Prosthetics and Orthotics International, 1994, 18, 174–179

A study of 200 cases of congenital limb deficiencies

S. K. JAIN

Artificial Limb Centre, Pune, India

Abstract

An analysis of 200 patients with congenital limb deficiency who attended the Artificial Limb Centre, Pune from January 1984 to April 1990 is presented. This group is representative of the congenital limb deficient population of the country. The commonest deficiencies were transverse phalangeal total/partial deficiency and transverse forearm partial deficiency (below elbow) in upper limbs, whereas transverse metatarsal total/partial deficiency and transverse leg partial deficiency (below knee) were commonest in lower limbs. Transverse forearm partial deficiency was more common in female, while transverse leg partial deficiency was more common in male children, 16 patients did not require any treatment, 6 needed only surgical correction. Some 30 patients needed surgery before prosthetic fitting, while 148 patients required only prostheses. Some 68% of patients achieved satisfactory to excellent results; 18% showed poor rehabilitation. No definitive cause for the deformities could be isolated; however, many parents believed that possible exposure to the eclipse during pregnancy was the cause of the deficiency. The eldest child was most affected.

Introduction

The Artificial Limb Centre in Pune, one of the biggest in the country and with its own kind of rehabilitation centre, attracts limb deficient patients from all parts of the country. Therefore the limb deficient children attending this Centre can easily be accepted as a representative sample of the total congenital limb deficient population of India.

All correspondence to be addressed to Colonel S K Jain, Artificial Limb Centre, Pune 411 040, India.

Material and methods

This study includes 200 consecutive patients with congenital limb deficiency, who came for treatment to the Artificial Limb Centre, Pune from January 1984 to April 1990.

Every patient was examined in detail and deficiencies recorded. Special care was taken to elicit history of maternal illness, consumption of drugs, exposure to radiation as well as hyperemesis, foetal trauma, vaccination, smoking and alcoholic habits of expectant mothers during pregnancy. Other factors such as the socio-economic status of the parents, family history, position of child in the family tree and history of any other sibling or close relative similarly affected, which could possibly throw light on the cause of the limb deficiency were also examined.

Patients requiring any surgical intervention before the prosthetic fitting were identified. Surgery was carried out wherever necessary and a prosthesis provided. The state of rehabilitation was assessed as excellent, good, satisfactory, or poor, based upon functional achievement with the help of a prosthesis. The group of patients which did not require any treatment surgical or prosthetic was also identified.

Observations and discussion

Incidence

The 200 congenital limb deficient patients who visited this Centre were from a total number of 5375 amputees making the incidence 37 per thousand amputees. In western literature the incidence is 30 per thousand (Vitali *et al.*, 1988).

Table 1. Associated causative factors

Causative factors (probable)	No of patients
Drugs	19
Previous abortions	8
Previous premature births	3
Previous Caesarean births	4
Injury to the abdomen during pregnancy	4
Radiation during pregnancy	4
Heredity	1
Exposure to eclipse	19
Total	62

Sex incidence

There were 130 (65%) male and 70 (35%) female patients (ratio 13:7), while the comparative sex incidence in various other studies (Lamber, 1971; Agarwal *et al.*, 1986) show this ratio to be 13:12.

Age at the time of reporting for treatment

It was seen that the number of males outnumbered females in all age groups except the 10-15 years age group, where the number of girls was more than the boys; maybe they and their parents became more conscious of the disability at this age.

Birth serial of the patients

The position of the child in the family lineage was recorded. It was seen that 82 (41%) of patients were the eldest in the family. Contribution of first 3 children was 84%.

Aetiological factors

An attempt was made to discover the exact cause which might have produced the disability. In only 62 patients was some sort of history noted which could probably be correlated with the limb deficiency, as shown in Table 1.

Most limb defects developed between the third and eight post-ovulatory week (Kenedy, 1967; Swanson, 1981) and only a few parents were aware of the pregnancy till late in the third month of gestation; fewer still could remember an intake of any drug or an illness suffered.

The noticeable finding was a history of previous abortions in 8 cases. This may indicate some kind of placental insufficiency which may have caused the previous abortions as well as the deficiency in the new born. Only 19 mothers could remember having taken drugs namely antiemetics, antibiotics, antispasmodics and antidepressants during the pregnancy. None of these drugs is specifically known to have been a cause of limb deficiencies (The pharmacological basis of therapeutics, 1985). However 168 mothers gave a history of taking haematenics and multivitamins during pregnancy.

An interesting finding was a history of exposure of expectant mother to the eclipse during pregnancy in 19 cases. Though no documentary evidence exists to correlate it with the deficiency in the literature, still many parents believe this to be the cause of limb deficiency in the new born.

