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Congenital limb anomalies and amputees Tayside, Scotland 1965-1994

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Abstract

The purpose of this study was to review the 68 patients who had been referred to Dundee Limb Fitting Centre during the period 1965-1994, with a congenital anomaly of a major limb requiring prosthetic replacement. A profile of the incidence of congenital anomalies, amputation levels and prosthetic fitting was obtained. During the period only 68 cases with 80 congenital anomalies were referred. During these 29 years, 20 cases required surgical amputation and overall 35 surgical procedures were performed in these cases, only 3 were in the upper limb. The incidence of upper and lower limb deficiency was similar. The patients represented a small proportion (1.6%) of the patients who were reported to have congenital anomalies. Figures indicated that about 8% of all live/still births have some form of anomaly. Prosthetic fitting and use was successful in all 68 cases but long term life follow-up is necessary to ensure continued prosthetic use.

Introduction

The incidence of limb deficiency, that is absence of part of a limb, present at birth has been variously reported as being 1:4264 in Canada (McDonnell, 1988) and 5:10,000 in Australia (Jones and Lipson, 1991).

Scottish figures from the Common Services Agency (CSA) of the Scottish Office report rates of 130:10,000 for 1988 (Scottish Health Service Common Services Agency, 1991) and 120:10,000 for 1989 (Scottish Health Service Common Services Agency, 1992) for all congenital anomalies. Tayside, Scotland, with a population of 400,000 inhabitants, had a high incidence of limb anomaly (limb absence or deficiency) at 3.1% (310:10,000) of all live/still births in 1988 and 3.3% (330:10,000) in 1989. This incidence of limb anomalies is much higher than previously reported. Care must be taken when comparing results of one centre with another since incidence of reporting may vary significantly from place to place.

However, in a 29 year period only 68 patients were referred for prosthetic fitting to Dundee Limb Fitting Centre (DLFC) which caters for the entire Tayside and North Fife area. All 68 patients were successfully fitted with a prosthesis (Tables 1, 2, 3, 4, 5 and 6).

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Method

The records of all patients who attended DLFC since its establishment in 1965 were reviewed. The centre maintains its own medical records and stores the files on site, retaining the records of those patients who have died or left the area. No files are destroyed.

Those patients who had undergone an amputation or had a limb deficiency at birth were identified and their records scrutinised. The level of amputation was defined using the nomenclature described by Day (1988) and now published as an international standard (International Standards Organisation, 1989).

Results

Table1 lists the levels of amputation in those with limb deficiency present at birth. There were 80 congenital limb anomalies in the 68 patients referred for prosthetic fitting and these were equally divided between upper (40 anomalies) and lower limb (40 anomalies). Fourteen patients had mixed limb anomalies and 10 patients had other anomalies.

Table 1. Congenital cases requiring a prosthesis presented at DLFC 1965-94 (N = 68)

Level of prosthetic fitting	Limb anomalies	Surgical amputation	Deceased
Wrist disarticulation (WD)	1		
Partial hand (PH)	13		1
Trans-radial (TR)	21	1	2
Elbow disarticulation (ED)	1		
Trans-humeral (TH)	4		
Ankle disarticulation (AD)	20	20	1
Trans-tibial (TT)	8	7	
Knee disarticulation (KD)	5	1	
Trans-femoral (TF)	5	4	
Hip disarticulation (HD)	2	2	
			1 case with clubhand
Total	80	35	5

Table 2 lists levels of prosthetic fitting on patients with a one sided anomaly of the lower limb. All 8 patients required a surgical amputation and 7 were subsequently fitted with an ankle disarticulation prosthesis and 1 with a trans-femoral prosthesis.

Table 3 lists the level of prosthetic fitting on patients with a one sided anomaly of the upper limb. Of the 32 patients 8 were fitted with a partial hand (PH) prosthesis, 1 with a wrist disarticulation (WD) prosthesis, 20 with a trans-radial (TR) prosthesis, 1 with an elbow disarticulation (ED) prosthesis and 2 with a trans-humeral (TH) prosthesis.

