# Congenital pseudarthrosis of the tibia

## Treatment and outcome at skeletal maturity in 10 children

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We present the clinical characteristics of 14 children with congenital pseudarthrosis of the tibia (CPT) treated in northern Finland at Oulu University Hospital during the years 1968–1996. The incidence of CPT in northern Finland was $3.5/10^5$ during years 1962–1996, which is higher than in previous reports. The estimated prevalence of pseudarthrosis was $1.8/10^5$. The first 10 children with a mean follow-up time of 19 years from the first grafting procedure and 14 years from the last grafts, all had a solid bony union and could walk without external support at the last follow-up. The best treatment, especially in unfavorable types, seems to be a radical resection of the pseudarthrosis and reconstruction with a free vascularized fibular graft.

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Congenital pseudarthrosis of the tibia (CPT) continues to be a perplexing orthopedic problem in which it is difficult to achieve and maintain bony union and good alignment after manifestation of the pseudarthrotic fracture (Masserman et al. 1974, Andersen 1978, Morrissy and Riseborough 1981, Boyd 1982).

Over 20 conventional bone grafting and fixation methods have been used, usually resulting in a more or less deformed leg or even a juvenile amputee (Sofield 1971, Boyd 1982, Murray and Lovell 1982, Umber et al. 1982, Jacobsen et al. 1983). Newer techniques such as electrical stimulation (Paterson and Simonis 1985), vascularized bone graft (Taylor et al. 1975, Chen et al. 1979, Hagan and Buncke 1982, Solonen 1982, Leung 1983, Paterson 1989, Pho et al. 1985), compression-distraction (Plawecki et al. 1990) and bone transport (Ilizarov and Gracheva 1971) seem more promising than earlier conventional graft procedures, at least in terms of achieving a primary bony union.

14 children with CPT have been treated in our hospital during the years 1968–1996. We present the clinical characteristics of these patients, calculate the incidence of CPT in northern Finland, and evaluate the long-term results of surgical treatment in the first 10 cases who have reached skeletal maturity.

## Patients and methods

### *Epidemiological study*

The study area was Oulu University Hospital's region, with a population of 733,000 (December 31, 1996) (Central Statistical Office in Finland, 1997). The basic epidemiological material consisted of patients attending the Department of Pediatric Surgery at Oulu University Hospital for treatment from 1968 onwards. Additional patients with a diagnosis of pseudarthrosis (International Catalogue of Disease (ICD) numbers 8–10) were traced from the records of the University Hospital. In addition, patients were traced by contacting pediatric surgeons and orthopedists in the region.

The incidence figures were calculated from the number of individuals born with pseudarthrosis in relation to the total number of live births in the region during 1962–1996. For prevalence figures, the number of affected individuals in the population at a particular time was related to the total population.

14 children (9 girls) with CPT were treated dur-

Table 1. Patient data

| A | B | C | D | E | F | G | H | I | J | K | L | M | N | O | P | Q | R |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 1968 | f | right | 5 y | no | II | 1 | 33 | limping | 7 | <5 | <5 | no | 2.5 | normal | solid | poor |
| 2 | 1972 | m | left | 1 y | no | III | 2 | 26 | normal | 2.5 | <5 | <5 | no | 0 | deficient | solid | good |
| 3 | 1972 | m | right | 1y 1m | yes | II | 1 | 25 | limping | 3.5 | <5 | 17 | no | 2 | deficient | solid | poor |
| 4 | 1977 | f | left | 4y 11m | no | III | 2 | 24 | normal | 1 | <5 | 14 | no | 0 | normal | solid | good |
| 5 | 1977 | f | right | 4y 4m | no | V | 3 | 22 | normal | 0 | <5 | <5 | 20 | 1.5 | deficient | solid | fair |
| 6 | 1978 | f | left | 1y7m | no | II | 4 | 19 | limping | 6 | <5 | <5 | no | 3.5 | deficient | solid | poor |
| 7 | 1978 | f | left | 7m | yes | II | 1 | 18 | normal | 1 | 22 | 30 | no | 2 | normal | solid | good |
| 8 | 1979 | m | right | 10d | no | I | 2 | 17 | normal | 0 | <5 | <5 | no | 0 | deficient | solid | good |
| 9 | 1981 | f | right | 7m | yes | III | 1 | 17 | normal | 0 | <5 | <5 | 15 | 0 | deficient | solid | good |
| 10 | 1982 | f | left | 6m | yes | II | 1 | 15 | normal | 2 | <5 | <5 | no | 0 | deficient | solid | good |
| 11 | 1992 | f | right | 7y | no | V | 1 | | | | | | | | | | |
| 12 | 1993 | m | right | 6m | yes | II | 2 | | | | | | | | | | |
| 13 | 1994 | m | left | 5m | yes | I | 2 | | | | | | | | | | |
| 14 | 1994 | f | left | 11m | no | II | 3 | | | | | | | | | | |