Despite the various factors mentioned, no definite cause could be isolated except in one case where the new born and the mother both had deficient/weak thumbs in both hands. The cause was perhaps genetic. However in this case no one else in the family was affected.

Previous treatment

Only 18 patients had received some treatment; 15 had undergone some surgical treatment and only 8 patients had received prostheses. This indicates ignorance or the lack of facilities to deal with limb deficient children,

Deficiencies

It is not practically possible to classify or group all the deficiencies encountered. However the simple classification of deficiencies followed earlier by the author (Jain *et al.*, 1989) has not been used in this article. It has been changed to conform to the International Standard Organisation (ISO) Classification (Day, 1991) as shown in Figures 1 and 2 for upper limbs and lower limbs respectively. This classification is descriptive and can be used under most circumstances.

The deficiencies are classified into two basic categories.

1. *Transverse Deficiency*: This resembles an amputated limb where the limb has developed normally up to a particular level and beyond which no bony element is present. Transverse deficiencies are:

(a) Transverse upper limb deficiencies:
   (i) phalangeal total/partial deficiency;
   (ii) metacarpal total/partial deficiency;
   (iii) carpal total/partial deficiency;
   (iv) forearm total/partial deficiency;
   (v) upper arm total/partial deficiency;
   (vi) shoulder total/partial deficiency.

(b) Transverse lower limb deficiencies:
- (i) phalangeal total/partial deficiency;
- (ii) metatarsal total/partial deficiency;
- (iii) tarsal total/partial deficiency;
- (iv) leg total/partial deficiency;
- (v) thigh total/partial deficiency;
- (vi) pelvis total/partial deficiency.

2. *Longitudinal Deficiency*: All other cases where an element or elements within the long axis of the limb is/are reduced or absent, are grouped in longitudinal deficiency.

(a) Longitudinal upper limb deficiencies:
- (i) radius total carpus partial ray 1 total deficiency;
- (ii) radius total/partial deficiency;
- (iii) ulna total carpus partial rays 2, 3, 4, 5 total deficiency;
- (iv) ulna total/partial deficiency;
- (v) radius ulna total/partial deficiency;
- (vi) humerus total deficiency;
- (vii) humerus total radius ulna total/partial deficiency.

(b) Longitudinal lower limb deficiencies:
- (i) tibia total tarsus partial ray 1 total deficiency;
- (ii) tibia total/partial deficiency;
- (iii) fibula total tarsus partial rays 2, 3, 4, 5 total deficiency;
- (iv) fibula total/partial deficiency;
- (v) tibia fibula total/partial deficiency;
- (vi) femur total/partial deficiency;
- (vii) femur total tibia fibula total/partial deficiency.

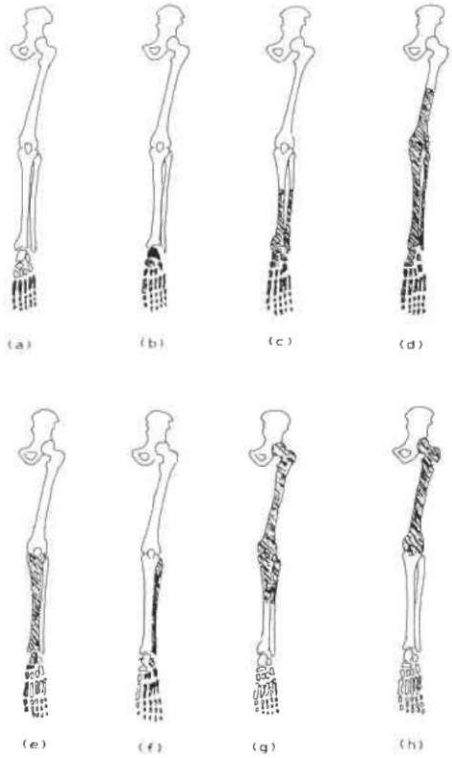

Fig. 1. Congenital upper limb deficiencies.
(a) Phalangeal total deficiency
(b) Carpal partial deficiency
(c) Fore arm partial deficiency
(d) Upper arm partial deficiency
(e) Radius total carpus partial ray 1 total deficiency
(f) Ulna total carpus partial rays 2, 3, 4, 5 total deficiency
(g) Radius total deficiency
(h) Ulna total deficiency

