

Laterality, gender and age differences in estimated frequency and actual registration of people with congenital upper limb absences

C. M. FRASER

Occupational Therapy Services, Addenbrooke's NHS Trust, Cambridge, UK

Abstract

Initial analyses from a survey of people with unilateral upper limb congenital absence registered with the Cambridge Disablement Services Centre (DSC) indicated differences related to laterality and gender. A postal survey of all DSCs in the UK was conducted and support for these findings was provided from the analysis of the information supplied by the 25 DSCs who could provide data in the format requested. Comparing statistics for the UK population with those gained from the 25 DSCs, estimates for the number of children and adults who should be registered with DSCs in the UK are made. From these figures it is suggested that the non-registration rate for adults with a congenital absence of an upper limb could be as high as 64%.

Introduction

A number of studies have been undertaken relating to congenital limb reduction defects (Stoll *et al.*, 1996; Brown *et al.*, 1996; Froster and Baird, 1992; Rogala *et al.*, 1974). These studies have commented on the frequency of all congenital defects of the upper limbs. Comparison between studies is difficult as each study subdivides cases into subgroups differently. None of these studies focused specifically on cases that would benefit from prosthetic care although Froster and Baird (1992) did find a significant preponderance of left-sided defects within the subgroup of transverse defects at the radius/ulna level.

All correspondence to be addressed to Carole Fraser, Occupational Therapy Services, Box 174, Addenbrooke's NHS Trust, Hills Road, Cambridge CB2 2QQ, UK. Tel: (+44) 1223 216586. E-mail: cf13@cam.ac.uk.

McDonnell *et al.* (1988) provided estimates for the incidence of congenital upper limb deficiencies by comparing data from a number of sources. This paper focused on the number of cases that would benefit from prosthetic care but did not comment on gender. Kyberd *et al.* (1997) focused on the upper limb amputee population attending Oxford DSC. This study included both acquired and congenital upper limb absences. Although laterality and gender differences were observed amongst the clinic population with congenital upper limb absences the study did not specifically focus on these. A recent report by the Amputee Medical Rehabilitation Society (1997) provides estimates for the number of new cases per annum of upper limb congenital amputees needing prosthetic care in England. These figures are extrapolated from data from the Manchester DSC but do not give details of laterality or gender.

Cambridge DSC survey

A profile of upper limb amputees registered at Cambridge DSC was undertaken as part of a research project designed to evaluate how cosmetic and functional prostheses are used (Fraser, 1998). Analyses of the data related to congenital upper limb absences revealed significant differences between the numbers of left and right congenital absences as well as gender differences. As a result it was decided to pursue these findings further by engaging in additional enquiries.

From the Cambridge DSC records a total of 70 upper limb congenital amputees who had unilateral upper limb absences at trans-humeral or trans-radial level were identified. Some 41 were aged between 16 and 80 years in May 1996 and 29 were aged 15 years and below (Table 1).

Table 1. Congenital amputees registered with the Cambridge DSC by age, gender and side of absence.

Gender	Male		Female		Total
	Left	Right	Left	Right	
Side of absence					
Congenital 16 years and above	19	4	10	8	41
Congenital 15 years and below	11	10	15	3	29

There was a significant difference among the number of adults with a left limb absence and those with a right limb absence. Some 29 (71%) had a left-sided absence and 12 (29%) had a right-sided absence ($X^2 = 7$, $df = 1$, $p < .01$). Curiously this difference did not appear to exist in the younger age group. Twenty-nine congenital amputees aged 15 years and below were registered; 13 (45%) had a right-sided absence and 16 (55%) had a left-sided absence. Although there was a slight tendency for more left limb absences than right the difference was not significant.

It was possible that the laterality and gender differences observed in the Cambridge DSC population could have been an idiosyncratic feature of that population therefore it was decided to undertake a national survey of DSCs.

National DSC survey

The managers of 42 DSCs in the Directory Centres for the UK were contacted and a request made for details of the number of patients with a unilateral upper limb congenital absence who were registered at their centres, differentiated by side of absence, gender and age group (15 years and below and 16 years and above). Out of the total of 42 centres 5 did not have upper limb amputees registered with them, 4 centres did not reply and 8 centres were unable to supply data in the format requested. Thus useable data were available from 25 of the possible 37 DSCs which provide a service for upper limbs. There appeared no reason to think that the 12 centres not contributing data were systematically different from the group who did provide usable

data. From inspection they were geographically spread and from a knowledge of the centres they were equally represented in size.

