or not.

The collected data were analyzed with SPSS version 11 software. Descriptive statistics were applied.

Results

We analyzed 143 children with limb deficiency from a total number of 2,393 amputees who visited the prosthetic rehabilitation clinic, making the incidence of 60 per thousand amputees. Females were more than males, n=81(56.6%), n=62(43.4%) respectively, with male to female ratio of 1.3:1. The mean age of children at the first visit to the clinic was 7.15 years ranging between six months to 15 years. The boys presented earlier to the clinic than girls. The boys' mean age was 6.3 years and the girls' mean age was 7.9 years.

Most children (n=124) were with unilateral limb deficiency, the majority of them were with upper limb involvement (n=95), and the most common deficiency was the transverse type (n=112) (Table I).

In the unilateral upper limb transverse deficiencies, the forearm partial (below elbow) deficiency (n=24) was the most common, followed by the carpal total (wrist disarticulation) deficiency[14] (Table II).

In the unilateral upper limb longitudinal type deficiencies, the metacarpophalangeal (Ray) deficiency (n=8) was the most common (Table III).

The longitudinal deficiencies (n=43) in the unilateral lower limbs were more than the transverse deficiencies.[14] The most common deficiency in the transverse type of unilateral lower limb deficiency was the leg partial (below knee) deficiency (n=8) (Table IV).

The most common longitudinal type in the unilateral lower limb deficiency was the femoral partial or proximal femoral focal (PFFD) deficiency (n=29) (Table V).

We found 19 patients suffered of multiple congenital limb deficiency; they had 46-limb deficiencies. One of the patients was a girl with quadriple type deficiency and had bilateral transverse arm totalis and bilateral thigh totalis. The patients with bilateral upper limb involvement were the most common in the multiple congenital limb deficiency, followed by the triple limb deficiency (Table VI).

Table II. Levels of unilateral congenital transverse upper limb deficiency

Level	Number of patients	Male	Female	Right	Left	No. of patients fitted with prosthesis
Arm total (shoulder disarticulation)	1	0	1	0	1	0
Arm partial (above elbow)	2	1	1	1	1	2
Forearm total (elbow disarticulation)	4	2	2	3	1	3
Forearm partial(below elbow)	24	9	15	10	14	20
Carpal total (wrist disarticulation)	14	7	7	6	8	11
Metacarpal phalangeal partial + total	11	5	6	5	6	6
Total	56	24	32	25	31	42 (75%)

Table III. Levels of unilateral congenital longitudinal upper limb deficiencies

Level	No. of patients	Male	Female	Right	left	No. of patients fitted with prosthesis
Humeral partial\ total	0	0	0	0	0	0
Ulnar partial	1	0	1	1	0	1
Radial total	1	0	1	0	1	1
Carpal total	1	1	0	1	0	1
Metacarpo-phalangeal partial (Ray)	8	1	7	5	3	5
Total	11	2	9	7	4	8 (72.5%)

Table IV. Levels of unilateral congenital transverse lower limb deficiencies

Level	No. of Patients	Male	Female	Right	Left	No. of Patients fitted with prosthesis
Thigh total (hip disarticulation)	2	1	1	1	1	1
Thigh partial	0	0	0	0	0	0
Leg total (knee disarticulation)	2	0	2	2	0	2
Leg partial (below knee)	8	3	5	5	3	8
Tarsal total	0	0	0	0	0	0
Metatarso-phalangeal partial	2	0	2	0	2	1
Total	14	4	10	8	6	13

Table V. Levels of unilateral congenital longitudinal lower limb deficiencies

Level	No. of patients	Male	Female	Right	Left	No. of patients fitted with prosthesis
Femoral partial/total (PFFD)	29	15	14	15	14	26
Fibular partial \ total	6	3	3	4	2	2
Tibial partial total	3	1	2	3	0	2
Tarso-metatarso-phalangeal partial \ total	5	2	3	3	2	3
Total	43	21	22	25	18	33 (76%)

