

CONGENITAL PSEUDARTHROSIS OF THE TIBIA AND NEUROFIBROMATOSIS

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On the basis of 21 cases of congenital pseudarthrosis of the tibia, which were radiologically typed and later personally examined, it was found that all cases of the dysplastic type also showed evidence of neurofibromatosis. No evidence of neurofibromatosis was found among the cystic and the clubfoot type of pseudarthroses.

Key words: congenital; pseudarthrosis; tibia; neurofibromatosis

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Congenital pseudarthrosis of the tibia seems to be related to neurofibromatosis. In order to establish the relationship, and if possible to make further distinctions among cases of congenital pseudarthrosis of the tibia the following investigation was carried out.

MATERIAL

Congenital pseudarthrosis of the tibia is taken to mean all congenital fractures of the tibia including pseudarthrosis after a pathological fracture or after osteotomy of a lower leg with congenital anterior angulation. From cases of congenital pseudarthrosis treated in Danish Orthopaedic Departments and at Shriner's Hospital, Chicago, there were 21 in which a complete series of X-ray films from immediately after birth or prior to the occurrence of fracture were available. For all cases complete records from the time of the first treatment were available. All 21 patients have later been questioned and examined for evidence of neurofibromatosis.

A diagnosis of neurofibromatosis was considered positive when the patient showed typical *cafe au lait* spots and had a positive family history of neurofibromatosis. In cases where the family history was negative the diagnosis of

neurofibromatosis was only considered positive when the child had more than five typical *cafe au lait* spots each of more than 0.5 cm in diameter. To obtain a more concise concept of the disease the three cases of pseudarthroses which had occurred after osteotomy were considered separately.

RESULTS

Of the 21 patients, 12 had a positive diagnosis of neurofibromatosis (Table 1). In five cases there was no family history of neurofibromatosis but all patients showed typical *cafe au lait* spots and in addition one patient also had multiple tumours of the skin. In four of these cases the parents were examined without finding any evidence of neurofibromatosis. In the fifth case the patient was adopted and had no knowledge of her family.

Of the 12 patients treated in Danish Departments six had a positive diagnosis of neurofibromatosis. Of the nine American patients six had a positive diagnosis (Table 2). The distribution of the radi-

ological types of pseudarthroses among the 21 cases is seen in Table 3. The various radiological types of pseudarthroses found among the 15 cases of congenital pseudarthrosis born in Denmark between 1940 and 1965 are seen from Table 4 (Andersen 1972).

Table 1. Number of patients with congenital tibial pseudarthrosis with and without evidence of neurofibromatosis.

	No. of patients
Evidence of neurofibromatosis in patient and history of neurofibromatosis in family	7
Evidence of neurofibromatosis in patient but no history of neurofibromatosis in family	5
No evidence of neurofibromatosis in patient or in the family	9
Total	21

Table 2. Cases of congenital tibial pseudarthrosis with and without neurofibromatosis in relation to geographical place of treatment.

	No. with neurofibromatosis	No. without neurofibromatosis
Treated in Denmark	6	6
Treated in Chicago	6	3

Table 3. The incidence of neurofibromatosis among the various radiological types of congenital tibial pseudarthrosis.

Radiological type of congenital tibial pseudarthrosis	Total no.	No. with neurofibromatosis
Dysplastic	8	8
Cystic	5	0
Sclerotic	1	1
Clubfoot	4	0
Pseudarthrosis after osteotomy	3	3

Table 4. Cases of congenital tibial pseudarthrosis born in Denmark between 1940–1965 in relation to radiological type and neurofibromatosis.

Radiological type of congenital tibial pseudarthrosis	Total no.	No. with neurofibromatosis
Dysplastic	5	5
Cystic	1	0
Sclerotic	1	0
Clubfoot	3	0
Unknown Pseudarthrosis after osteotomy	2	1
	3	3

DISCUSSION

The relationship between congenital pseudarthrosis of the tibia and neurofibromatosis was first noticed by Ducroquet (Ducroquet & Cottard 1939) who found that 10 out of 11 of his patients also showed evidence of neurofibromatosis. This relationship has since been accepted by several authors although the occurrence of neurofibromatosis among cases of congenital pseudarthrosis of the tibia has shown a considerable variation. Van Nes (1966) thus concluded that among 22 patients treated by him there were no cases of neurofibromatosis. In recent articles (Duhaine et al. 1972, Hardinge 1972, Masserman et al. 1974, McBryde & Stelling 1972, Sofield 1971) the percentage has varied from 38 to 70. On the basis of the possible relationship between neurofibromatosis and congenital pseudarthrosis, Aegerter (1950) suggested a relationship between neurofibromatosis and fibrous dysplasia.

The discrepancies in the recorded instances of neurofibromatosis might in some cases be due to inadequate clinical data, but it might also be due to the fact that it is difficult, especially in young children, to establish the diagnosis (Fienman & Yakovac 1970).

In the present investigation it can be seen that in all cases of the so-called

Figure 1. Dysplastic type of congenital tibial pseudarthrosis before occurrence of fracture.

Figure 2. Sclerotic type of congenital tibial pseudarthrosis with pseudarthrosis of the fibula and imminent fracture of the tibia.



dysplastic type of congenital pseudarthrosis (Andersen 1973) (Figure 1) a positive diagnosis of neurofibromatosis could be established. Among the cystic cases there was no evidence of neurofibromatosis. This type is often looked upon as a localized form of fibrous dysplasia. As a biopsy has not been routinely performed in the present cases this diagnosis of fibrous dysplasia in the cystic cases could not be confirmed. If Aegerter's previously-mentioned theory is accepted it would seem that there might exist two types of cystic congenital pseudarthrosis. This has not been confirmed so far and the present investigation was also unable to corroborate this view.

In the only case of the so-called sclerotic type of pseudarthrosis in the present investigation (Figure 2), a positive diagnosis of neurofibromatosis was found. As other cases of this type of pseudarthrosis have been seen without any evidence of neurofibromatosis it might be possible that they are not all alike. The clubfoot type of pseudarthrosis which has a much more favourable prognosis (Andersen 1973) showed no evidence of neurofibromatosis.

The present investigation has thus confirmed that clinically there seems to exist at least three different types of congenital pseudarthrosis. The pseudarthrosis connected with neurofibromatosis is easily recognized at the primary

X-ray films because of its dysplastic appearance and its antero-lateral bowing.

Among the three cases of pseudarthrosis which arose after osteotomy all had a positive diagnosis of neurofibromatosis. The X-ray films from before the osteotomy showed typical antero-lateral bowing but no obvious dysplasia. Although it has been stressed previously it should be emphasized that children with congenital antero-lateral bowing of the lower leg and with neurofibromatosis should not have corrective osteotomies unless the indications are very well substantiated and the extremely great risk of pseudarthrosis has been seriously considered.

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