Results of national survey

As can be seen in Table 2, of the 2075 patients with a congenital upper limb absence registered with the 25 participating DSCs, 363 (41%) aged 15 and below had a right limb absence and 515 (59%) had a left absence. In the 16 and above age group 467 (39%) had a right absence and 730 (61%) had a left absence. There were significantly more congenital amputees registered with the DSCs with a left absence than a right absence and this significant difference is observable in amputees aged 15 years and below ($X^2 = 25.6$, $df = 1$, $p < .000$) and those aged 16 years and above ($X^2 = 58$, $df = 1$, $p < .000$). There was a slight tendency towards a higher percentage of left congenital absences amongst the 16+ group when compared to the 15- group, but this difference was not significant ($X^2 = 3.07$, $df = 1$, $p > .05$).

If the data from the 25 DSCs are categorised by gender, there were significantly more males (54%) than females (46%) overall ($X^2 = 19.6$, $df = 1$, $p < .001$) (Table 3). If men and women were equally likely to have congenital absences of an upper limb and given that in 1995 the ratio of males to females in the UK was 1:1.04 (Population Trends 88, 1997) then it would be expected that in this sample there would be 1017 males and 1058 females. In fact there were 1118 males and 957 females. The finding of more males than female absences holds for children with right absences, children with left absences

Table 2. Congenital upper limb absences registered with 25 participating DSCs, by age group and side of absence.

Age	Left	Right	Total
16 years and above	730 (61%)	467 (39%)	1197 (100%)
15 years and below	515 (59%)	363 (41%)	878 (100%)
Total	1245 (60%)	830 (40%)	2075 (100%)

Table 3. Data from 25 DSCs divided into age groups, gender and side of absence.

	Left		Right		Total
	Male	Female	Male	Female	
16 years and above	355 (29%)	375 (31%)	276 (23%)	191 (15%)	1197 (100%)
15 years and below	284 (32%)	231 (26%)	203 (23%)	160 (18%)	878 (100%)
Total	639 (31%)	606 (29%)	479 (23%)	351 (17%)	2075 (100%)

and adults with right absences. In the case of adults with a left absence a significantly higher percentage of females is observed than would have been expected ($X^2 = 12.77$, $df = 1$, $p < .001$).

Age differences and estimates of non-registration

It might be assumed that all children (aged 15 and below) with a congenital absence of a limb who are registered with a DSC would continue to use the service at 16 years and beyond. However this would appear not to be the case. For the most recent year 1995, for which data are available, the ratio of 15- to 16+ for the UK population was 1:3.84 (Population Trends 88, 1997). There has been no significant increase in congenital upper limb absence in the last 16 years (Chappell, 1992) and if the life expectancy of people with congenital upper limb absences is similar to the general population it could be assumed that there should be 3.84 times as many adults (16 years and above) with a congenital upper limb absence as children with such a limb absence. From the 878 children registered it could be expected that 3,372 adults should be registered with the 25 DSCs. As only 1,197 adults were recorded as being registered this suggests that 64.5% of people with upper limb congenital absences who are aged 16 years and above are not registered with a prosthetic clinic and probably do not wear prostheses.

Although there is currently no central database for recording the number of people with upper limb congenital defects, attempts have been made to determine what the overall figure might be. McDonnell *et al.* (1988) reviewed various data sources for the UK and North America and suggested a figure of approximately 1:9400 for whom prosthetic provision could be considered. Kyberd *et al.* (1997) suggested that a figure of around 1:13,500 would best correspond to the upper limb congenital absence rate per year for the Oxford population who would benefit from prosthetic provision. From the report by the Amputee Medical Rehabilitation Society (AMRS) (1997) it is possible to derive a figure of 1:4,670 cases who would need prosthetic provision. A very rough estimate of the expected number of people with congenital limb absences in the UK, using the total population figure for the UK for 1995 of 58,606,000 and McDonnell's estimates would be around 6,235. If the Oxford figure was used then the number would be 4,341. If the AMRS data are used the number would be 12,307. Assuming that the 25 centres providing usable data were representative of the 37 centres in the UK who have people with congenital upper limb absences registered at their centres it could be estimated that approximately 3,071 people with congenital upper limb absences would be registered. Using the figure and comparing it with the figures gained from McDonnell, Kyberd and AMRS estimates it

Table 4. Alternative estimates of upper limb congenital absences extrapolated from different sources of observed data.

Age	Registered nos. for 25 UK DSCs	Est. registered nos. for 37 UK DSCs	Est. population from McDonnell <i>et al.</i> (1998)	Est. population from Kyberd <i>et al.</i> (1997)	Est population from AMRS (1992)
15-	878	1299	1288	897	2543
16+	1197	1772	4947	3444	9767
Total	2075	3071	6235	4341	12307

could be assumed that between 1,270 and 9,236 people with congenital upper limb absences are not registered with a prosthetic clinic (Table 4).