A Patient no.
B Year at diagnosis
C Sex
D Side of CPT
E Age at presentation
F NF1
G Boyd's classification
  I frank pseudarthrosis in the tibia at birth
  II anterior bowing and a sclerotic hourglass constriction of the tibia
  III cystic lesion in the tibia
  IV sclerotic segment with stress fracture
  V fibular dysplastic type II changes preceded tibial ones
  VI intraosseus neurofibroma
H Andersen's classification
  1 dysplastic
  2 cystic
  3 sclerotic
  4 mixed type
I Age at last follow-up, years
J Ability to walk without support
K Leg length inequality, cm
L Tibial deformity, degrees valgus
M Tibial deformity, degrees procurvatum
N Ankle valgus deformity, degrees
O Difference in leg circumference, cm
P Ipsilateral fibula
Q Tibial union
R Results at the last follow-up

ing 1968–1996. Admissions of these children were divided rather unequally between the four decades (Table 1). Clinical manifestation of the disease occurred at less than 2 years of age in all but 4 children . The patients were classified on the basis of the primary radiographs, as proposed by Boyd (1982) and Andersen (1973) (Table 1).

Neurofibromatosis type 1 (NF1) was diagnosed in 6 children (Table 1), using the criteria of the National Institutes of Health (NIH) (Stumpf et al. 1988, Gutmann et al. 1997). Two of the following criteria were needed for NF1: 6 or more café au lait macules; 2 or more neurofibromas of any type or one plexiform neurofibroma; multiple freckles in the axillary area or the inguinal regions; optic glioma; 2 or more Lisch nodules (iris hamartomas); a distinct osseous lesion, such as sphenoid dysplasia, or thinning of the bone cortex, with or without pseudarthrosis; a first-degree relative (parent, sibling, or offspring) who meets the above criteria for NF1.

### *Treatment in the 10 children who have reached skeletal maturity*

3 of the first 10 children were treated only with traditional autogenous bone transplantation methods (Table 2). The commonest technique consisted of onlay chip autografts from the iliac crest to the partially excised pseudarthrosis. In 2 cases, massive dual allografts taken from the mother's tibia were also used (Figure 1). Fixation in connection with conventional grafting was performed by intramedullary nailing, plate or screws.

A free vascularized pedicle graft with radical resection of pseudarthrosis was used in 7 children , usually after one or more unsuccessful conventional graftings (Table 2). A microvascular iliac crest graft was used in one patient treated at the Orthopaedic Hospital of the Invalid Foundation in Helsinki in 1980 (Solonen 1982). A vascularized pedicle graft from the contralateral fibula using Taylor's technique (Taylor et al. 1975) was undertaken at our hospital in 6 cases during the 1980s

