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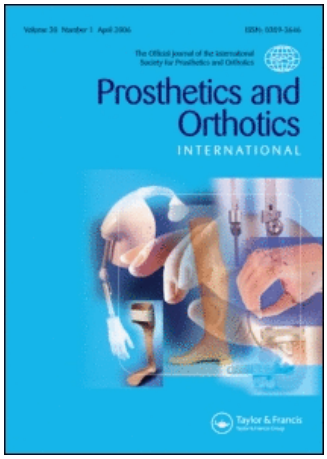

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Lower limb deficient children in the Netherlands: Epidemiological aspects

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Lower limb deficient children in the Netherlands: epidemiological aspects

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Abstract

Information on the characteristics of children with limb deficiencies and amputations in the Netherlands is largely lacking. The present study aimed to collect data about the prevalence of congenital deficiencies, the ratio of congenital to acquired limb deficiencies, types of lower leg deficiency or amputation and male/female ratios.

Data were obtained from a regional birth defects registry for the northern part of the Netherlands (EUROCAT-NNL) and from a national survey. Inclusion criteria for the selection of the EUROCAT data were: children/foetuses with lower leg deficiencies born in 1981-1986. Inclusion criteria for the survey data were: children aged 1-18 years with congenital deficiencies or acquired amputations of the leg, excluding toe deficiencies/amputations.

Both the regional birth defects registry and the national survey only yielded small numbers of children, which limits the validity of the authors' findings. The Eurocat data show a prevalence of lower leg deficiencies at birth Of 2.07/10,000. Fifty-five (55) children/foetuses were included in the present study. The male/female ratio was 1:1. Of the live-born children, 30% also had defects of the upper limbs, while 38% had bilateral lower limb deficiencies. The national survey included 89 children, of whom 73% had congenital deficiencies, while the others had undergone amputations: of which 37% were due to malignancies, 29% to traumata, 13% to infections and 21% to other pathology. The male/female ratio was 7:3 for the children with congenital deficiencies. versus 6:4 for the children with acquired amputations. In the group of congenital deficiencies, fibula deficiency was most frequently seen (36%), while in the group with acquired amputations trans-femoral amputation, knee disarticulation and trans-tibial amputation were seen with equal frequency (21%). In 40% of the children with congenital deficiency and in 8% of the children with acquired amputations the arm was also affected. Both legs were affected in 37% of the children with congenital deficiencies and in 8% of the children with acquired amputations.

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Introduction

Paediatric patients with limb deficiencies have either congenital deficiencies or acquired amputations, A congenital limb deficiency means the partial or total absence of one or more skeletal elements of the limbs at birth. The majority of such cases are due to failure of formation of parts (Day, 1991). In most patients the aetiology of congenital limb deficiency is unknown (Svoboda, 1992; Hubbard *et al.*, 1997).

In general, approximately 60% of the amputations in childhood are congenital

(Cummings and Kapp, 1992; Tooms, 1992). In a census of 4105 children in the USA, Krebs and Fishman (1984) reported 67% congenital deficiencies and 33% acquired paediatric amputees. Among congenital anomalies, the International Clearinghouse (1996) reported upper limb deficiencies in 68%, lower limb deficiencies in 23% and a combination of the two in 9%, while Jain (1994) reported 51%, 39% and 10% respectively.

Of the acquired amputations approximately 70% are due to trauma and 10-15% to malignancy, while the remaining 15% are caused by infection, vascular disease, etc. (Aitken and Frantz, 1953; Cummings and Kapp, 1992; Tooms, 1992). There are no precise figures on the numbers of children with congenital or acquired lower limb deficiencies in the Netherlands. The present study aimed to collect data about epidemiological aspects of lower limb deficient children, *viz.* the prevalence of congenital deficiencies, the ratio of congenital to acquired limb deficiency, type of lower leg deficiency or amputation, as well as sex and some other patient characteristics.

The survey was approved by the Medical Ethical Committee of the University Hospital of Groningen and of the "De Hoogstraat" Rehabilitation Centre in Utrecht.

Materials and methods

Subjects

Two approaches were used to collect data: a regional birth defects registry and a national survey. First, the prevalence of congenital lower limb reduction defects was studied by analysing the data of the European Registry of Congenital Anomalies (EUROCAT) for children born between 1981 and 1996. EUROCAT is a network of registries. The authors used data from the Northern Netherlands (NNL) EUROCAT centre. The registry includes congenital anomalies in live-born and stillborn children. Infants can be entered into the registry by various health professionals, including midwives, obstetricians, general practitioners, paediatricians and rehabilitation physicians, provided informed consent from the parents is obtained. The registration methods have been described elsewhere (Cornel *et al.*, 1992; Lechat and Dolk, 1993).

