Surgical correction of severe dystrophic neurofibromatosis scoliosis: an experience of 32 cases

Wael Koptan · Yasser ElMiligui

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Abstract The surgical management of severe rigid dystrophic neurofibromatosis curves is a demanding procedure with uncertain results. Several difficulties are present in such patients including a poor bone stock, sharp angulation of these dystrophic curves and dural thinning or ectasia. The aim of this work was to review the clinical and radiographic outcomes of three-dimensional correction of severe rigid neurofibromatosis curves analyzing its efficacy, safety and possible complications. Thirty-two patients were followed up for an average of 6.5 years (range 3–9 years). The average age at surgery was 14 years (range 11–19 years). All patients had typical dystrophic curves, and the apex of the deformity was thoracic ($n = 13$), thoracolumbar ($n = 14$) and lumbar ($n = 5$). All patients had a two-staged procedure; an anterior release followed latter by posterior hybrid instrumentation augmented by sublaminar wires. Two wires were usually placed immediately below the proximal anchor, and several sublaminar wires were always passed at the apex of the deformity. There were a total of 142 wires with an average of 6.5 wires/patient (range 5–8 wires). The mean preoperative Cobb angle of the scoliotic curve was 102.2° (range 71° – 114°) corrected to an average of 39° (range 16° – 49°), and the loss of correction had an average of 4° . The mean

preoperative sagittal plane deformity was 49° corrected by an average of 61%, and rotation was corrected by an average of 34%. There were no dural tears during passage of the sublaminar wires, no implant-related complications and no permanent neurologic deficits. The use of extensive and vigorous anterior release with posterior hybrid instrumentation has proved useful and effective in the treatment of these severe deformities; sublaminar wires allow safe gradual correction and even distribution of forces over multiple anchor points improving the correction achieved and decreasing implant-related complications.

Keywords Neurofibromatosis · Scoliosis · Dystrophic curves · Hybrid instrumentation

Introduction

Neurofibromatosis is one of the most common genetic disorders, affecting 1/3,000 people [1]. Two clinical forms of neurofibromatosis are generally recognized: peripheral neurofibromatosis (NF Type 1) and central neurofibromatosis (NF Type 2) [2]. Spinal deformities were first reported in patients with NF Type 1 by Gould [3], and are considered its most common skeletal manifestation with an incidence ranging from 10 to 60% [4, 5].

The spinal deformity occurs in two forms: a regular curve similar to idiopathic scoliosis and a characteristic curve pattern described as a short (including 4–6 vertebrae), sharp angular curve, usually located in the thoracolumbar region and associated with dystrophic changes [6, 7]. The more severe the dystrophic changes, the more rapid are the deterioration and curve progression.

Several authors reported the need for early and aggressive surgical intervention for dystrophic curves due to the

W. Koptan · Y. ElMiligui
Department of Orthopaedic Surgery,
Faculty of Medicine, Cairo University, Cairo, Egypt

W. Koptan (✉)
60 Gezzirat El Arab Street, Mohandeseen, Geiza 12411, Egypt
e-mail: waelkoptan@yahoo.com

Y. ElMiligui
139 Tahrir Street, Dokki, Geiza 12411, Egypt
e-mail: el_miligui@yahoo.com

inability of brace treatment to halt their progression [7–9]. Nevertheless, surgical correction encountered several difficulties including a poor bone stock, sharp angulation of these curves, dural ectasia and the presence of spinal dislocation. Loss of correction and curve progression were reported even in the presence of solid fusion [10, 11].

A few series described the use of multisegmental posterior spinal instrumentation for correction of these deformities [12, 13]; however, little was mentioned on their use combined with multiple sublaminar wires whose passage was a major concern in those patients.

The aim of this work was to review the clinical and radiographic outcomes of surgical correction using multisegmental instrumentation, including multiple sublaminar wires, for severe rigid neurofibromatosis curves analyzing its efficacy, safety and possible complications.

Patients and methods

This study, approved by the ethics committee and the Institutional Review Board, included 32 patients with neurofibromatosis scoliosis who gave their informed consent to be incorporated in this research. It consisted of a consecutive series of 32 NF Type 1 patients with severe rigid spinal deformities treated between 1998 and 2006.

