

Case Reports & Case Series

Management of a very severe Dystrophic Kyphoscoliosis on a patient with
Type 1 Neurofibromatosis - CASE report

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ABSTRACT

Introduction: Neurofibromatosis (NF) is a group of multisystem diseases with an autosomal dominant inheritance pattern. It affects the neural crest cells, being able to compromise all tissues. NF type 1 is the most prevalent, also known as the peripheral type. This includes multiple musculoskeletal manifestations, such as severe and difficult-to-manage scoliosis. Including two types: dystrophic and non-dystrophic.

Case report: 40-year-old man with a history of NF1, grave spine deformity, extreme back pain, and severe functional limitation. The initial diagnostic images of the spine showed a thoracolumbar kyphoscoliosis over 80 degrees which collapsed the thoracolumbar junction and generated spinal cord compression. He was managed with a Vertebral Column Resection (VCR) at the thoracolumbar junction level, thoracoplasty, and posterior thoracic to lumbosacral arthrodesis. The patient required hospitalization and long rehabilitation therapy. Currently, he has good clinical and diagnostic imaging results after one year of following.

Discussion: This patient's case corresponds to dystrophic kyphoscoliosis. This group of patients requires more aggressive surgical management, considering the high risk of progression and deterioration even after surgery.

1. Introduction

Neurofibromatosis (NF) is a group of multisystem diseases with an autosomal dominant inheritance pattern [1], affecting 2500 individuals per year [2]. It is the most common single-gene hereditary disease in humans [3]. Despite, this being a disorder of the neural crest cells [4], it can affect tissues from the three primary germ layers: ectoderm, mesoderm, and endoderm [1,3].

There are two clinical types of NF: type 1 (NF1) or peripheral and type 2 (NF2) or central [1,3,4]. NF1 is the most prevalent type, associated with a mutation on the gen 17q11.2. This gene codes for a protein called Neurofibromin. This protein is expressed in multiple cell groups and down-regulates the Ras signal transduction pathway, causing a decrease in cell proliferation [5].

Clinically, this disease has variable expressions from mild cutaneous

lesions to life-threatening conditions [2,5]. Among the most affected tissues is the musculoskeletal system, with focal or systemic presentations [1,6]. These include bone demineralization disorders, osteomalacia, short stature, and macrocephaly. Spinal deformity and skeletal dysplasia are less frequent presentations but with higher morbidity and mortality [1,6].

The most frequent focal musculoskeletal manifestation is scoliosis, highly associated with dystrophic kyphosis mainly in the thoracic region [4]. Scoliosis in patients with NF1 can be classified into two groups: non-dystrophic and dystrophic. It must also be differentiated from idiopathic scoliosis, which has similar imaging findings to dystrophic scoliosis. This classification is so important to determine prognosis and treatment [1,4,7].

Dystrophic scoliosis is the uncommon type but is much more resistant to treatment. It is frequent that despite spinal fusion, dystrophic

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curves can progress [3]. In trying to avoid the progression of these patients after surgery, different management has been proposed. Posterior-only approaches could be considered especially when the deformity measures less than 50° [3,7,8]. In cases where the curve exceeds 50°, the anterior/posterior approach with intervertebral body fusion is described more frequently; including severe thoracic kyphosis [8,9]. These surgeries can be very extensive and include different techniques, materials, and a highly specialized team to correct spinal deformity. Moreover, these patients experience preoperative limitations in pulmonary function, which increases the susceptibility to anesthetic complications, causing performing this surgery carries a high perioperative risk [10]. The following article corresponds to a case report of a patient with severe kyphoscoliosis in the context of NF1, his surgical management, and outcomes, carried out in Bogota-Colombia.

2. Case report

2.1. Initial assessment

A 40-year-old man with a history of NF1 and ischemic heart disease arrives for a neurosurgery assessment for grave spine deformity, severe back pain, neurological claudication, and postural inability. On physical examination, he presented many *café au lait* spots over the skin, a big hump, and limitation of flexion–extension movements, without significant compromise of muscular strength, sensitivity, or stretch reflexes.

The initial CT scan (Fig. 1) of the spine showed a structured right vertex thoracolumbar kyphoscoliosis over 80 degrees which collapsed the thoracolumbar junction. And the MRI (Fig. 2) showed a spinal cord compression that could explain the disability of this patient but questioned the possibility of moving the legs without neurological deficit. He already had a history of thoracic laminoplasty years ago performed in another hospital.

A neurosurgical meeting was taken, where the patient was considered a candidate for surgical management with Vertebral Column Resection (VCR) at the thoracolumbar junction level, right

thoracoplasty, and thoracic to lumbosacral arthrodesis.