Secondly, a survey was done in all parts of the Netherlands about limb deficient children. In the Netherlands, the care for lower limb deficient children is mostly provided by rehabilitation physicians and their teams. All rehabilitation physicians in the Netherlands were asked to invite children and their parents to take part in the survey. Children aged 1-18 years with a congenital reduction defect or acquired amputation of the lower limb were included. The epidemiological study was part of a survey about the functional capacity of lower limb deficient children. Toe amputations or toe deficiencies were excluded, because these can be expected to have relatively little influence on the functional capacity of the child. It was assumed to be too much of a burden for the parents and their children to ask them about the deficiency just after the birth or amputation. Therefore the family was approached at least 1 year after the event.

After they and/or their parents had given informed consent, all children were visited at home. Data were collected by means of structured interviews. The lower extremities were examined if the child allowed this to happen. If necessary, further information was obtained from the referring physician.

Deficiency and amputation classification

Classification of the lower limb deficiencies was based on the International Standards Organisation (ISO)/International Society for Prosthetics and Orthotics (ISPO) classification system for congenital limb deficiencies (Day, 1991). In this system, the congenital limb deficiencies fall into two groups: transverse and longitudinal. The former resemble a stump after amputation, in that the limb has developed normally to a particular level, beyond which no skeletal elements are present. All other cases are classified as longitudinal, in which there is reduction or absence of an element or elements within the long axis of the limb (Day, 1991). The classification used in the present survey is shown in Table 2. In the present study, a deficiency was classified as total if only a small residual part was present (Källén *et al.*, 1984). The acquired amputations were included in the transverse category. In addition, rotationplasty was distinguished as a separate category.

Data analysis

All data were analysed using descriptive statistics in the SPSS statistical analysis

programme. A test for goodness of fit was used to analyse whether the observed frequency was significantly different from the frequency expected. The level of significance was set at $p<0.05$.

Results

The regional birth defects registry (EUROCAT-NNL)

Between 1981 and 1996, 55 children/foetuses with congenital deficiencies of the leg were registered, including 47 live-born infants. Over these years, 227, 259 children were born alive in this EUROCAT region, yielding a prevalence of 2.07/10,000 live births. Of these 47 children, 8 died before their first birthday. Of the 47 live-born, 19 only had one or more deficient toes, 3 of whom died before their first birthday.

After exclusion of children with toe defects only those who did not survive for more than one year, 23 children with congenital limb reduction defects remained. None of these had chromosomal defects. Two (2) of these 23 however, were registered with FFU complex (Lenz, *et al.*, 1993), 1 with possible FFU complex, 1 with amniotic bands syndrome and 1 with oromandibular/limb hypogenesis.

Of the total of 55 children/foetuses, 18 (33%) had deficiencies of one or both hands or arms. Of the 47 live-born children, 14 (30%) had defects of hands or arms, while 18 (38%) had bilateral lower limb reduction defects. The group of 23 children who were still alive after their first birthday and who did not suffer solely from deficiencies of the toes, included 4 (17%) children who also had arm deficiencies and 6 (26%) with both legs affected. Of the 55 children/foetuses with a lower limb deficiency, 27 (49%) were male, while 12 (52%) of the 23 children who were still alive after their first birthday and who did not suffer solely from deficiencies of the toes, were male.

The national survey

The rehabilitation physicians referred 90 children to the survey. One (1) child could not be contacted, so 89 children were included in the survey. It is not known how many children or parents refused their participation or how many physicians did not check their parent files.

All limb deficient children (n=89):

The mean age at the time of the study was 8 years and 11 months (SD 5.2; range 1.5-18.5).

Table 1. Aetiology of lower limb deficiency in 89 children.

Aetiology	*n*	%
Congenital	65	73.3
Trauma	7	7.8
Malignancy	9	10.0
Infection	3	3.3
"Other"	5	5.6

Congenital deficiencies were seen in 65 children (73%). The other 24 children had acquired amputations. Table 1 presents an overview of the aetiology. The category of "other" mentioned in this table includes pseudo-arthrosis, congenital vascular deformity, mortified lower limb femoral thrombosis, congenital lower limb deformity without deficiency.