The study included 36 patients with neurofibromatosis scoliosis; 4 patients were lost during the follow-up period; the remaining 32 were followed for a minimum of 3 years. All patients presented with two or more criteria [14] to diagnose NF. The series included 18 males and 14 females with an average age of 14 years (range 11–19 years) and a positive family history in 8 patients (25%). Before being referred to us, 20 patients were ineffectively braced for an average period of 11 months (range 6 months–2 years).

Clinical examination included a thorough neurologic examination and assessment of curve flexibility. All patients were neurologically intact and presented with markedly rigid curves.

Radiologic examination included standard plain X-rays; frontal and sagittal curve measurements were made by Cobb's technique; apical axial rotation was determined by the method of Pedriolle and Vidal [15]. All patients had typical dystrophic curves and at least 3/5 of the following criteria including vertebral scalloping, penciling of the ribs, severe apical vertebral rotation, spindled transverse processes and foraminal enlargement. The apex of the deformity was thoracic ($n = 13$), thoracolumbar ($n = 14$) and lumbar ($n = 5$). MRI showed dural ectasia in six patients; there was no cord compression, intra spinal anomalies or neurofibromas.

All patients had a two-staged procedure; an anterior release/fusion followed by posterior correction/instrumentation

augmented by sublaminar wires. The procedure was performed at two sessions in the first 20 patients and as a single-staged procedure in the remaining 12.

The anterior release included as many levels as possible with an average of 4 levels (range 3–6 levels) centered over the apex of the deformity. The apical vertebrae were approached from the convex side through a standard approach, and the diaphragm was not divided except in thoracolumbar curves ($n = 14$); the released levels were only packed with morselized rib autograft in thoracic and thoracolumbar curves ($n = 27$) and morselized iliac crest autograft in lumbar curves ($n = 5$).

Posteriorly, extreme care was taken during exposure due to occasional thinning of the laminae. Posterior hybrid instrumentation included a claw on either side proximally, 2–6 pedicle screws distally; sublaminar wires were placed immediately below the proximal anchor, and several wires were placed at the apex of the deformity (Fig. 1). There were a total of 142 wires with an average of 6.5 wires/patient (range 5–8 wires). Aggressive facetectomies were executed; gradual correction was done with a combination of translation/derotation maneuvers. Meticulous decortication was performed and generous autografting using rib/iliac crest autograft. All patients had a positive wake-up test. Postoperatively, all patients were braced for an average of 3 months, and then gradually weaned off the brace.

Fig. 1 Intraoperative photograph of the construct configuration. It includes hooks, pedicle screws (a) and sublaminar wires (b) following final correction

Table 1 Comparison between Groups 1 and 2

	Group 1	Group 2
Number of patients	19	13
Operative notes		
Average blood loss (cc)	870	1,050
Average operative time (min)	375	415
Coronal plane		
Mean pre-op deformity (°)	106	98
Mean post-op deformity (°)	41	37
Average correction (%)	63	62
Average loss of correction (%)	2.5	4.5
Sagittal plane		
Mean pre-op deformity (°)	26	72
Average correction (%)	68	54
Average loss of correction (%)	2	4
Instrumented levels	12	13
Score for (SRS)-30 questionnaire	124	112

Results

Patients were followed up for an average of 6.5 years (range 3–9 years). The operative notes were analyzed, and the clinical and radiological results of surgery were evaluated including complications related to the surgical procedure. They were divided into two groups (Table 1) depending on their sagittal plane deformity: 19 patients had kyphosis less than 45° (Group 1) and 13 patients had kyphosis more than 45° (Group 2). There was no significant difference in average age, sex distribution or family history between both groups.

Operative notes

The total blood loss had a mean of 960 cc (range 650–1,500 cc), and was significantly less in Group 1 with an average of 870 cc (range 650–1,100 cc) compared to Group 2 with an average of 1,050 cc (range 725–1,500 cc) ($P < 0.01$). The total operative time had an average of 395 min (range 310–500 min), and was considerably less in Group 1 with an average of 375 min (range 310–425 min) than Group 2 with an average of 415 min (range 385–500 min) ($P < 0.01$).