There were 14 cases of patients with multiple deficiency of which 1 was bilateral upper limb, 6 upper and lower limb, 4 bilateral lower limb and 3 mixed anomaly. Fifteen surgical amputations were carried out on 11 of the patients. Twenty-six prostheses were fitted (Table 4). The details of the individual patients are outlined in Table 5.

Table 6 lists the 14 patients with limb anomalies with a specific diagnosis. Twelve surgical amputations were carried out on 10 of the patients.

In Tayside in 1988 there were a total of 4850 live and still births. Table 7 gives information with regards congenital anomalies reported in Tayside in that year (Scottish Health Service Common Services Agency, 1991).

In Scotland as a whole there were 66,569 live and still births in 1988 with 5,974 having some form of anomaly. Table 8 lists information about congenital anomalies reported in Scotland in that year (Scottish Health Service Common

Table 2. Level of prosthetic fitting on patients* with a one sided anomaly of the lower limb (N=8)

Prosthetic fitting	Anomaly	Number of cases
AD	Clubfoot	2
	Metatarsal dysplasia	1
	Longitudinal femur partial	1
	Longitudinal fibula total	2
	Longitudinal fibula total, tarsus partial, rays 4 + 5 total	1
TF	Hypoplastic limb	1

*All patients had surgical amputation.

Table 3. Level of prosthetic fitting on patients with a one sided anomaly of the upper limb (N=32)

Prosthetic fitting	Anomaly	Number of cases
PH	*Longitudinal*	
	Ulna total, carpus partial, metacarpals/phalanges partial.	1
	Radius partial, carpus total, 1-3 metacarsal/phalanges partial.	1
	Metacarpals 4+5 total, 3rd phalanges total.	1
	Transverse	
	Carpus partial.	3
	Phalanges partial.	2
WD	Longitudinal carpus partial 2-4 rays total.	1
TR	*Longitudinal*	
	Radius total, ulna partial, carpus total, phalanges partial.	2
	Radius total, ulna partial, carpus total.	1
	Radius partial, carpus total, metacarpal total, 1-4 phalanges total, 5 phalanges partial.	1
	Transverse	
	Radius partial, ulna partial carpus total.	2
	Forearm upper.	3
	Forearm middle (with bud).	1
	Forearm middle.*	1
	Forearm middle.	4
	Forearm lower.	3
	Carpus partial.	2
ED	Forearm total.	1
TH	Arm middle.	2

*Case requiring surgical amputations

Table 4. Level of prosthetic fitting on patients (N=14) with multiple deficiency

Prosthetic fitting	Number of fittings
Ankle disarticulation (AD)	9
Trans-tibial (TT)	4
Knee-disarticulation (KD)	4
Trans-femoral (TF)	1
Partial hand (PH)	5
Trans-humeral (TH)	2
Trans-radial (TR)	1
Total	26

Services Agency, 1991).

Table 9 lists surgical events other than the amputation performed on 15 patients. All told there were a further 30 surgical events.

Table 10 lists incidence of the surgical events in the 68 cases.

Discussion

The incidence of limb anomaly has previously been reported as 7:10,000 in Japan (Kakurai and Kida, 1991), 5:10,000 in Australia (Jones and Lipson, 1991) and a similar rate in the United Kingdom (Evans *et.al.*, 1991)

The overall incidence of anomaly as reported by the Scottish Health Service Common Services Agency (1991 and 1992) for 1988 and 1989 were much higher with rates of 8.97% and 7.47% respectively (900:10,000 and 750:10,000) but these figures included all anomalies not only limbs. The limb anomalies rate was 1.3% of all live/still births (130:10,000) but only a relatively small number are referred for prosthetic management.

Tayside figures are the highest reported by the CSA with 3.1% (310:10,000) of all live/still births in 1988 having some limb anomaly (Table 7). There were 4,850 live/still births in 1988 resulting in 150 babies having a limb anomaly. It was surprising that only one of these 150 cases was referred for prosthetic supply.

These figures are much higher than previously reported and thus of considerable interest (Table 8). Previously in the UK the reported figure was 1:4400 for both congenital and acquired amputations (2.3:10,000) (Evans *et al.*, 1991). Similarly a review by Rogola *et al* (1974) of 52,000 live births in Scotland reported a rate of 1:3000 for congenital upper and lower limb amputation (3.3:10,000).