Table VI. Distribution of multiple congenital limb deficiencies

Limb involved	No. of Patients	No. of limb deficiency	Male	Female	Right	Left	Transverse	Longitudinal	No. of patients Fitted with prosthesis
Bilateral upper limb	8	16	4	4	8	8	16	0	5
Bilateral lower limb	3	6	2	1	3	3	6	0	2
Double (upper \lower)	2	4	1	1	1	3	3	1	2
Triple limb deficiency	4	12	2	2	6	6	9	3	2
Quadriple (four limb)	2	8	1	1	4	4	8	0	0
Total	19	46	10	9	22	24	42	4	11

Table VII. Prosthetic fitting of patients with congenital limb deficiency

Side of deficiency	No. of patients	No. of fitted patients	%	No. of limb involved	Transverse deficiency	Prosthetic fitting	%	Longitudinal deficiency	Prosthetic fitting	%
Unilateral upper limb	67	50	74.5	67	56	42	75	11	8	72.5
Unilateral lower limb	57	46	81	57	14	13	93	43	33	76
Multiple limbs	19	11	58	46	42	25	59.5	4	2	50
Total	143	107	75	170	112	80	71.5	58	43	74

Table VIII. The suspected incriminated etiological factors in congenital limb deficiency

Etiological factor	Number of patients involved	%
Radiation during pregnancy	9	6.3
Drugs	18	12.6
German measles	2	1.4
Heredity	2	1.4
Cousin marriages	16	11.2
Injury to abdomen, vaginal bleeding, UTI during pregnancy	28	12.6
Subtotal	75	52.5
Non claimed causes	68	47.5
Total	143	100

One hundred and seven (75%) patients with limb deficiency were fitted with prostheses. Fifty-seven (81%) of the unilateral congenital lower limb, 67 (74.5%) of the unilateral congenital upper limb, and 19 (58%) of the multiple congenital limb deficiencies were fitted with prostheses (Table VII).

Seventy five (52.5%) children's mothers claimed exposure to some of the incriminated etiological factors, like exposure to x-ray, drugs, German measles, vaginal bleeding, urinary tract infections etc. (Table VIII).

Discussion

In this study, we reviewed the incidence and demographic characteristics of children with congenital limb deficiency, types, levels and some of the incriminated etiological factors. We also presented the percentage of patients who were fitted with prostheses. The results in this study had similarities and differences with literature. The incidence per thousand of amputees in this study (60 per thousand) was less than a local epidemiological study on amputee population in the RMS-Jordan,[14] and higher than Indian, Japanese, Australian, and

other Western studies which were about 30 per thousand of amputees.[2,3,7,11-13,15]

The increase of the incidence in this study maybe explained by the fact that our centre (RRC) is a referral centre for pediatric orthopedic, plastic surgery and rehabilitation; we receive patients from all regions of the country, which we think increased the number of the congenital limb malformations in our services.

We suggest a comprehensive survey for all newborns in our country for few years to estimate the exact incidence of the limb malformations and other types of congenital malformations in accordance of the international epidemiological studies.

Male children (n=62) were less than female children (n=81) with male to female ratio of 1:1.3. The mean age of females (7.9 years) was higher than male mean age (6.3 years). This presented similarities with local, and Turkish studies and differences with Australian, Japanese, Tunisian, Venezuelans, Indian and Dutch studies.[2,4,5,8,12-14,16,17] We may explain that in our society the parents are more aware for their daughters with congenital malformations than their son's malformations and feel ashamed to bring them from birth for medical consultation but eventually are obligated to face the fact and bring them to the clinic for that the number of girls was more and the girls were older than boys at presentation to the clinic for prosthetic rehabilitation.