Again using these very rough estimates it could be assumed that slightly more than one fifth of the number of people with congenital upper limb absences derived from the McDonnell, Kyberd and AMRS figures would be aged 15 and below i.e. 1,288, 897 and 2,543 respectively. The estimated number of children 15 and below registered with the 37 UK DSCs is 1,299 which is very similar to the estimate derived from McDonnell. This figure would lead to the conclusions that, if the McDonnell figures were used then all children in the UK for whom prosthetic treatment should be considered appear to be registered with a DSC and that the discrepancy between the number of congenital upper limb absences actually registered with DSCs in the UK and the number that could be expected to be registered results entirely from the adult group.

If the Kyberd figures were to be used then it would have to be concluded that more children are registered in UK DSCs than were born with a congenital absence. The Kyberd figures were calculated for the Oxford population and may not reliably apply to the UK population as a whole. Nevertheless, also from the Kyberd figures, there would appear to be evidence for a lower number of adults registered with the Oxford DSC than might have been expected.

Using the numbers derived from both the Kyberd and McDonnell figures for the 16+ population there appears to be a non-registration of between 49% and 64% of adults with a congenital absence.

The AMRS figures appear to bear no relation to the other estimates. These figures are extrapolated from one high profile DSC which has a strong reputation for prosthetic care of children. In fact there is evidence from the data collected from the 25 DSCs to suggest that the centre has attracted twice the number of children with congenital upper limb absences who would have been expected from the catchment area. Interestingly if the AMRS estimate for children is halved it is comparable with the McDonnell figures and the estimated figure for the UK DSCs.

Discussion

A bias towards more left-sided upper limb congenital absences has been commented on in a

number of papers (Kyberd *et al.*, 1997; Froster and Baird, 1992; McDonnell *et al.*, 1988; Scotland and Galway, 1983). There appear to be no definitive explanations for this. Corballis and Morgan (1978) suggest that the developing embryo is under the influence of a left-right maturational gradient which seems to favour earlier or more rapid development on the left than on the right. Brown *et al.* (1989) raises the possibility that the asymmetric development of the cardio-vascular system leads to subtle differences in vessels serving the left and right limbs and that the most likely explanation for the mechanism of induction of unilateral limb defects may lie in the vascular supply to the limbs. These factors could be important if embryos are more likely to be exposed to negative influences earlier rather than at a slightly later stage in their development.

There is some evidence to suggest that people with a left-sided absence are more likely to wear prostheses. A slight but non-significant tendency towards a higher percentage of left absences in the 16+ age group as compared to the 15- group was observed in this study (Table 3). Dlugosz *et al.* (1988) found evidence of an apparent association between congenital limb reduction defects of the upper limb and learning difficulties. Children with learning difficulties may have problems in learning to use prostheses and this may lead to a higher rejection rate in this group. It would be interesting to explore this finding further. There was also a significantly higher number of 16+ females registered than would have been expected from the sample as a whole. Kyberd *et al.* (1997) also reports a significantly larger number of females with a left-sided congenital absence registered with the Oxford DSC. These findings might suggest that people and particularly females with a left absence are more likely to continue to use prosthetic services in their adulthood and therefore be good wearers of prostheses. Interestingly in the Cambridge study undertaken to evaluate how cosmetic and functional prostheses are used; amongst the possible participants, who included both congenital and acquired amputees, 57 (47%) had a left-sided absence and 64 (53%) had a right absence but amongst the actual participants in the study 36 (55%) had a left absence and 30 (45%) had a right absence. This might also support the idea that people with a left-sided upper limb absence

are more likely to wear prostheses than those with a right absence. McDonnell *et al.* (1988) suggests that a prosthesis would be more successful when fitted on the side that would have been dominant and as 85 per cent of the population is thought to be right hand dominant it could be expected that more people with a right-sided absence would become wearers of prostheses. From the findings reported in this paper this would not appear to be so.

Significantly more males than females were found to be registered with DSCs. This finding is supported by other studies (Jones and Lipson, 1991; Gregory-Dean, 1991; McDonnell *et al.*, 1988) but not commented on. Other studies have included amputees with an acquired absence. As higher numbers of males and a higher number of right-sided absences are seen amongst amputees with an acquired absence it could be that the laterality and gender differences amongst the congenital group have not been fully appreciated. This apparent discrepancy between the number of children and adults registered with DSCs could reflect an improvement over the last 20 years in the referral to prosthetic services of children with upper limb absences. Many adults over the age of 40 may never have been referred to a limb clinic. Before 1970 children were often only referred to a clinic when they went to school. Scotland and Galway (1983) demonstrated a dramatic increase in the acceptance rate in prosthetic wear in children fitted before two years of age. If this is the case and if as this study suggests most children with an upper limb absence are being registered with limb clinics it could be expected that as these children reach adulthood a higher number will continue to use their prostheses. There could then be a marked overall increase in the number of people using upper limb prosthetic services.