**Table 2. Treatment of the first 10 children**

| Patient no. | Treatment to obtain bony union: Age at first/ last grafting | Convent. procedures, number and methods | Free vascularized bone graft | Later treatment for deformity or complications: Problem | Procedure |
|---|---|---|---|---|---|
| 1 | 5y/12y | 7, onlay chips<br>1, cortical dual graft | VIC [a] | Genu valgum | Tibial osteotomy |
| 2 | 1y 6m/8y 9m | 3, onlay chips (last w IMN) [a] | – | – | – |
| 3 | 1y 2m/5y 1m | 2, cortical dual grafts<br>3, onlay chips (last w IMN) [a] | – | Valgus ankle in the affected limb | Distal tibial osteotomy |
| 4 | 5y 1m | 1, onlay chips with IMN [a] | – | – | – |
| 5 | 4y 4m/9y 11m | 2, onlay chips to fibular pseudarthrosis | VFG [a] | Valgus ankle in the affected limb | Distal tibial osteotomy |
| 6 | 1y 8m/10y 4m | 7, onlay chips (the last to distal fusion of VFG)* | VFG [a] | Shortening of the tibia | Tibial osteotomy<br>Tibial elongation |
| 7 | 1y 2m/6y 11m | 2, onlay chips | VFG [a] | Valgus ankle in the affected limb | Distal tibial osteotomy |
| 8 | 10d/2y 11m | 2, onlay chips | VFG [a] | Valgus ankle in donor limb<br>Shortening of the tibia | Distal tibial osteotomy<br>Tibial elongation<br>Epiphyseodesis in the opposite femur |
| 9 | 4y 8m/5y | 1, onlay chips to the distal fusion of VFG | VFG | Valgus ankle on both limbs | Distal tibial osteotomy bilaterally |
| 10 | 7m/6y 6m | 1, onlay chips | VFG [a] | Shortening of the tibia | Tibial osteotomy × 2<br>Tibial elongation |

[a] last grafting procedure: IMN with intramedullary nail, VFG vascularized fibular graft, VIC vascularized iliac crest graft