Congenital lower limb deficiency:

In this sub-group (n=65) 45 children were male (69%) and 20 female (31%). The mean age was 7 years and 10 months (SD 5.2; range 1.5-18.1). In 24 children both legs were affected (36%). The level of the limb deficiencies in this group is shown in Table 2. Fibula deficiency is most frequent, with 32 of the 89 affected legs (36%). PFFD was seen in 21 cases. The category "other" in Table 2 includes a lower leg with one unidentified osseal structure and deformed foot, partial fibula deficiency with short toes on both sides, partial tibia deficiency with no ankle and a foot with 5 rays, fibula deficiency with partial tibia deficiency (no foot) and one unknown deformity which had already been converted into a trans-tibial amputation. In addition, 26 children (40%) also had upper limb deficiencies. In the children with unilateral deficiencies the right lower limb (n=51, 57%) was more often affected than the left (n=38, 43%), though the difference failed to reach significance ($P>0.05$).

Acquired lower limb amputation (n=24):

There were 15 boys and 9 girls (63% versus 37%) in this group. The mean age at the time of the study was 11 years and 10 months (SD 4.2; range 3-18). Two children had both legs affected; one as a result of extended trauma and the other caused by meningococcal sepsis. Two (2) children had an upper limb amputated as a complication of meningococcal sepsis. The

Table 2. Level of congenital lower limb deficiency in 65 children.

Congenital lower limb deficiency	Right	Left
Hemipelvectomy	0	0
Hip disarticulation	4	1
Trans-femoral amputation	0	0
Knee disarticulation	1	0
Trans-tibial amputation	4	4
Ankle disarticulation	3	2
Tarsal amputation	0	1
Pelvic deficiency	0	0
Proximal focal femoral deficiency (PFFD)	8	6
Proximal focal femoral deficiency (PFFD), with fibula deficiency	5	2
Tibia deficiency	3	2
Fibula deficiency, without bowing tibia	8	8
Fibula deficiency, with bowing tibia	11	5
Foot deficiency in 1 ray or more	2	3
"Other"	2	4
Total	51	38

mean age at which the amputation took place was 6 years and 6 months (SD 3.8; range 0-14 years).

Amputation levels are listed in Table 3. Twenty-six (26) lower limbs were affected, evenly divided over the left and right sides.

Discussion

The aim of the present study was to investigate epidemiological aspects of children with either congenital lower limb deficiencies or acquired leg amputations.

A regional birth defects registry and a national survey were used, but both yielded only small numbers of children who could be included. This obviously limits the validity of the results.

Based on the birth defects registry data, it was found that there was a prevalence of congenital lower limb deficiencies of 2.07/10,000 live births when toe deficiencies were included and 1.01/10,000 live births when toe deficiencies were excluded. This prevalence was found for the northern part of the Netherlands, but there are no reasons to expect a different prevalence in the other parts of the Netherlands.

Table 3. Level of acquired amputations in 26 lower limbs of 24 children.

Amputation level	Right	Left
Hemipelvectomy	0	2
Hip disarticulation	2	0
Trans-femoral amputation	4	1
Knee disarticulation	1	4
Trans-tibial amputation	2	3
Ankle disarticulation	1	2
Rotationplasty	3	1
Total	13	13

The region included in the EUROCAT-NNL covers approximately 10% of the Netherlands, hence it might be expected that about 230 children would be included in the survey if it had been possible to include all children with lower limb deficiencies. The Dutch National Medical Registration (SIG, 91-95) provides data about surgical amputations. The average incidence in children younger than 15 years is 12 cases annually (excluding children suffering only defects of toes). If all these cases have been included in this study, the population of children with acquired amputations should number approximately 200 cases. It is not known how many of these children have died within one year after the amputation or whether children with congenital deficiencies followed by conversion procedures are included. However, it may be concluded that from both the EUROCAT data and the SIG data that the population in the national survey does not include all of the children with lower limb deficiencies or amputations living in the Netherlands.

A child with either a congenital deficiency or an amputation is only likely to be referred to a

physician if it needs a prosthesis or if it encounters difficulties in activities of daily life. Some of the children who could have been included will not be referred to a physician. Moreover, it is likely that some of the children were not identified from the patient files of the physicians, because not all patient files have been computerised.

The mean age of the children with congenital deficiencies was lower than would be expected if all children with leg deficiencies had been included. It is likely that more older children were missed than younger children. From the sex ratio found in the EUROCAT data, it may be concluded that more girls have been missed than boys.

The study did, however, include the largest group of lower limb deficient children ever studied in the Netherlands.

None of the studies found in the literature about the prevalence of limb deficiencies analysed the data on lower limb deficiencies separately, although the tables presented in a few papers allowed prevalence rates for congenital lower limb deficiencies to be calculated or estimated (Froster-Iskenius and Baird, 1989; Kakurai and Kida, 1991).