Radiologically

The radiographs analyzed were those obtained preoperatively, immediate postoperatively and at the last follow-up.

Coronal plane

The mean preoperative scoliotic curve was 102° (range 71°–114°); Group 1 had an average of 106° (range

75°–114°) while Group 2 had an average of 98° (range 71°–111°) ($P = 0.09$). All scoliotic deformities had a flexibility index of less than 40% on bending films. The mean postoperative deformity had an average of 39° (range 16°–49°); Group 1 (Fig. 2a–d) had an average of 41° (range 38°–49°) yielding 63% correction; Group 2 (Fig. 3) had an average of 37° (range 16°–48°) yielding 62% correction ($P > 0.99$). At final follow-up, there was an average of 2.5% correction loss in Group 1 compared with 4.5% in Group 2 ($P < 0.001$).

Sagittal plane

The mean preoperative sagittal plane deformity was 49° (range 2°–91°); Group 1 (included mainly thoracic and thoracolumbar curves) had an average of 26° (range 2°–43°) while Group 2 (included mainly thoracolumbar and lumbar curves) had an average of 72° (range 45°–91°) ($P < 0.01$). The overall correction had an average of 61%; Group 1 (Fig. 2e–f) had an average correction of 68%, while Group 2 (Fig. 3) had an average correction of 54% ($P < 0.001$). At final follow-up, loss of correction had an average of 2% in Group 1 compared with 4% loss in Group 2 ($P < 0.01$).

Rotation

The apical vertebral body rotation was corrected by an average of 34% with no significant difference in correction between Group 1 and 2.

Instrumentation

There was no significant difference in the number of instrumented levels or number of sublaminar wires used; Group 1 had an average of 12 levels (range 8–15) while Group 2 had an average of 13 levels (range 8–14) ($P = 0.09$); there were a total of 142 wires with an average of 6.5 wires/patient (range 5–8 wires).

Clinically

At final follow-up, the scores of the Scoliosis Research Society (SRS)-30 questionnaire in Group 1 ranged from 92 to 140 with an average of 124 and for Group 2 ranged from 88 to 128 with an average of 112 ($P < 0.001$).

Complications

- Two patients had superficial infection that responded well to antibiotics. One patient had a deep wound infection that required surgical debridement and copious irrigation.

◀ **Fig. 2** A 16-year-old female with a left dystrophic curve (**a, b**) pre-op and follow-up AP X-rays revealing a 113° scoliosis corrected down to 40° (**c, d**) pre-op and follow-up PA photographs (**e, f**) pre-op and follow-up lateral X-rays revealing a 7° thoracic hypokyphosis corrected to 29° (**g, h**) pre-op and follow-up lateral photographs

- Two patients had a definite pseudoarthrosis presenting after 6 and 10 months, respectively, with persistent pain without instrumentation failure; tomography confirmed the diagnosis; exploration and re-grafting were performed, and the rest of the follow-up was uneventful.
- There were no permanent neurologic injuries. One patient experienced a transient weakness in the left lower limb that was not detected during the wake-up test; it fully recovered within 3 months.
- There were no dural tears during passage of the sublaminar wires, and there were no metal failures or implant-related complications.

Discussion

The surgical management of spinal deformities in neuro-fibromatosis is a major challenge with a high incidence of pseudoarthrosis and curve progression [16, 17]; the presence of dystrophic changes, the magnitude of the scoliotic deformity and sagittal plane alignment are of utmost importance in determining the ideal surgical approach for their treatment [8, 9, 17]. Patients with a scoliotic deformity of 20°–40° and kyphosis <50° are simply managed with posterior spinal instrumentation and fusion [2]. On the other hand, management of severe rigid scoliosis associated with hyperkyphosis is extremely demanding with uncertain outcome.