3. Surgery and perioperative process

The first surgery was performed in May 2022 by two expert neurosurgeons with a qualified surgical team. Severe thoracolumbar scoliosis, kyphosis deformity with meningocele, and severe cord compression were described as intraoperative findings.

With the patient in a prone position, a single midline incision was made from T7 to L3. They began with posterior spinal instrumentation at a thoracic level from T7. Decompression of the spinal cord was made by laminectomy and foraminotomy from T11 to L3. Followed by a posterior column osteotomy at the level of T11 to L2. Then bilateral extra cavitory approach surgery T11-T12-L1-L2-L3 was made to perform resection of the transverse process, head of ribs, and finally, corpectomy at those vertebral levels, preserving the anterior longitudinal ligament. An expandable spinal cylindrical cage with autologous and heterologous bone grafts was anchored at the level of T10 and L4. Followed by reduction maneuvers to achieve adequate alignment of vertebral bodies, as well as instrumented posterior arthrodesis with semi-polyaxial screws and cobalt chrome bars.

Finally, the transpedicular instrumentation of the lumbosacral spine was completed, including S2 alar iliac pelvic fixation (Fig. 3). The decision to use iliac screws was due to the severity of the coronal and sagittal imbalance, severe pelvic retroversion, and the proximal extension of the surgery. Additionally to the incipient involvement of the L5-S1 intervertebral disc.

Both coronal and sagittal deformity correction was achieved by direct and indirect maneuvers and reinforced with double lateral bars anchored with connectors. Dural tears were corrected by autologous grafts and fibrin sealants. The total time of surgery was 17 h, with 1500 cc of bleeding whence, required a transfusion of 2 units of red blood cells and an albumin unit (Fig. 4).

Neuromonitoring was used throughout the procedure to ensure spinal cord and nerve integrity. Post-surgery, the patient was transferred to

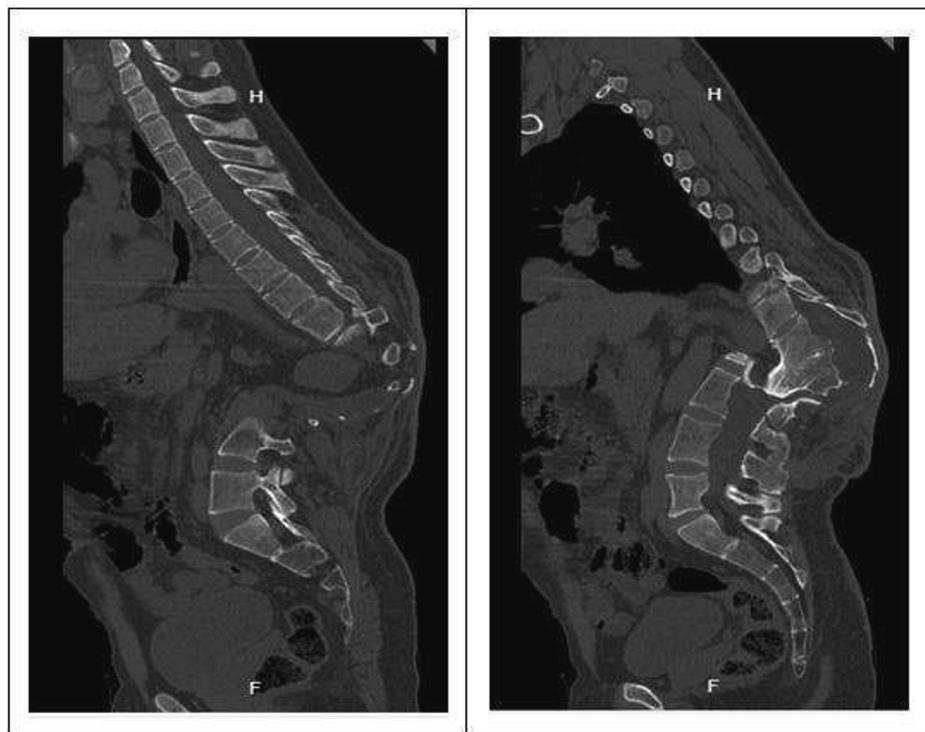


Fig. 1. Sagittal plane of preoperative CT scan. A severe sagittal deformity is shown, particularly at the thoracolumbar junction, with significant compromise of the spinal canal.