The prevalence of 2.07/10,000 found in the present study from the EUROCAT data is comparable with that found in Canada, 2.1/10,000 (Froster-Iskenius and Baird, 1989) but lower than that found in Japan, approximately 2.7/10,000 (Kakurai and Kida, 1991). The sex ratio (male:female) among children with congenital deficiencies in the EUROCAT data is about 1:1. Froster and Baird (1993) found a ratio of 11:8 for live-born children with congenital deficiencies, but this was not significantly different from the male:female ratio of the general population at birth for the same period. In the Dutch national survey a male:female ratio was found for the congenital group of 9:4, while the ratio among children with acquired amputations was 5:3. Krebs and Fishman (1984) also found boys to be over-represented in their survey, with a ratio of 9:7 in the congenital group and one of 7:4 for the group with acquired amputations

Bilateral lower leg deficiencies in children older than 1 year were found in 27% of all cases in the EUROCAT data. The survey data showed bilateral lower leg deficiencies in 36% of the children with congenital deficiencies, and in 8% of the children with acquired amputations, i.e., 29% of the total group. This is higher than the percentages found by Krebs and Fishman (1984). In their study of children with congenital deficiencies and acquired amputations they found bilateral deficiencies or amputations in 13% of cases. Krebs and Fishman (1984) obtained the population for their study through prosthetic clinics in the USA. As a result, they may have missed children with proximal bilateral amputations who are more likely to be wheelchair-bound. In a group of Canadian children, Froster and Baird (1993) found that 17% had congenital bilateral lower limb deficiencies, which is again lower than the percentages in this study. The differences may have been caused by the fact that the EUROCAT data regard toe deficiencies of the contralateral side as bilateral deficiencies, which may not have been the case in the other studies. Jain (1994) reported 28% bilateral lower leg deficiencies in Indian children, which seems comparable with the Netherlands findings.

In the Dutch national survey children with congenital deficiencies represented 73% of cases, while 27% of the children had undergone amputations. Krebs and Fishman (1984) included 2,739 children with congenital deficiencies and 1,366 children with acquired amputations, i.e., 67% congenital deficiencies and 33% acquired amputations. The distribution in the present survey seems to be comparable.

The EUROCAT data showed 8 of the 47 live-born children (17%) to have died before their first birthday. It is known from the literature that children with congenital deficiencies are more likely to die in the first year than children without abnormalities (Froster and Baird, 1993; Källén *et al.*, 1984). Froster and Baird (1993) found a percentage of 4.5. However, their study excluded the children with both upper and lower limb deficiencies, as well as children with diagnoses of a syndromic nature, which means that their study is only partly comparable with the authors' study. Including all children with both upper and lower limb deficiencies in their study resulted in a percentage of 13% neonatal deaths (Froster-Iskenius and Baird, 1989).

The EUROCAT data record arm deficiencies in 18 (33%) of the children born alive with lower leg deficiencies (including defects of the toes only), while 18% of the children still alive after one year (excluding defects of the toes only) had

arm deficiencies. The corresponding figure in the Dutch national survey was 40%. Jain (1994) found a percentage of 10% and Krebs and Fishman (1984) one of 12%. This means that children with arm as well as leg deficiencies are over-represented in the present survey. Perhaps these children are more likely to be referred to a rehabilitation physician, because the treatment of children with both leg and arm deficiencies is more complex than that of children with leg deficiencies only.

The most common type of deficiency in the survey was that of the fibula (Table 2). This is an agreement with the findings of Källén *et al.*, (1984). Conventry and Johnson (1952) also mentioned the fibula as one of the bones most frequently absent or deficient at birth, but they referred to a publication from 1925. Stewart and Jain (1995) mentioned ankle disarticulations as the most frequent type of deficiency. It is not clear from their data how often such ankle disarticulations had resulted from conversion procedures. Most children with fibular deficiencies require ankle disarticulation as a conversion procedure (Kruger, 1992). In Venezuela (Fernandez-Palazzi *et al.*, 1991) the most common type of congenital lower limb deficiencies was found to be that of proximal femoral deficiencies (27%) followed by fibula deficiencies (23%).

Differences in epidemiological data found in different studies may reflect real differences between countries, but the variations are more likely to be caused by differences in assessment (i.e., the methods by which data were obtained), by the use of different definitions or by random variations. Future studies may shed more light on this problem. An uniform classification system of congenital deficiencies is one of the conditions for making studies from different countries more comparable.

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