Before the introduction of third generation spinal implants, surgical correction of dystrophic curves had disappointing results; several authors reported a poor outcome even with the use of Harrington instrumentation in some of their patients. In these series, overall results were collectively reported without segregating patients who had posterior instrumented fusions. Sirois and Drennan [6] reported a 38% incidence of pseudoarthrosis in 9 out of 23 patients with severe dystrophic spinal deformities and an average curve progression of 12.7°; an overall failure of 72% occurred in patients with a kyphotic deformity of >50°. Similarly, Winter et al. [7] reported a 64% incidence of failure when curves with a kyphotic deformity of >50° were treated with posterior fusion alone; to the contrary, a fusion rate of 80% occurred when additional anterior fusion was performed.

Later, Hsu et al. [10] reported a 7.5% incidence of failure in 13 dystrophic curves managed by combined

Fig. 3 A 10-year-old male with a right dystrophic curve (a, b) pre-op and follow-up AP X-rays revealing an 80° scoliosis corrected down to 17° (c, d) pre-op and follow-up lateral X-rays revealing a 79° thoracolumbar kyphosis corrected to 7° (e, f) pre-op and follow-up PA photographs

anterior and posterior fusion; all five patients with angular kyphoscoliosis had progression of the deformity during their follow-up. The largest series by Parisini et al. [18] included 56 patients with an overall fusion failure of 53% in patients who had posterior instrumented fusion alone compared to 23% who had an additional anterior fusion. Moreover, kyphoscoliotic patients ($n = 31$) who were managed posteriorly alone had a fusion failure of 63%.

The results of surgical correction considerably improved with the advent of Cotrel-Dubousset multisegmental instrumentation. It was first reported by Holt and Johnson [12] in a series of five patients; their scoliosis ranged from 50° to 110° and kyphosis ranged from 20° to 67°. Despite instrumentation and fusion, three patients showed a progression of their scoliosis. In the same year, Shufflebarger [13] reviewed the results of 12 patients, and achieved 69% frontal correction, 10° of sagittal improvement and 33% axial derotation. Loss of correction was minimal (mean 0.8°) with no pseudoarthrosis. The majority of his patients

(10/12) had non-dystrophic curves which can justify these excellent results.

To our knowledge, very limited series [19, 20] were ever since reported on the use of multisegmental instrumentation; the most recently available by Li et al. [21] reported 19 patients with NF-1 treated surgically with long, posterior instrumented fusion. Sixteen had dystrophic curves in which the Cobb's angles in the coronal and sagittal planes before, after surgery and at follow-up were 68° and 31°, 27° and 28°, and 33° and 30°, respectively. The mean intraoperative blood loss was 807 ml, and the mean duration of surgery was 190 min. There were no cases of coronal or sagittal decompensation, neurologic complications, or infections. There were eight (42.1%) complications; pseudoarthrosis with instrumentation failure that required revision surgery occurred in one (5.2%) patient.

Our series included 32 patients with severe rigid dystrophic curves of a mean of 102° (range 71°–114°). Although we did not find a published definition of 'severe'

curves in NF patients, we relied on the work of Lenke et al. [22] who defined it as a Cobb angle $\geq 70^\circ$ in AIS patients; later, Greiner et al. [23] reported that AIS patients did not exhibit clinically significant respiratory symptoms until their curves were 60° – 100° , thus defined severe scoliosis as Cobb angle $>60^\circ$. Rigid scoliosis was defined as a coronal deformity with less than 40% flexibility index on bending films.

Despite the severity and rigidity of the deformity, both groups similarly yielded a correction comparable to the best achieved by other authors, as well as the few degrees of correction lost at follow-up. Group 2 patients had significantly more loss of correction in both coronal and sagittal planes. On the other hand, Group 1 patients had a significantly less total blood loss and total operative time and a better score of the Scoliosis Research Society (SRS)-30 questionnaire.

Due to the severity of the scoliotic deformity, all patients first had an anterior release and grafting. Anterior arthrodesis was performed not only to increase the fusion rate but also to prevent the development of crankshaft phenomenon in skeletally immature patients. Anterior approaches to the spine are associated with several complications including pulmonary, vascular and even sexual complications, with a rate varying between 50 and 100% [24–26]. In our study, we had no similar complications. Our total blood loss was acceptable, and excessive bleeding from the plexiform venous channels and consequent postoperative hemorrhage were avoided by meticulous hemostasis.