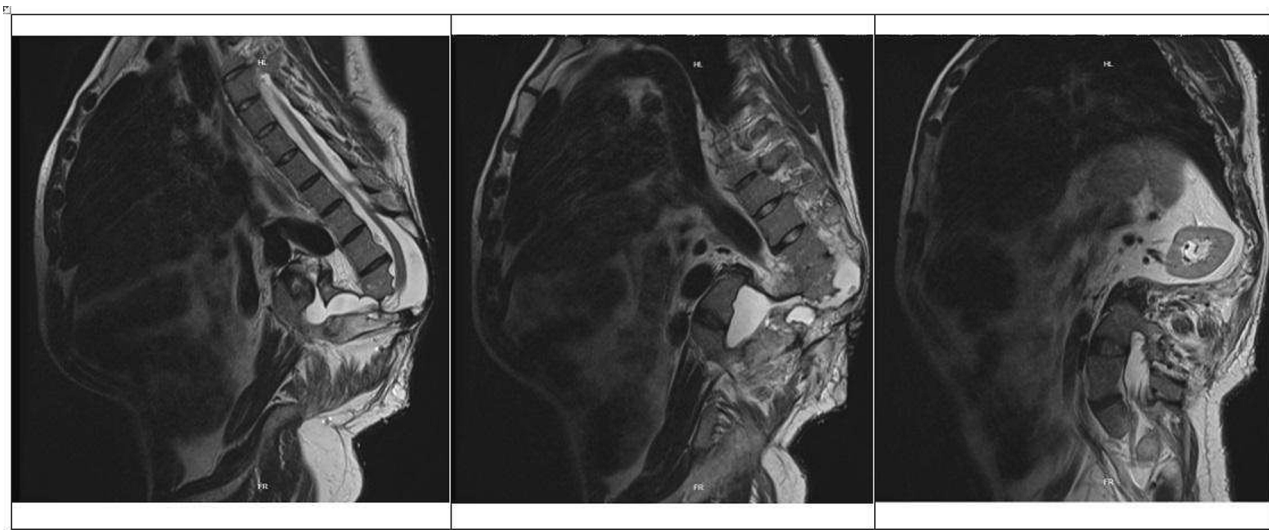


Fig. 2. Sagittal plane of preoperative MRI. The compromise of the spinal canal is even more evident, with the magnitude of the sagittal and even coronal deformity.

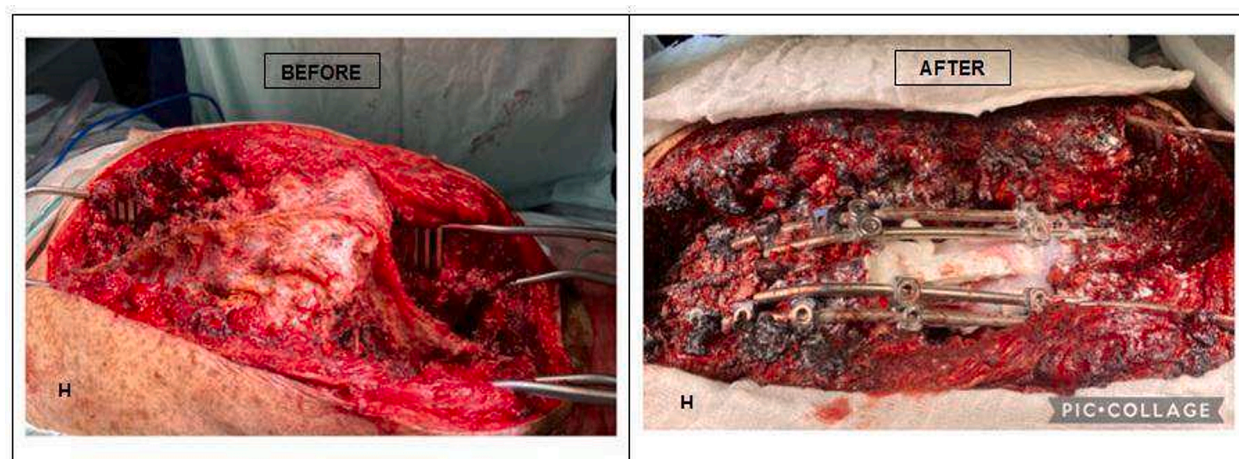


Fig. 3. Intraoperative view before and after vertebrectomy and spinal instrumentation, demonstrating immediate correction of the deformity.