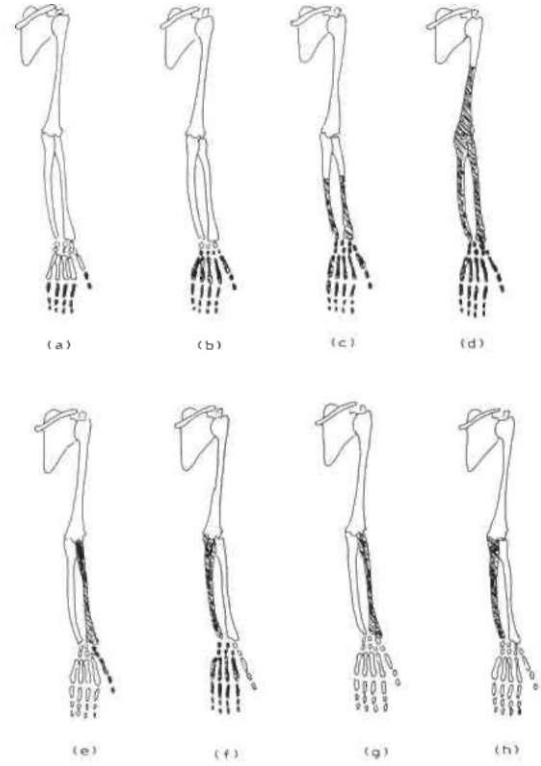

Fig. 2. Congenital lower limb deficiencies.
(a) Metatarsal total deficiency
(b) Tarsal total deficiency
(c) Leg partial deficiency
(d) Thigh partial deficiency
(e) Tibia total tarsus partial ray 1 total deficiency
(f) Fibula total tarsus partial rays 2, 3, 4, 5 total deficiency
(g) Femur total tibia fibula partial deficiency
(h) Femur total deficiency

Deficiency in upper limbs

Table 2 shows deficiencies observed in the upper limbs. It was interesting to note that the most common deficiency was transverse forearm partial deficiency (below elbow), found in 57 patients (2 bilateral cases). Some 34 (60%) were females and 23 (40%) were males. The left side was a little more affected than the right side. Transverse phalangeal deficiency was seen in 23 patients (14 single-sided and 9 bilateral), where 15 (65%) were males and 8 (35%) were females.

Table 2: Upper limb deficiencies

Deficiencies	Rt	Lt	Bil	No of patients	No of limbs
Transverse deficiency					
Phalangeal total/partial deficiency	7	7	9	23	32
Metacarpal total/partial deficiency	2	6	–	8	8
Carpal partial deficiency	7	5	–	12	12
Carpal total deficiency	2	4	–	6	6
Forearm partial deficiency	23	32	2	57	59
Forearm total deficiency	3	2	–	5	5
Upper arm total/partial deficiency	4	5	–	9	9
Shoulder total/partial deficiency	1	–	–	1	1
Longitudinal deficiency					
Radial total carpal partial ray 1 total deficiency	2	–	–	2	2
Radius total/partial deficiency	1	–	–	1	1
Ulnar total carpal partial rays 2, 3, 4, 5 total deficiency	–	2	3	5	8
Ulna total/partial deficiency	–	1	1	2	3
Radius ulna total/partial deficiency	–	–	–	–	–
Humerus total deficiency	–	–	–	–	–
Humerus total radius ulna total/partial deficiency	–	1	–	1	1
Total	52	65	15	132	147

Table 3: Lower limb deficiencies

Deficiencies	Rt	Lt	Bil	No of patients	No of limbs
Transverse Deficiency					
Phalangeal total/partial deficiency	2	–	–	2	2
Metatarsal total/partial deficiency	4	12	6	22	28
Tarsal partial deficiency	1	3	–	4	4
Tarsal total deficiency	2	2	5	9	14
Leg partial deficiency	10	11	–	21	21
Leg total deficiency	–	2	–	2	2
Thigh total/partial deficiency	4	–	–	4	4
Pelvis total/partial deficiency	–	–	–	–	–
Longitudinal deficiency					
Tibial total tarsal partial ray 1 total deficiency	3	–	–	3	3
Tibia total/partial deficiency	–	–	–	–	–
Fibular total tarsal partial rays 2, 3, 4, 5 total deficiency	2	6	2	10	12
Fibula total/partial deficiency	1	–	–	1	1
Tibia fibula total/partial deficiency	–	3	–	3	3
Femur total/partial deficiency	–	5	–	5	5
Femur total tibia fibula total/ partial deficiency	6	5	–	11	11
Shortening					
Tibiofibular	4	7	–	11	11
Femoral Tibiofibular	5	2	–	7	7
Total	44	58	13	115	128

Table 4: Distribution of deficiencies

Limbs involved	No of patients	Upper limb	Lower limb
One upper limb	84	84	–
One lower limb	59	–	59
Bilateral upper limbs	17	34	–
Bilateral lower limbs	20	–	40
One upper and one lower limbs	8	8	8
Three limbs	6	9	9
Four limbs	6	12	12
Total	200	147	128

Deficiency in lower limb

Table 3 shows the deficiencies in the lower limbs. The commonest deficiency was transverse metatarsal deficiency in 22 patients (6 bilateral). Another common deformity was transverse leg partial deficiency (below knee). It was found in 21 patients (no bilateral case) of which 13 (62%) were males and 8 (39%) were females.

