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THE INCIDENCE OF SCOLIOSIS DUE TO NEUROFIBROMATOSIS

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Cobb in 1950 stated that a high proportion of scolioses, classified as idiopathic, may be due to neurofibromatosis. This paper describes a retrospective study of a series of 3209 scoliosis cases to determine the incidence of neurofibromatosis. It was found to be just over 3 per cent. Two different clinical pictures of scoliosis were seen among the neurofibromatosis cases.

Key words: scoliosis; neurofibromatosis

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Neurofibromatosis or von Recklinghausen's disease is generally a dominant and rarely a recessive hereditary disease (Kleind 1970, Robert 1970). The exact aetiology is still unknown. The incidence of the disease is one in 2500–3000 births. The disease is characterised by neuromata, pigmented "café au lait" spots on the skin, visceral abnormalities and bone lesions. Spinal deformity is the most frequent manifestation of bone involvement (Law & Pallis 1963, Cotrel 1970). The incidence of scoliosis associated with neurofibromatosis varies from 4 to 43 per cent in 248 cases reported in the literature (Table 1) (Hagelstam 1948, James 1955, Scott 1965, Moe et al. 1972, Biot et al. 1974).

In 1950, Cobb stated that a high proportion of scolioses classified as idiopathic may be due to neurofibromatosis. The exact figure for such an incidence could not be found in the literature. The purpose of this study was to determine the incidence of neurofibromatosis in a large series of 3209 cases of scoliosis.

MATERIAL

In 1972, with the kind permission of Dr. Cotrel, the author had the opportunity to study retrospectively the details of the records and the roentgenograms and to examine a large number of the 3017 patients who had been admitted to the scoliosis service of the Institut Calot, Berckplage, France, during the previous 23 years. To this were added a further 192 scoliosis patients who had been admitted to the Shafa Rehabilitation Hospital in Tehran in the previous 4 years.

Altogether there were 98 cases of scoliosis due to neurofibromatosis in this series of 3209 scolioses. The incidence in this series, therefore, is slightly over 3 per cent (Table 2). There were 51 girls and 47 boys and thus neither sex was dominant. The age at the time of appearance of the scoliosis ranged from 6 months to 36 years. However in the majority it occurred between 11 and 15 years (Table 3). The criteria for diagnosis were family history, which was present in only 18 per cent of cases, skin lesions, neuromata, six or more "café au lait" spots or both

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Table 1. Cases of scoliosis associated with neurofibromatosis reported in the literature.

Year	Author(s)	No. of cases
1948	Haglestam collected from literature	77
1951	Kessel	2
1955	James	33
1959	Casman	3
1962	Philippart	16
1865	Scott	19
1969	Curtis et al.	8
1972	Moe et al.	55
1974	Biot et al.	35
	Total from the literature	248
	Present reported series	98
	Total no. of cases reported	346

(Veliskalis 1960) and spinal deformity, which was present in 100 per cent of cases (Table 4, Figure 1). The spinal lesions were in the form of scoliosis, kyphosis or kyphoscoliosis, but not lordosis (Table 5).

All the roentgenograms and notes of the 98 patients have been carefully examined and assessed. Twenty-three patients complained of pain, seven were suffering from dyspnoea, and the deformity of the spine with or without one or both of the other two factors were the complaints of the remaining 60 cases (Table 6). The curvature of the spine was generally a short curve with the acute angle involving less than five vertebrae in the majority of cases. Destruction and distortion of vertebrae were as a rule part of the radiographical picture (Figure 2). While scalloping of vertebrae and a widening of intervertebral foramina were rather common,

Table 2. Incidence of neurofibromatosis in the two scoliosis series.

Name of hospital	Total no. of scoliosis cases	No. of scolioses due to neuro-fibromatosis	Per cent
Institut Calot Berck-plage, France	3017	89	3
Shafa Rehabilitation Hospital, Tehran, Iran	192	9	4½
Total	3209	98	Just over 3

Table 3. Age at the onset of scoliosis in neurofibromatosis.