We had only 5/32 complications (15.6%) with no metal failures or implant-related complications. Only two patients (6.2%) had a definite pseudarthrosis that was successfully managed. The low incidence of pseudarthrosis can possibly be due to multiple factors; this included the combined approach, multisegmental instrumentation and the extreme care taken in preparing the bone graft bed. Although some authors recommend routine tomographic evaluation 6 months postoperatively [7, 9, 27], we only used tomography in these two patients. We only performed a wake-up test in all patients, and did not experience any permanent neurologic deficits; only one patient experienced a transient weakness which, although missed during the wake-up test, later fully recovered. Unfortunately, the use of continuous spinal cord monitoring was not available.

A major difficulty encountered in NF1 patients is the poor bone stock and remarkable osteoporosis [28, 29] which can result in metal failure, hook dislodgment and consequent loss of correction [19, 30]. On the other hand, several series with long follow-up have showed increased rate of pseudarthrosis in NF patients [11, 16, 17]. Our use of multisegmental third generation instrumentation systems allowed multiple anchor points to correct these severe rigid curves. Sublaminar wires were extensively placed

immediately below the proximal claw and at the apex of the deformity, with an average of 6.5 wires/patient. Sequential tightening of these wires allowed gradual correction without cutting through the laminae and provided even distribution of forces over multiple anchor points. The presence of dural ectasia was not a problem, and no dural tears occurred during passage of sublaminar wires. All pedicle screw constructs were not used because of the extremely thin pedicles in NF patients and the lack of gradual correction provided by hybrid instrumentation using sublaminar wires; these hybrid constructs allowed excellent correction with relatively simple and inexpensive instrumentation.

Conclusion

The greater the kyphotic component of the deformity reflected a significantly higher loss of correction in both coronal and sagittal planes, more blood loss, longer operative time and a worse score of the Scoliosis Research Society (SRS)-30 questionnaire.

The use of combined approach for the management of severe dystrophic spinal deformities is mandatory. Extensive and vigorous anterior release with posterior hybrid instrumentation, including multiple sublaminar wires, has proved efficiency in the treatment of these difficult cases. Despite the recent trend for all pedicle screw constructs, sublaminar wires provide a cost-effective useful alternative.

Conflict of interest statement None of the authors has any potential conflict of interest.

References

1. Littler M, Morton NE (1990) Segregation analysis of peripheral neurofibromatosis (NF1). *J Med Genet* 27:307–310
2. Kim HW, Weinstein SL (1997) Spine update: the management of scoliosis in neurofibromatosis. *Spine* 22:2770–2776
3. Gould EP (1918) The bone changes occurring in von Recklinghausen's disease. *Q J Med* 11:221–228
4. Akbarnia BA, Gabriel KR, Beckman E, Chalk D (1992) Prevalence of scoliosis in neurofibromatosis. *Spine* 17(Suppl):244–248
5. Funasaki H, Winter RB, Lonstein JB, Denis F (1994) Pathophysiology of spinal deformities in neurofibromatosis. *J Bone Joint Surg Am* 76:692–700
6. Sirois JL, Drennan JC (1990) Dystrophic spinal deformity in neurofibromatosis. *J Pediatr Orthop* 10:522–526
7. Winter RB, Lonstein JE, Anderson M (1988) Neurofibromatosis hyperkyphosis: a review of 33 patients with kyphosis of 80° or greater. *J Spinal Disord* 1:39–49
8. Savini R, Vincenzi G (1976) Deformities of the spine in neurofibromatosis. Clinical and radiographic study of 46 cases. *Ital J Orthop Traumatol* 2:37–50
9. Winter RB, Moe JH, Bradford DS, Lonstein JE, Pedras CV, Weber AA (1979) Spine deformity in neurofibromatosis. A review of 102 patients. *J Bone Joint Surg Am* 61:677–694