Distribution of deficiencies

More than one limb was involved in 57 patients. Two limbs were involved in 45 patients (22.5%) three in 6 (3%) and all four in 6 patients (3%). In the study by Kay (1974) the percentage of two, three and four limbs was 15, 5 and 10 respectively. Details of the distribution of the deficiency are shown in Table 4. Involvement of the upper limb was more common than involvement of the lower limb in single limb deficiency, while bilateral deficiency was more common in lower limbs.

Associated defects

Some 21 patients were found to have various associated defects as shown in Table 5. Constriction ring was the commonest defect noticed, which could be the cause of the limb deficiency. Three children with talipes equinovarus deformity had deficiency in the upper limbs.

Table 5. Associated defects

Defects	No of patients
Constriction rings	8
Talipes equinovarus	3
Syndactyly	6
Spina bifida	1
Dislocation of hip	1
Craniostenosis	1
Absent pectoralis major and minor	1
Total	21

Management

Of the 200 patients in this study 16 patients did not require any treatment, 6 patients needed only surgical correction of deformity while 30 patients required surgical treatment such as amputation and the release of a constriction ring before a prosthesis was fitted. Surgery was performed only in the lower limbs. Some 148 patients required no other treatment except prosthetic fitting.

Prostheses were provided in 178 cases. The commonest upper limb prosthesis was trans-radial (57 cases) while in lower limbs it was trans-tibial (21 cases). Extension prostheses for shortening of the lower limb was given to 25 patients.

Rehabilitation status

The rehabilitation status was assessed not only in respect of function achieved but also in appearance which had a definitive role in improving self-confidence. The results are shown in Table 6.

Satisfactory to excellent results were achieved in 68% of cases, while in 18% of cases rehabilitation was poor, mainly representing patients with upper limb deficiency, since rehabilitation is difficult in upper limb deficiency as a rule. These results are similar to the study carried out by Jain *et al.* (1989).

Table 6. State of rehabilitation

	Limb involved				
State of rehabilitation	Upper	%	Lower	%	Overall
Excellent	8	5%	30	23%	14%
Good	36	24%	60	48%	36%
Satisfactory	44	30%	8	6%	18%
Poor	34	23%	15	12%	18%
Not known	13	9%	3	2%	5%
Not relevant	12	9%	12	9%	9%
Total	147		128		

REFERENCES

AGARWAL AK, SHARMA VP, SHARMA RK, VERMA S (1986). Congenital limb deficiencies; proceedings of the XIV Annual Conference *Indian Assoc Paediatr Med Rehab,* p56–63.

DAY HJB (1991). The ISO/ISPO classification of congenital limb deficiency. *Prosthet Orthot Int* **15,** 67-69.

JAIN SK, SANYAL NC, POONEKAR PD, RAVISHANKAR R, HIRWANI S (1989). A study of 100 cases of congenital limb deficiencies. *Med J Armed Forces India* **45,** 77-83.

KAY HW (1974). A proposed international terminology for the classification of congenital limb deficiencies. *ICIB* **13(7),** 1-16.

KENNEDY W (1967). Epidemiological aspects of the problem of congenital malformations. *Birth Defects* **31(1),** 22-25.

LAMBERT CN, PELLICORE RJ, HAMILTON RC, SCIORA J, BIRCH A (1971). Twenty-three years of clinic experience. *ICIB* **15 (3/4),** 15-20.

The pharmacological basis of therapeutics. (1975)./ edited by LS GOODMAN, A GILMAN./5th edition. – London: Macmillan Publications.

SWANSON AB (1981). Congenital limb defects – Classification treatment. *Clin Symp* **33(3),** 3-32.

VITALI M, ROBINSON KP, ANDREWS BG, HARRIS EE (1978). Amputations and Prostheses. – London: Bailliere Tindall. p11.

ERRATUM

The address for correspondence for the paper *A new modular six-bar linkage trans-femoral prosthesis for walking and squatting* by J. K. Chakraborty and K. M. Patil published in the last issue of Prosthetics and Orthotics International was incorrect. All correspondence concerning this article should be addressed to

Professor K. M. Patil
Department of Applied Mechanics
Indian Institute of Technology
Madras 600 036
INDIA