Age in years	No. of patients	Per cent of patients
0–5	20	20
6–10	14	14
11–15	48	50
Over 16	13	12
Unknown	6	5

biopsies from destroyed vertebrae gave a positive picture of neurofibromatosis in only seven cases. Lesions in bones other than the spine were seen in six cases (Figure 3). The curvature was in the cervical, thoracic and thoraco-lumbar region but not in the lumbar region with the majority being in the thoracic region (Table 7). The convexity in 71 cases was to the right; a hump was present in 76 cases. Details are recorded in Table 8.

It is important to note that more than 50 per cent of scolioses due to neurofibromatosis first appear at approximately the same age as the idiopathic type. But unlike the idiopathic form they involve a limit number of vertebrae, generally producing a short acute angle curve with more rotation, severe distortion and in some cases a considerably more rapid progress.

TREATMENT

All patients with curvature between 15° and 40° were treated with bracing and dynamic exercises. For those who deteriorated under this regime surgical treatment was inevitable. However, it was considered advisable to start surgical treatment without delay in patients whose curvature was over 60°. Sixty-two cases received conservative treatment, which consisted of bracing, correction with localizer Risser cast or E.D.F. Cotrel cast, together with active exercises. Of these, 32 cases proved that their curve was stable even

Table 4. Criteria for diagnosis of neurofibromatosis.

Criteria	No. of patients	Per cent of patients
Family history	18	20
"Café au lait" spots (6 or more)	94	95
Neurofibroma	35	34
Spinal deformity	98	100

Figure 1. Thoraco-lumbar scoliosis with a large "café au lait" spot.

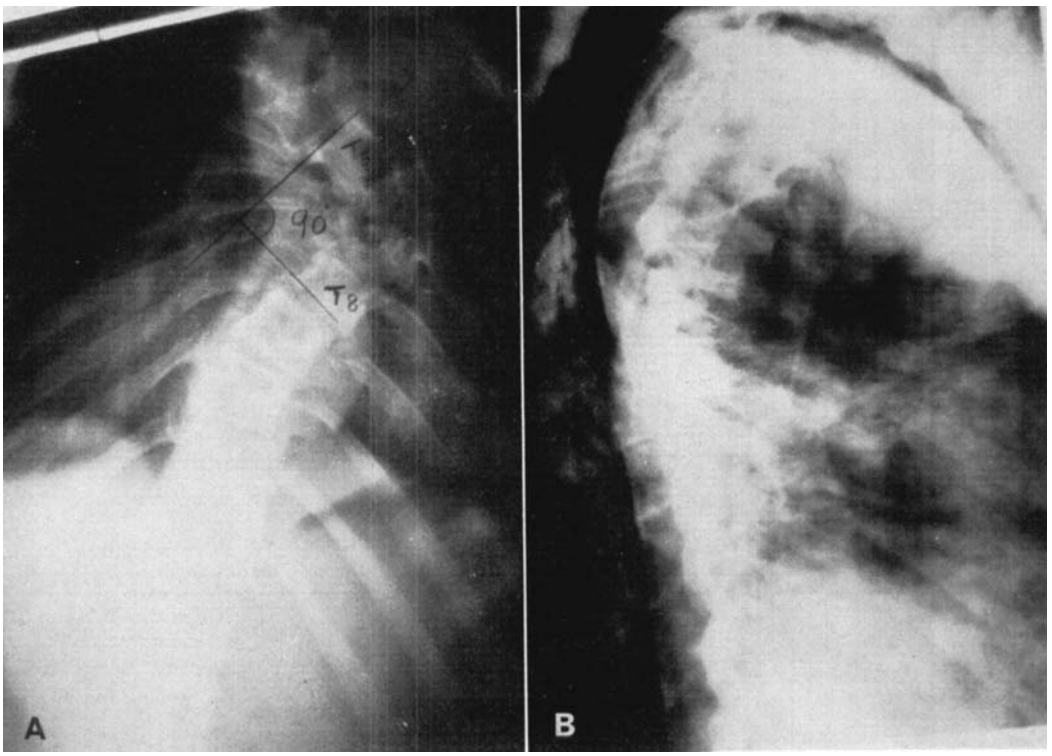

Figure 2. Typical X-rays of kyphoscoliosis due to neurofibromatosis showing severe distortion and acute angle curve. a. Anteroposterior view. b. Lateral view.