The unilateral congenital upper limb deficiencies predominated the unilateral congenital lower limb deficiencies, which presented similarities with Japanese and Indian studies, and differences with Turkish, Venezuelan, and Dutch studies.[2,4-8,13,18]

The transverse type deficiencies were more than the longitudinal type deficiencies. The transverse deficiencies predominated in the unilateral upper limb and in the multiple congenital limb deficiencies, while the longitudinal deficiencies predominated in the unilateral lower limb congenital deficiencies, which had similarity with some studies[2,3,4,8] and difference with other studies.[5,6,13,16,17] The most common transverse level in unilateral upper limb deficiencies was the forearm partial (below elbow) deficiency which was similar to many studies.[2,13,18] The longitudinal metacarpophalangeal (Ray) deficiency was the commonest in the unilateral longitudinal upper limb deficiencies, in contrast with the majority of literatures were the commonest type was the longitudinal radial partial/total deficiency.[17,19] In unilateral congenital lower limb deficiencies the leg partial (below knee) level was the commonest transverse deficiency, which presented similarity with published studies.[7,12,13]

The femur total/partial deficiency presented the majority in the longitudinal unilateral lower limb deficiencies which was similar of the Venezuelan study[17] while the fibular total/partial deficiency was the most common in the majority of the published studies.[2,5,6,13,16,20] This may be explained by that many of the children with fibular total deficiency referred to us by the orthopaedic surgeon after doing a below knee amputation for them, and we considered those as transverse not longitudinal type because of deficit of old information for the patients. We reviewed all patients with leg length discrepancy for compensation, which allowed all patients with proximal femoral focal deficiency to attend the clinic for assessment. This may explain that the femur total/partial deficiencies predominant in the longitudinal lower limb deficiency in our study.

The right side were more than the left side deficiencies, which was similar of some studies[5,6] and different than other studies.[2,18] In the multiple congenital limb deficiencies the bilateral upper limb deficiencies were the commonest followed by the triple limb deficiencies, which was inconsistent with some of the literature.[2,3,13] This maybe explained by the fact that some of those with simple and complicated deformities of the lower limb, didn't attend the prosthetic rehabilitation clinic, and were satisfied with orthopaedic management or didn't appear in both clinics because of family problems and traditions. One hundred and seven children accounted 75% of all limb deficient children who received prostheses. The percentage of unilateral lower limb deficient children who received prostheses (81%) were more than unilateral upper limb (74.5%), inconsistent with many authors who reported that all children with limb deficiency were provided with prostheses, although not all use it. Concerning the use of prostheses, they found that about 30-45% of upper limb deficient children use their prostheses and 69-85% of lower limb deficient children use their prostheses.[2,3,4,6,12,13,18,21] This presented some similarity with our study because we offer the prosthesis for the child who we know he may use it for the lower limb deficient children and we offer the cosmetic type prostheses for the

majority of upper limb deficient children.

In relation to the incriminated etiological factors like x-ray exposure, drugs, heredity etc., we found that 52.5% of children's family claimed exposure to some of these. Nine mothers were exposed to x-ray in the second and third trimester, where no proved limb malformation happen in this period. According to literature, most limb defects develop between the third and eighth post-ovulatory weeks.[21]

Eighteen mothers ingested drugs like antibiotics, antipyretics, non thalidomide antiemetics and tonics with no proved iatrogenic effect.

Sixteen children's parents were relatives and presented traditional cousin marriages. Cousin marriages are not implicated definitely in congenital limb deficiencies in any study apart of a Turkish study in a small number of patients which related close cousin marriages that may increase the longitudinal upper limb and the transverse lower limb deficiencies.[4] Twenty eight children's mothers were exposed to urinary tract infection, vaginal bleeding and mild injury to the abdomen during pregnancy, which were not proven to produce definite congenital limb malformations. Two mothers were exposed to German measles in the second trimester, with no definite effect. Two children, one girl and one boy, had transverse carpal partial deficiency were cousins (the boy's mother was the sister of the girl's father). This raised the possibility of hereditary factor but they hadn't any other associated organic involvement and no other malformations.

Despite the various factors mentioned, no definitive cause could be isolated except for the two cousin children were heredity maybe implicated and this calls for a definitive genetic study.

Conclusion

The most common deficiency in our study was the transverse fore-arm partial in upper limb and the longitudinal femoral partial deficiency in the lower limb. The majority of our congenital limb deficient children were fitted with prostheses although not all use it functionally. No etiological factors definitely could be incriminated in the congenital limb deficiency.