The CSA in their reports of 1988 and 1989 drew attention to the difficulties in comparing one statistic to another from different studies. This is mainly due to different degrees of anomaly being reported. The CSA results are drawn from information obtained from a form

Table 5. Multiple cases (no specific diagnosis) indicating level of prosthetic fitting.

1.	(a)	KD	Transverse leg total.
	(b)	KD	Transverse leg total.
2.	(a)	KD	Transverse leg total.
	(b)	KD	Transverse leg total, polydactaly
3.	(a)	PH	Longitudinal 3-4 metacarpal partial, phalanges partial.
	(b)*	AD	Longitudinal fibula total, tarsal/metatarsal/phalanges total.
	(c)		Longitudinal fibula total, tarsus partial, 4 and 5 rays total.
	(d)		Breast different sizes.
4.	(a)	TH	Transverse arm middle 1/3rd.
	(b)	TH	Transverse arm middle 1/3rd.
5.	(a)*	AD	Longitudinal tarsus partial, 3, 4 and 5 metatarsal/phalanges partial.
	(b)*	AD	Longitudinal tarsus partial, 3, 4 and 5 metatarsal/phalanges partial.
6.	(a)*	AD	Longitudinal fibula total, tarsus partial, 4 and 5 rays total.
	(b)		Ilium abnormal, longitudinal ulna total, carpus partial 4 and 5 rays total.
7.	(a)*	TT	Longitudinal metatarsal partial, 3, 4 and 5 rays total.
	(b)*	TT	Longitudinal metatarsal partial 3, 4 and 5 rays total.
	(c)		Pelvic dysplasia.
8.	(a)	PH	Transverse carpus total.
	(b)	PH	Longitudinal carpus partial.
	(c)*	AD	Longitudinal 2, 3 and 4 metatarsals partial.
9.	(a)		Deformed foot – dysplasia of fibula, longitudinal tibial partial, metatarsal fusion 2 and 3.
	(b)*	AD	Longitudinal tibia total.
	(c)	PH	Transverse carpus total.
	(d)	PH	Longitudinal metacarpal 4 and 5 rays total.
10.	(a)		Clubhand. Longitudinal radius total, carpus partial, 4 and 5 rays total.
	(b)*	AD	Clubfoot. Converted to AD.
11.	(a)*	AD	Clubfoot.
	(b)*	TF	Clubfoot initial amputation AD.
12.	(a)*	AD	Dysplastic femur, fibula total.
	(b)*	TR	Transverse forearm middle.
13.	(a)*	TT	Clubfoot.
	(b)		Dysplastic radius.
14	(a)*	TT	Dysplastic limb.
	(b)		Hand polydactaly.

* Surgical amputations performed.

Table 6. Cases with a specific diagnosis (N = 14).

	Diagnosis	Deformity	Level of prosthetic fitting
1.	Coxa vara	Short limb	Inclusion prosthesis
2.	Spina bifida	Neuropathy and foot deformity	TT*
3.	Spina bifida	Scoliosis and foot deformity	AD*
4.	Spina bifida	Shortened limb and foot deformity	TF*
5.	Spina bifida	Paralytic dislocated hip	HD*
6.	Spina bifida	Avascular necrosis of talus	TT*
7.	Chromosomal 16	Abnormality	KD
8.	Neurofibromatous		TT
9.	Femoral atresia	Umbilical cellulitis	TF*
10.	Scleroderma	Abnormal ilium	HD*
11.	VATER association	Transverse leg total Abnormal pelvis, spinal and renal abnormalities, anal artresia	TF
12.	Arthrogryphosis multiplex	Bilateral clubfeet	AD* TT*
13.	Drug induced (Thalidamide)	Limb dysplasia	KD*
14.	Drug induced (debendox)	Transverse tarsus total Transverse tarsus total (webbed hand, carpus metacarpal fusion)	AD* AD*

* Surgical amputations performed.

(SMR 11) which is completed by clinicians when an anomaly is noted, using the Manual of the International Classification of Diseases, Injuries and Cause of Death (1977). Minor and major anomalies are recorded on this form, therefore, widely differing report rates have emerged. "The overall congenital anomaly rate varies greatly between Boards (Health Boards) almost entirely because of the differences in noting minor anomalies such as skin tags etc" (Scottish Health Service Common Services Agency, 1991).