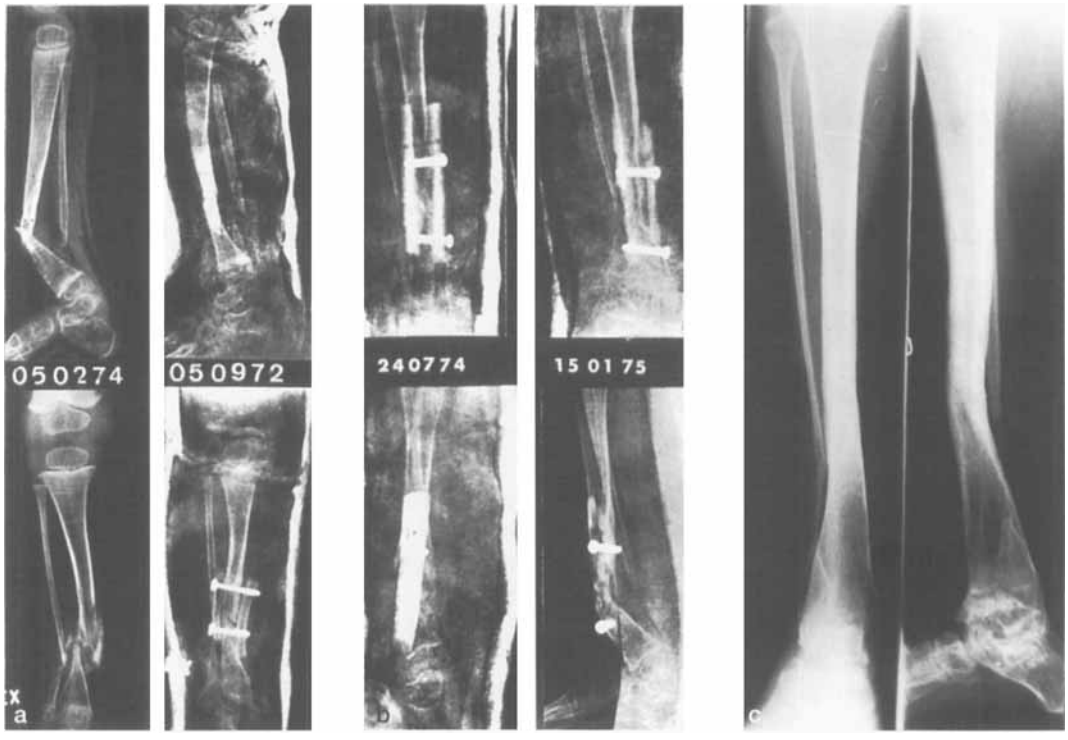


Figure 1. Patient 3, with Boyd's group II, Andersen's dysplastic type of CPT and NF1. Dual rib autografts were placed across the CPT at the age of 1 year and 2 months and osteolysis of the grafts was complete after a year (a). A massive dual homograft from the mother's tibia was implanted at the age of 3 years and after 6 months the grafts had disappeared almost entirely, because of an aggressive osteolysis (b). Solid bony union was achieved after partial resection of CPT with onlay chip grafts and stabilization with a transtarsal intramedullary nail at the age of 7 years. The final result was poor because of 3.5 cm shortening and atrophy of the limb and ankle rigidity at the age of 25 years (c).



Figure 2. Patient 8, with neonatal CPT (Boyd's group I) without NF. The leg was edematous and fractured at birth (a). Radiographs after 2 conventional onlay grafts and a rib pedicle graft with a broken intramedullary nail is shown on the left in Figure b at the age of 2 years and 10 months, immediately after resection at the age of 2 years and 11 months and transfer of a vascularized pedicle graft from the contralateral fibula in the middle, and the situation 2 years later on the right. Following tibial elongation at 6 years of age, contralateral femoral epiphyseodesis at 12 years of age, and tibial osteotomy to correct ankle valgus at the age of 15 years, the final result 14 years after the fibular transplantation at the age of 17 years was good (c).

(Table 2). The transferred fibula graft was telescoped into the tibial ends and postoperative immobilization was carried out with a plaster cast in 2 children (Figure 2) and external fixation followed by plaster cast or splint support in 4 children (Figure 3).

2 of the 6 children treated with vascular transfers from the contralateral fibula needed an additional graft procedure to achieve persistent union at the fusions between the graft and the ends of the resected tibia (Table 2). Fibular lesions were not operated on in most patients.

A total of 40 bone grafting operations were performed (Table 2). Numerous additional surgical procedures were needed. Nails, plates, screws and fixation devices were removed, large numbers of plaster casts and glass fiber bandages were prepared postoperatively or after removal of the fixation devices, and various braces were used for early ambulation and the prevention of recurrent angulation. Stress fractures in the graft occurred in 2 children after solid union of the tibia, requiring further treatment with casts, braces and bandages.

All but 2 of the 10 children underwent one or more corrective surgical procedures for tibial and/or ankle deformities after permanent bony union. 2 of the 6 patients with a vascularized pedicle graft taken from the contralateral fibula needed correction with distal tibial osteotomy for a significant valgus deformity of the ankle in the donor leg.

Evaluation of the final outcome in the first 10 children was based on discussion with the patients about their symptoms and on clinical radiographic findings enumerated in Table 1. Radiographs were taken of the leg and ankle to assess the union of tibia and fibula, angulation of the tibia and the valgus deformity of the ankle. The leg length inequality was measured clinically and radiological-


Figure 3. Patient 7, with Boyd's group II and Andersen's dysplastic type of CPT and NF1. A good bony union was achieved 2 months after radical resection of CPT and transfer of a vascularized pedicle graft from the contralateral fibula at the age of 7 years (a). The recurrent procurvatum has increased over the years until skeletal maturity, but not thereafter. At the last follow-up at the age of 18 years, it was 30° (b).

ly. The findings were scored as follows: ability to walk: normal = 2, limping = 0; inequality of leg length: < 1.5 cm = 2, 1.5–4 cm = 0, > 4 cm = –1; tibial valgus/procurvatum: < 5° = 2, 5°–20° = 1, > 20° = 0; ankle valgus deformity: no = 1, yes = 0; difference in leg circumference: 0–1 cm = 1, > 1 cm = 0; ipsilateral fibula: normal = 1, deficient = 0; tibial union: solid = 1. The total scores of the patient qualified the result at the last examination as follows: 0–4 = poor, 5–6 = fair and 7–10 = good.

## Results

### *Epidemiological study*

In Oulu University Hospital's region, a total of 399,616 children were born during 1962–1996. The incidence of CPT at birth was 14/399,616 or 1/28,544, i.e., $3.5/10^5$. By the prevalence day (31 December, 1996), 1 of the known 14 pseudarthrosis patients had moved out of the region. Based on the remaining 13 children, the prevalence of pseudarthrosis in northern Finland was 13/733,037 or $1.8/10^5$.