This study was a cornerstone for further studies in the congenital limb deficiency in our country. Furthermore, in may help in future planning and management of material, facilities, budget needs for children with limb deficiency.

References

1. **HJB Day.** The ISO/ISPO classification of congenital limb deficiency. *Prosthet & Orthot Int* 1991; 15: 67-69.
2. **Jain SK.** A study of 200 cases of congenital limb deficiencies. *Prosthet & Orthot Int* 1994; 18:174-179.
3. **Stewart CPU, Jain AS.** Congenital limb anomalies and amputees, Tayside, Scotland 1965-1994. *Prosthet & Orthot Int* 1995; 19: 148-154.
4. **Sener G, Yigiter K, Bayar K, Erbahceci F.** Effectiveness of prosthetic rehabilitation of children with limb deficiencies present at birth. *Prosthet & Orthot Int* 1991; 23: 130-134.
5. **Rijnders LJM, Boonstra AM, Groothoff JW, *et al.*** Lower limb deficient children in Netherlands; epidemiological aspect. *Prosthet & Orthot Int* 2000; 24: 13-18.
6. **Boonstra AM, Rijnders LJ, Groothoff JW, Eisma WH.** Children with congenital deficiencies, or acquired amputations of the lower limbs; functional aspects. *Prosthet & Orthot Int* 2000; 24: 19-27.
7. **Sharma VP.** The care of the limb deficient child in India. *Prosthet & Orthot Int* 1991; 15: 146-151.
8. **Yiğiter K, Ülger Ö, Şener G, Akdoğan S, *et al.*** Demography and function of children with limb loss. *Prsthet & Orthot Int* 2005;29(2): 131-138.
9. **Svoboda J.** Psychological Considerations in Pediatrics; use of amputee dolls. *Prosthet & Orthot Int* 1992; 4: 207-212.
10. **Hirons RR, Williams KB, Amor RF, Day HJB.** The prosthetic treatment of lower limb deficiency. *Prosthet & Orthot Int* 1991; 15: 112-116.
11. **Evans DGR, Thakker Y, Donnai D.** Heredity and dysmorphic syndromes in congenital limb deficiency. *Prosthet & Orthot Int* 1999; 15: 70-77.
12. **Jones LE, Lipson A.** The care of the limb deficient child in Australia. *Prosthet & Orthot Int* 1991; 15: 140-142.
13. **Kakurai S, Kida M.** The care of limb deficient child in Japan. *Prosthet & Orthot Int* 1991; 15: 146-151.
14. **Al-Worikat A.** Amputees referred to the Royal Medical Services-Jordan. *JCRPO Journal* 1996; 2: 174-175.
15. **Vitali M, Robinson KP, Andrews BG, Harris EE.** Amputations and prosthetic London, Bailliere Tindall 1978; 11.
16. **Sliman N, Mrabet A, Daghfous S, Douik M.** The care of limb deficient child in North Africa. *Prosthet & Orthot Int* 1991; 15: 152-155.
17. **Fernandez–Palazzi F, De Gutierrez DP, Paladino R.** The care of the limb deficient child in Venzuela. *Prosthet & Orthot Int* 1991; 15: 156-159.
18. **Kuyper MA, Breedijk M, Mulders AHM, *et al.*** Management of children in the Netherlands with upper limb deficiencies. *Prosthet & Orthot Int* 2001; 25: 228-234.
19. **Lamb DW.** The treatment of longitudinal radial deficiency. *Prosthet & Orthot Int* 1991; 15: 100-103.
20. **Shatilov OE, Rozkov AV, Cheminova TV.** Reconstructive surgery for fibular deficiency. *Prosthet & Orthot Int* 1991; 15: 137-139.
21. **Jones, LE.** Prosthetic limb use in Australia 1981-1985 under the free limb scheme. *Prosthet & Orthot Int* 1989; 13: 76-81.
22. **Swanson AB.** congenital limb defects- classification treatment. *Clin Symp* 1981; 33(3): 3-32.