Historically no universally acceptable nomenclature of limb deficiency has been produced. Franz and O'Rahilly (1961) devised a widely accepted classification, but other also appear in various publications which creates difficulty when comparing results.

Table 7. Congenital anomalies in Tayside in 1988 from a total of 4850 live and still births as reported by Scottish Health Service Common Services Agency (1991).

Anomaly	Number
Some anomaly	801
Spina bifida	4
Polydactyly	4
Syndactyly	6
Anomalies of feet	135
Reduction of limb	1

Table 8. Congenital anomalies in Scotland in 1988 as reported by Scottish Health Service Common Service Agency (1991).

Anamolies	Live births	Still births	Rates of anomalies/10,000 live/still births			
			Live births		Still births	
			Male	Female	Male	Female
Anomalies of feet	664	2	91.6	109.5	–	117
Polydactyly	55	1	7.9	8.7	53.8	–
Syndactyly	66	4	11.7	8.1	107.5	117
Reduction of deformity	19	3	4.1	1.6	161.3	–
Other anomalies of limbs	206	6	33.5	28.6	215.1	117
Multiple congenital anomalies	41	2	6.8	5.6	53.8	58.5

Table 9. Surgical events other than single amputation (N = 30).

Problem	Level of amputation or absence	Number of operations
1. Clubfoot, radial Clubhand	AD	1 Rotation osteotomy of femur.
2. Spina bifida	KD	2 Osteotomy femur, bone graft.
3. Bilateral "deformed" feet, left hand has finger buds	AD	3 Fusions of 2, 3, 4, metatarsals. Revision of stump.
4. Spina bifida	TT	2 Multiple foot operations.
5. Radial clubhand, partial radius, scoliosis sublaxation of patella	TT	1 Hip replacement
6. Spina bifida, aplastic hip, neuropathy and foot deformity	TT	3 Multiple revisions to stump
7. Spina bifida, deformed feet	AD	1 Valgus tibia osteotomy.
8. Arthrogryphosis multiplex, clubfeet	AD x 2 (later AD/TT)	3 Stump level revision. Bilateral hip replacements.
9. Scoliosis, limb shortening	TF	1 Stump level revision.
10. Fibula absence	AD	5 Tibial osteotomy x 3. Femoral osteotomy x 1. Lateral muscle release.
11. Clubfoot	AD	3 Arthrodesis, tendon transplant. Amputation Neuroma excised.
12. Syndactyly (hands and feet)	Bilateral AD	2 Ankle disarticulations x 2. Hand operations.
13. Fibula loss "flipper" arm	AD	1 Ankle disarticulation and pollicization.
14. "Abnormal" leg (Triphalyngeal thumbs)	TT	1 Tibia ankylosed to femur
15. Absent fibula	AD	1 Ankle disarticulation. Tibial osteotomy.

* Surgical amputations performed.

O'Rahilly (1971) emphasized the need for an internationally accepted nomenclature. Day's (1988) nomenclature dispensed with ambiguous terms such as "amelia" and "macromelia". Clear terms such as "transverse", "longitudinal", "partial" and "complete", have been described and accepted as the Standard by the International Standards Organization (1989).

Table 10. Summary of surgical events.

Of 68 patients, 35 (50%) had surgical amputations and 3 had bilateral surgical amputations (in total there were 64 surgical events, 1.1 per patient).
Of 20 patients who had an AD prosthesis fitted, 4 (20%) had an osteotomy, 5 of the tibia, 2 of the femur. There were 21 surgical events (one had 5 performed).
Of these patients only 3 had upper limb surgery, 3 of 35 (8.6%).
Of these 5 cases with spina bifida, 5 (100%) had a surgical amputation performed on the lower limb, in addition 2 had a varus osteotomy of the tibia and 2 had surgical amputation at trans-tibial level, one at trans-femoral.

During the 29 year period reported here only 68 patients were referred although one can presume that there were an approximate 4,350 cases of limb anomalies (i.e. 150 x 29). Only 14 of these have multiple anomalies with 15 amputations in 11 patients (Table 4 and Fig. 1).