Neurofibromatosis was diagnosed in 6 children by clinical examination and excluded in the remaining 8 cases. 3 NF1 patients were familial and 3 sporadic cases. All NF1 patients with pseudarthrosis also had typical skin manifestations of NF1. 1 of the 6 cases with NF was placed in Boyd's favorable group III, while 4 were classified as Andersen's unfavorable dysplastic type.

### *Outcome at the last follow-up in the 10 skeletally mature patients*

The average age at the first bone grafting procedure was 2 years (10 days–5 years) and at the last graft 7 years (2–12 years) (Table 2). The average follow-up time was 19 years (11–27 years) from the first grafting procedure and 14 years (8–19 years) from the last grafts. All 10 children had reached skeletal maturity by the time of the last follow-up examination. The youngest patient was 15 years old by that time and the oldest 33 years.

A permanent solid bony union was achieved in all 10 patients. They could walk without external support and 7 patients even without limping. The

final successful grafting procedure consisted of onlay autogenous chip grafts, with an intramedullary nail in 3 patients (Figure 1) and a vascularized pedicle graft in 7, although 2 of these (patients 6 and 9) needed additional bone chips on the distal fusion of the fibular graft (Table 2) (Figures 2 and 3). At the follow-up, 1 patient (no. 2) complained about pain on the lateral side of his leg. He had a fragile fibular pseudarthrosis, for which osteosynthesis was done afterwards. The others had had only transient discomfort during motion.

Limping, shortening, recurrent or residual tibial bowing, valgus deformity of the ipsilateral or contralateral ankle, hypotrophy of the affected leg and foot were the main factors worsening the results. The outcome was good in 6 cases, fair in 1 and poor in 3 (Table 1).

The final result was good in the 3 cases of Andersen's favorable cystic form (Andersen 1973) and the patients belonging to Boyd's corresponding group III (Boyd 1982) also had good results.

## Discussion

### *Epidemiological study*

We believe that these 14 patients constitute most of the pseudarthrosis patients in northern Finland. In addition to the patient diagnosis list of University Hospital, cases were sought by inquiring among the pediatric surgeons and orthopedists in the study area about their pseudarthrosis patients. Patients diagnosed and treated only in the Orthopaedic Hospital of the Invalid Foundation in Helsinki, may have been missed in our search. In addition, mildly affected undiagnosed cases might exist in the study region. Thus the figures calculated in this study represent the minimal incidence and prevalence.

The incidence ($3.5/10^5$ live births) of CPT in northern Finland seems to differ from the estimate of 1/190,000 live births presented by Andersen (1978) and even more from the figure of 1/250,000 live births reported by Fairbank (1994). The reason is unknown. Our calculated prevalence of pseudarthrosis was $1.8/10^5$. This must be considered as only a rough estimate, due to the small number of cases.

The clinical findings of CPT in our 14 patients corresponded well to those reported previously with respect to sex, side, age at presentation, presence of NF and types of lesions (Boyd and Sage 1958, Andersen 1973, Masserman et al. 1974, Boyd 1982, Murray and Lovell 1982, Umber et al. 1982, Crawford and Bagamery 1986, Fairbank 1994).

### *Treatment*

As in previous series, it was difficult to achieve and maintain persistent union and good alignment (Sofield 1971, Morrissy and Riseborough 1981, Murray and Lovell 1982, Umber et al. 1982). Only 2 patients had pseudarthrotic fractures that could easily be treated merely by conventional grafting with intramedullary nailing. Permanent bony union of the affected tibia was usually achieved at the cost of multiple operations and hospital stays, as an average of 4 bone grafting procedures was needed with subsequent removal of screws, plates, nails or external fixation devices and applications of plaster casts, glass fiber bandages and various braces. In addition, an average of 1.6 corrective operations per patient was performed after permanent bony union of the tibia for recurrent or residual bowing of the tibia, shortening of the limb, or ankle valgus deformities.