Table 5. Type of spinal deformity in neurofibromatosis cases.

Deformity	No. of patients	Per cent of patients
Scoliosis	44	43
Kyphosis	2	2
Kyphoscoliosis	53	55
Lordosis	0	0

after discontinuation of treatment. There has been little or no change in their condition even up to 10–23 years later. It is important to realize that only curves under 50° remain stable with little change. Among these patients there was a girl aged 14 years who had a typical scoliosis due to neurofibromatosis with a right thoracic curve between T_4 and T_{12} of 97°. She developed paraplegia at the age of 17 years at the level of T_8. Her symptoms were treated successfully with skeletal traction and cast. Examination was carried out 13 years after discontinuation of treatment. Her spine was stable with a curve of 67°. There were no neurological signs. She had married and had two healthy daughters, aged 3 and 5 years, who were without scoliosis. Such an observation was rare indeed.

Surgical treatment was carried out for all aggressive curves and also for all curves over 60°. Surgery consisted essentially of posterior fusion with cancellous bone grafting in 27 cases, with bone graft and Harrington instrumentation in 28 and with cancellous and cortical tibial bone graft (Cotrel special technique) in 11 cases (Figure 4). All patients had at least 9 months postoperative spinal immobilization in a body cast. Good fusion was achieved in all except two cases. Secondary operations were required for the repair of pseudoarthroses in these two cases.

Complications of operative treatment

1. Death in one case due to cardiac arrest immediately after application of the body cast.
2. Pseudoarthroses requiring a second operation in two cases.
3. Deep infection with re-exploration in one case; superficial infection in four cases.
4. Permanent paraplegia in two cases, one in each series. In both cases Harrington instrumentation with mild correction had been carried out.

Figure 3. Shows extensive destruction of the upper end of the humerus, ulnar and radius in a girl aged 9 years with severe thoracic scoliosis due to neurofibromatosis.

Table 6. Functional complaints of neurofibromatosis patients.

Complaint	No. of patients	Per cent of patients
Pain	23	20
Shortness of breath (Dyspnoea)	7	5
Deformity	52	52
Combination of the above complaints	16	18

Table 7. Location of the curvatures in neurofibromatosis scoliosis.

Location	No. of patients	Per cent of patients
Cervical	5	5
Thoracic	75	75
Thoraco-lumbar	10	10
Lumbar	0	0
Combination of the above locations	8	8