Thirty-five (50%) required surgical amputation. The 35 who had surgery had 64 surgical procedures (1.1 per patient) but only 3 had upper limb surgery and this was pollicization of the fingers in two cases. These are listed in Table 9. It was found that the commonest operation was an osteotomy to correct limb alignment.

Fig. 1. Child with transverse deficiency at right radius and ulna level.

Kakurai and Kida (1991) reported that in a children's hospital where 101 upper limb amputees were presented. 6 (5.9%) required a surgical procedure whereas 44 of the 87 lower limb amputees (50.6%) had some surgical procedure. These figures are different to those of DLFC where 34 of the 38 lower limb amputees (85%) and 2 of the 40 (2.5%) with upper limb anomalies required surgery (Table 9).

The incidence of abnormal limbs has been reported by O'Rahilly (1971) as being commonest in fibula, femur, tibia, ulna and lastly humerus. Coventry and Johnson (1952) reported that the fibula was the most common deficit.

In keeping with other studies, Williams (1962) reported that fibular loss was the most common, the classical picture being one of a short limb with tibial bowing (skin dimple over the point of greatest angulation) with later delays in ossification at both ends of the tibia (Fig 2). Osteotomy may be required to correct

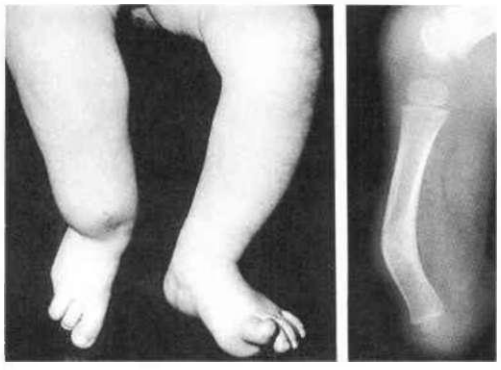

Fig. 2. (a) Child with fibular loss and tibial bowing (skin dimple over the point of greatest angulation).

(b) Delayed ossification at both ends of the tibia.

this bowing and in the period of the present study 7 such surgical procedures were performed in 4 patients, 5 in the tibia and 2 in the femur. Prosthetic fitting was achieved in all these cases.

It is important to remember that the group in the present study was highly selected as the patients were only those referred to the Centre for prosthetic fitting. In this group carpal anomaly was the most common congenital deficiency with ulna and radius being the next most common. Fibular deficiency was specifically present in 7 of the 68 cases (10%).

Humeral deficiency was less common in this group (2 cases: 3%) which was lower than O'Rahilly's (1971) figures. It is interesting that the CSA figures for Tayside, however, show that in live/still births the most common anomaly is in the foot (Table 7).

A specific diagnosis relating to the congenital problem could only be made in a small number of cases (4) (Table 6). Spina bifida was recorded in 5 cases the levels being 1 ankle disarticulation, 2 trans-tibial, 1 trans-femoral and 1 hip disarticulation. All of the five cases (100%) required surgery (Table 10). All were fitted satisfactorily with a prosthesis.

One other case was described as having an "abnormal limb" and was unable to recall why a trans-tibial amputation had been performed.

The current policy is to fit children with upper and lower limb deficiency at approximately one year old. This allows limb use to coincide with the normal developmental milestone. On average a three-monthly review is carried out with prosthetic replacement as and when necessary. Once skeletal maturity has been reached, attendance for review is as required, usually at the patient's own request. From the records it would appear that at this stage the prosthesis lasts approximately 2 years before a replacement is required.

Four children had been fitted with myoelectric prostheses for upper limb deficiency and the prosthetic follow-up has been carried out at the Scottish National Centre in Edinburgh. All have continued to use their prostheses but one patient is only an occasional user.

In 2 cases an inclusion prosthesis was used, one with coxa vara who had a short limb and one who had muscle fibrosis. Neither had an amputation but were considered as prosthetic

cases as they had anomalies requiring a prosthesis for ambulation.

Five of the 68 patients have died, 20 have left the area and 43 currently attend on a regular basis.

This review shows that only a small number of patients with limb deficiency at birth are referred for prosthetic fitting. Those patients who are referred may require surgery during their life time although functional prosthetic fitting has been successful in all those referred.

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