The non-registration of adults could also reflect the influence of parents who put pressure on their children to wear a prosthesis. Once in their teens they may reject their prostheses. Scotland and Galway (1983) and Brooks and Shaperman (1966) report an increase in the rejection rate amongst teenagers. Another reason for rejecting a prosthesis could be that people with a limb absence choose careers and lifestyles that minimise the need for 'two hands'. In the Cambridge study evaluating the use of cosmetic and functional prostheses, 54% of the participants reported that the choice of their

career had been influenced by their limb absence.

The wide discrepancies in the different estimates of people with congenital upper limb absences reflect the dearth of reliable sources for data collection. Drawing conclusions about the population of congenital upper limb absences by extrapolating data from one limb deficiency centre is likely to prove unreliable. It is unlikely that a reliable estimate for the number of people with upper limb deficiencies will be obtained until accurate records are available from all upper limb deficiency centres and the progress of both people who choose to use prosthetic services and those who choose not to are monitored.

Conclusions

This study has shown the need for detailed centralised records for upper limb absences in order to gain accurate and reliable information about this group. The number registered in any single clinic is small and it is only when larger numbers are used that certain patterns become apparent and significant. It will be important to monitor the numbers registered in DSCs over the next few years as children who should have benefited from early fitting of prostheses and improved follow-up become adults. Multicentre studies would provide a larger population of people with upper limb absences for study and more reliable information could emerge which would benefit both those who use prosthetic services and those who provide and plan the services

Acknowledgements

This research was supported by a grant from the Anglia and Oxford Regional Health Authority.

The author would like to thank the Cambridge DSC for their help and all the other DSCs who co-operated by responding to the questionnaire.

I would also like to thank Prof. Alan Wing and Dr Stephen Kirker for their help and comments in the preparation of this paper.

REFERENCES

- AMPUTEE MEDICAL REHABILITATION SOCIETY (1997). Congenital limb deficiency. Recommended standards of care. - London: Amputee Medical Rehabilitation Society (11 St. Andrew's Place, Regents Park, London).

- BROOKS MB, SHAPERMAN J (1966). Infant prosthetic fitting. *ICIB* 5(11), 16-26.
- BROWN N, LUMLEY J, TICKLE C, KEENE J (1996). Congenital limb reduction defects. - London: HMSO.
- BROWN NA, HOYLE CI, MCCARTHY A, WOLPERT L (1989). The development of asymmetry: the sidedness of drug-induced limb abnormalities is reversed in situs inversus mice. *Development* 107, 637-642.
- CHIAPPELL PH (1992). Arm amputation statistics for England 1958-88: and exploratory statistical analysis. *Int J Rehabil Res* 15, 57-62.
- CORBALLIS MC, MORGAN MJ (1978). On the biological basis of human laterality: 1. evidence for a maturational left-right gradient. *Behav Brain Sci* 2, 261-269.
- DLUGOSZ LJ, BYERS T, MSALL ME, MARSHALL J, LESSWING A, COOKE RE (1988). Relationships between laterality of congenital upper limb reduction defects and school performance. *Clin Pediatr* 27, 319-324.
- FRASER C (1998). An evaluation of the use made of cosmetic and functional prostheses by unilateral upper limb amputees. *Prosthet Orthot Int* 22, 216-223.
- Froster U, Baird P (1992). Upper limb deficiencies and associated malformations: a population-based study. *Am J Med Genet* 44, 767-781.
- GREGORY-DEAN A (1991). Amputations: statistics and trends. *Ann R Coll Surg Engl* 73, 137-142.
- Jones LE, Lipson A (1991). The care of the limb deficient child in Australia. *Prosthet Orthot Int* 15, 140-142.
- KYBERD PJ, BEARD DJ, Morrison JD (1997). The population of users of upper limb prostheses attending the Oxford limb fitting service. *Prosthet Orthot Int* 21, 85-91.
- MCDONNELL PM, SCOTT RN, MCKAY LA (1988). Incidence of congenital upper limb deficiencies. *J Assoc Child Prosthet Orthot Clin* 23(1), 8-14.
- POPULATION TRENDS 88 (1997). - London: Office of Population Census and Surveys. Office for National Statistics.
- ROGOLA E, WYNNE-DAVIS R, LITTLEJOHN A, GORMLEY J (1974). Congenital limb anomalies: frequency and aetiological factors. *J Med Genet* 11, 221-233.
- SCOTLAND TR, GALWAY HR (1983). A long-term review of children with congenital and acquired upper limb deficiency. *J Bone Joint Surg* 65B, 346-349.
- STOLL C, CALZOLARI E, CORNEL M, GARCIA-MINAURS S, GARNE E, NEVIN N (1996). A study on limb reduction defects in six European regions. *Ann Genet* 39(2), 99-104.