10. Hsu LCS, Lee PC, Leong JCY (1984) Dystrophic spinal deformities in neurofibromatosis treated by anterior and posterior fusion. *J Bone Joint Surg Br* 66:495–499
11. Wilde PH, Upadhyay SS, Leong JCY (1994) Deterioration of operative correction in dystrophic spinal neurofibromatosis. *Spine* 19:1264–1270
12. Holt RT, Johnson JR (1989) Cotrel-Dubousset instrumentation in neurofibromatosis spine curves. *Clin Orthop* 245:19–23
13. Shufflebarger HL (1989) Cotrel-Dubousset instrumentation in neurofibromatosis spinal problems. *Clin Orthop* 245:24–28
14. National Institutes of Health Consensus Development Conference (1988) Neurofibromatosis. Conference statement. *Arch Neurol* 45:575–578
15. Pedriolle R, Vidal J (1987) Morphology of scoliosis, three dimensional evaluation. *Orthopedics* 10:909–915
16. Betz RR, Iorio R, Lombardi AV, Clancy M, Steel HH (1989) Scoliosis surgery in neurofibromatosis. *Clin Orthop* 245:53–56
17. Calvert PT, Edgar MA (1989) Scoliosis in neurofibromatosis. *J Bone Joint Surg Br* 71:246–251
18. Parisini P, DiSilvestre M, Greggi T, Paderni S, Cervellati S, Savini R (1999) Surgical correction of dystrophic spinal curves in neurofibromatosis. *Spine* 24:2247–2253
19. Vandenbroucke J, van Ooy A, Geukers C, van der Linden AJ, Hoogmartens M (1997) Dystrophic kyphoscoliosis in neurofibromatosis type I: a report of two cases and review of the literature. *Eur Spine J* 6:273–277
20. Singh K, Samartzis D, An HS (2005) Neurofibromatosis type I with severe dystrophic kyphoscoliosis and its operative management via a simultaneous anterior-posterior approach: a case report and review of the literature. *Spine J* 5:461–466
21. Li M, Fang X, Li Y, Ni J, Gu S, Zhu X (2009) Successful use of posterior instrumented spinal fusion alone for scoliosis in 19 patients with neurofibromatosis type-1 followed up for at least 25 months. *Arch Orthop Trauma Surg* 129(7):915–921
22. Lenke LG, Bridwell KH, Blanke K, O'Brien MF, Baldus C (1994) Preoperative spinal canal investigation in adolescent idiopathic scoliosis curves $\geq 70^\circ$. *Spine* 19:1606–1610
23. Greiner KA (2002) Adolescent idiopathic scoliosis: radiologic decision-making. *Am Fam Physician* 65:1817–1822
24. Moe JH, Purcell GA, Bradford DS (1983) Zielke instrumentation (VDS) for the correction of spinal curvature: analysis of results in 66 patients. *Clin Orthop* 180:133–153
25. Kostuik JP, Enico TJ, Gleason TF (1986) Techniques of internal fixation for degenerative conditions of the lumbar spine. *Clin Orthop* 203:219–231
26. Ogilvie JW (1988) Anterior spine fusion with Zielke instrumentation for idiopathic scoliosis in adolescents. *Orthop Clin North Am* 19:313–317
27. Crawford AH, Parikh S, Schorry EK, Von Stein D (2007) The immature spine in type-1 neurofibromatosis. *J Bone Joint Surg Am* 89:123–142
28. Illes T, Halmay V, de Jonge T, Dubousset J (2001) Decreased bone mineral density in neurofibromatosis patients with spinal deformities. *Osteoporos Int* 12:823–827
29. Elefteriou F, Kolanczyk M, Schindeler A, Viskochil DH, Hock JM, Schorry EK, Crawford AH, Friedman JM, Little D, Peltonen J, Carey JC, Feldman D, Yu X, Armstrong L, Birch P, Kendler DL, Mundlos S, Yang FC, Agiostratidou G, Hunter-Schaedle K, Stevenson DA (2009) Skeletal abnormalities in neurofibromatosis type 1: approaches to therapeutic options. *Am J Med Genet A* 149A(10):2327–2338
30. Tsirikos AI, Saifuddin A, Noordeen MH (2005) Spinal deformity in neurofibromatosis type-1: diagnosis and treatment. *Eur Spine J* 14:427–439