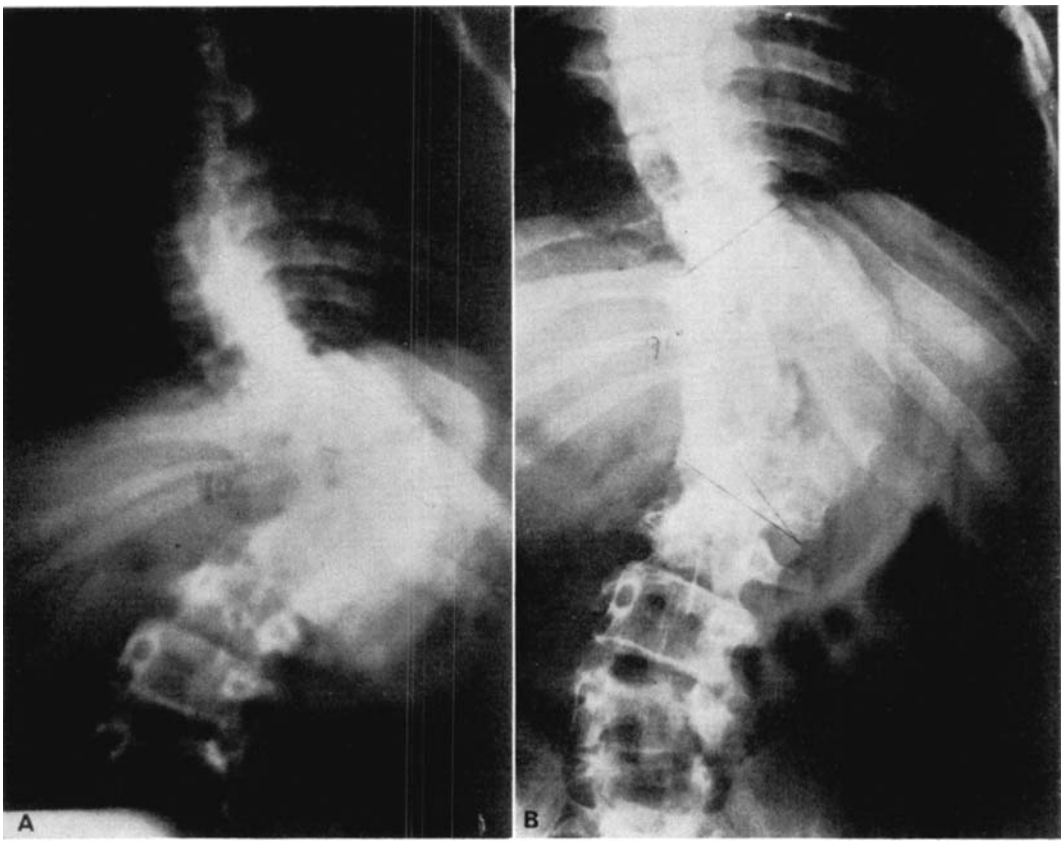

Figure 4. A typical acute angle curve in scoliosis due to neurofibromatosis. a. Preoperative. b. Postoperative. Note the heavy cancellous graft in the fusion area.

Table 8. Details of the scoliosis in neurofibromatosis.

	No. of patients	Per cent of patients
Convexity to the:		
Right	71	70
Left	8	8
Right and left	19	20
Measurement of the curve:		
$> 50°$	16	15
$< 50°$	82	85
No. of vertebrae involved:		
> 5	68	70
< 5	30	30
Hump:		
5 mm–15 cm	76	75
Rotation:	68	70
+ ++++		

Immediately after the operation paraplegia ensued and removal of the Harrington rod in the second case did not change the neurological state. In this case the actual tumour condition had eroded the lamina and the cord and dura were under the tumoral tissue. It is thought that paraplegia in this case was the result of vascular damage due to the removal of tumour tissue. However, from this bitter experience it was learnt that correction should never be done under general anaesthesia.

DISCUSSION

In this retrospective study of altogether 3209 scoliosis cases, 98 cases definitely due to neurofibromatosis were found. Thus the incidence of neurofibromatosis scoliosis in this series is just over 3 per cent. In the majority of cases the de-

formity of the spine appeared between 11 and 15 years of age. The minimum follow-up period was one year and the maximum was 23 years.

A study of the development of these 98 cases revealed that at least two distinct forms of scoliosis due to neurofibromatosis could be traced. A"moderate form" with a slow onset in the majority of cases, appears early in life between 5 and 10 years of age, and responds comparatively well to conservative measures. Some 30 per cent of the cases were in this category. The other form designated the "severe form" appears in most cases at a later age (10–15 years of age) and has a progressive onset. This form, even under the best orthopaedic management and bracing, deteriorates. It soon leads to severe kyphoscoliosis and unlike the idiopathic form it may lead to paraplegia. Unfortunately, it has not been possible to find definite criteria for differentiating these two forms.

Spinal fusion was the treatment of choice for all cases with progressive curves and also for all curves over 60°. The goal of surgery must be prevention of aggravation rather than the correction of aggravated curves. Correction of curves under anaesthesia may lead to irreversible paraplegia and regrettable results.

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