Despite the difficult treatment with many complications, significant positive signs could be found in our long-term results. All 10 patients were walking independently at the last follow-up examination, with no protective supports, and their mobility was sufficient for a normal working life and everyday activities.

A stable fixation with plates or intramedullary nails (Charnley 1956, Sofield 1971, Andersen 1973, Murray and Lovell 1982, Umber et al. 1982, Dormans et al. 1990, Baker et al. 1992) and external devices (Hagan and Buncke 1982) have been considered the principal prerequisite for sound healing.

In 7 of the 10 cases, a vascular pedicle autograft had been used to replace the tibial defect following radical resection of the pseudarthrosis. The vascularized grafts responded well to the increased stress, with hypertrophy and tibialization in accordance with Wolff's law (Figures 2 and 3). Only 2 of these patients needed an additional graft

to the distal fusion to attain permanent union. We feel that the vascularized fibular and iliac crest grafts were of value to our patients, as they evidently helped to avoid further conventional bone graftings which would have resulted in prolonged hospitalization, more dystrophic limbs and less satisfactory long-term results.

Progressive tibial angulation after consolidation of the fibular pedicle grafts was a late complication in some patients, and it seemed to us that more prolonged external support would have been needed to avoid this pitfall. To obviate recurrent angulation during weight bearing, running and jumping, Crawford and Bagamery (1986) recommended that grafted legs should be protected with braces, even up to the time of skeletal maturity. We agree with this statement.

Another late complication was ankle valgus deformity in both the affected and the donor legs. This shows that, in addition to tibial surgery, distally located fibular lesions must be treated. Tibiofibular fusion (Langenskiöld 1967, Umber et al. 1982) or transfixation (Paterson 1989) can be used to prevent progressive valgus deformity of the ankle. Such procedures would have been important for the donor leg in 2 of our patients, although the remaining distal part of the fibula represented more than one quarter of the whole length of the bone, which has been estimated to be sufficient for stabilization of the ankle (Taylor et al. 1975).

Prior to the introduction of newer surgical techniques, the long-term outcome achieved in the treatment of CPT was unsatisfactory and amputation was indicated in many cases since it gave a better outcome than that of a dystrophic limb with severe shortening (Andersen 1973, Morrissy and Riseborough 1981, Murray and Lovell 1982, Jacobsen et al. 1983). The severest disadvantages suffered by our 3 patients with a poor result were shortening and dystrophy of the limbs. These patients had undergone 5–9 unsuccessful conventional graft operations before the final procedure that induced permanent union. Bony union was finally achieved by conventional autografting with transtarsal intramedullary nailing in one of these cases and by means of a pedicle vascular fibular graft in 2 patients. Prolonged inactivation because of multiple operations and premature distal tibial growth arrest could probably have been avoided in these patients, if the vascularized fibular graft operations had been performed earlier. These 3 patients belonged to Boyd's unfavorable group II and Andersen's dysplastic and mixed types.

We agree with other authors who state that a radical resection of pseudarthrosis and substitution of the tibial defect with a pedicle vascularized fibular graft from the opposite limb is a valid although relatively complex method for treating difficult types of CPT (Taylor et al. 1975, Chen et al. 1979, Pho et al. 1985, Dormans et al. 1990). Vascularized grafts should obviously be used more frequently, to avoid multiple unfavorable traditional operations. However, Baker et al. (1992) have recently stressed once again the value of conventional graftings with intramedullary nail fixation (Charnley 1956, Boyd and Sage 1958, Umber et al. 1982). The late complications that have now emerged (recurrent angulation and ankle valgus deformity) could probably have been avoided by using long-term protective supports postoperatively and performing prophylactic tibiofibular fusions.

It has been difficult to find firm evidence to support the assumption that CPT patients with NF should have a poorer outcome than those without NF (Crawford and Bagamery 1986, Fairbank 1994). The main reasons are that the series of patients are too small and that the lesions are highly heterogeneous. This is also true of our series. Our findings indicated detrimental effects of neurofibromatosis on the outcome, however, and confirmed that the children belonging to the unfavorable types as defined by both Andersen and Boyd seemed to have a poorer prognosis.

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