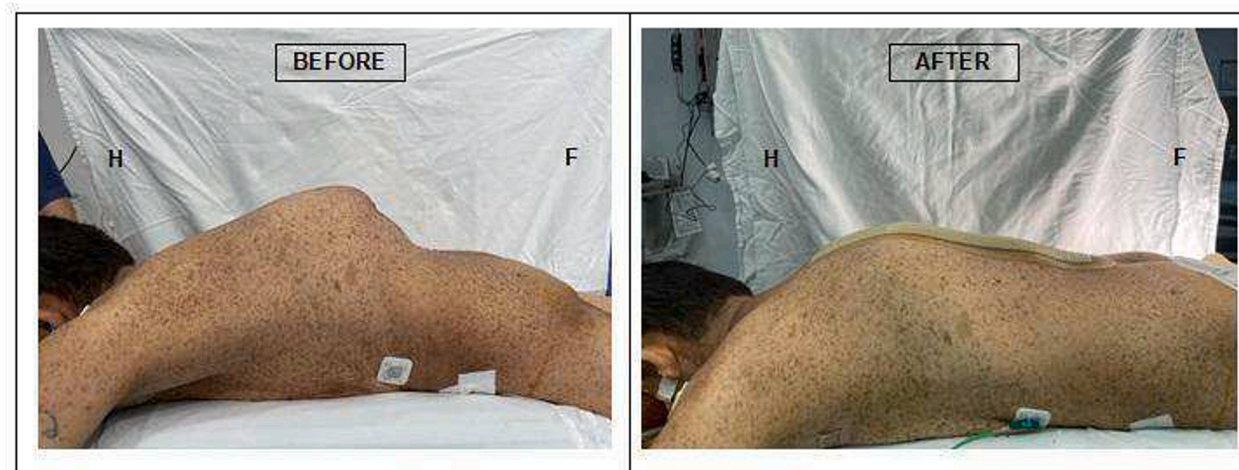


Fig. 4. Patient on the surgical table just before and after surgery, showing the correction of the spinal deformity.

the ICU and received sedation, mechanical ventilation, and comprehensive rehabilitation. Lower limb weakness, intact sensory function, and urinary retention were observed. A postoperative CT scan confirmed the successful correction of the sagittal spine deformity using an expandable spinal cylindrical cage (Fig. 5).

Approximately 5 days after the initial intervention, the patient developed a cerebrospinal fluid (CSF) fistula through the surgical wound, despite treatment with Acetazolamide. As a result, the patient underwent placement of a CODMAN HAKIM ventriculoperitoneal shunt. Should be considered that any patient undergoing multilevel vertebrectomy with dural sac reconstruction, such as this case, may require CSF diversion from the first surgical procedure.

The patient also required increased analgesic medications and follow-up care at the wound clinic. Twelve days following the initial surgery, he was discharged from the hospital with instructions for at-home rehabilitation.

However, one week later, the patient returned to the clinic with a profuse CSF fistula through the surgical wound. The patient underwent surgical closure of the fistula by duroplasty, wound debridement, and placement of a Hemovac drain. During the procedure, secretion cultures were taken. Initially, the patient was treated with Cefepime and Vancomycin antibiotics because a deep tissue infection was suspected. However, the cytochemical study of the CSF was not compatible with an infection, in addition to the negative result of the cultures taken during surgery. Instead, the CSF showed an abundant amount of proteins, which given the magnitude of the dural sac defect or “dead space” post-correction of the deformity, was considered to correspond to a strong healing process.

The patient was discharged to continue physical and occupational therapy at home. He was also prescribed nutritional supplements. At that moment, the patient had $2/7$ proximal strength and $3/5$ distal strength in the lower limbs, with normal strength and sensory in the upper limbs. He was able to stand with the support of a walker and orthoses but had limitations during gait.

4. Follow-up

The patient has completed a 1-year follow-up period, his condition has shown marked improvement during follow-up at the outpatient clinic. He has demonstrated the ability to walk with a walker and climb some stairs. His overall quality of life has improved with reduced pain and increased mobility. While the patient has made significant progress, he continues to undergo daily therapy to further enhance his rehabilitation.

Nine months after the surgery, control radiographs (Fig. 6) were taken which confirm the success of the correction achieved and maintained in this challenging case of severe deformity. Although residual thoracic kyphosis is still evident, the correction to the initial deformity is highly satisfactory. The patient has experienced a significant improvement in sagittal balance and remains free from axial pain one year after surgery.

5. Discussion

Orthopedic disorders are commonly observed in around 50% of patients with NF1, according to studies [7]. However, the exact cause of spinal deformities in these patients remains unclear. Some researchers speculate that it may be related to localized neurofibromas eroding or infiltrating the bone, bone metabolic deficiencies, osteomalacia, or mesodermal dysplasia [1,2]. Further research is needed to better understand the underlying mechanisms of these orthopedic disorders in patients with neurofibromatosis.

NF1-associated scoliosis is classified into two types: dystrophic and non-dystrophic [1,2,7]. Non-dystrophic scoliosis presents findings similar to those of Adolescent idiopathic scoliosis (AIS) but with an earlier age of onset, worse prognosis, and higher risk of developing

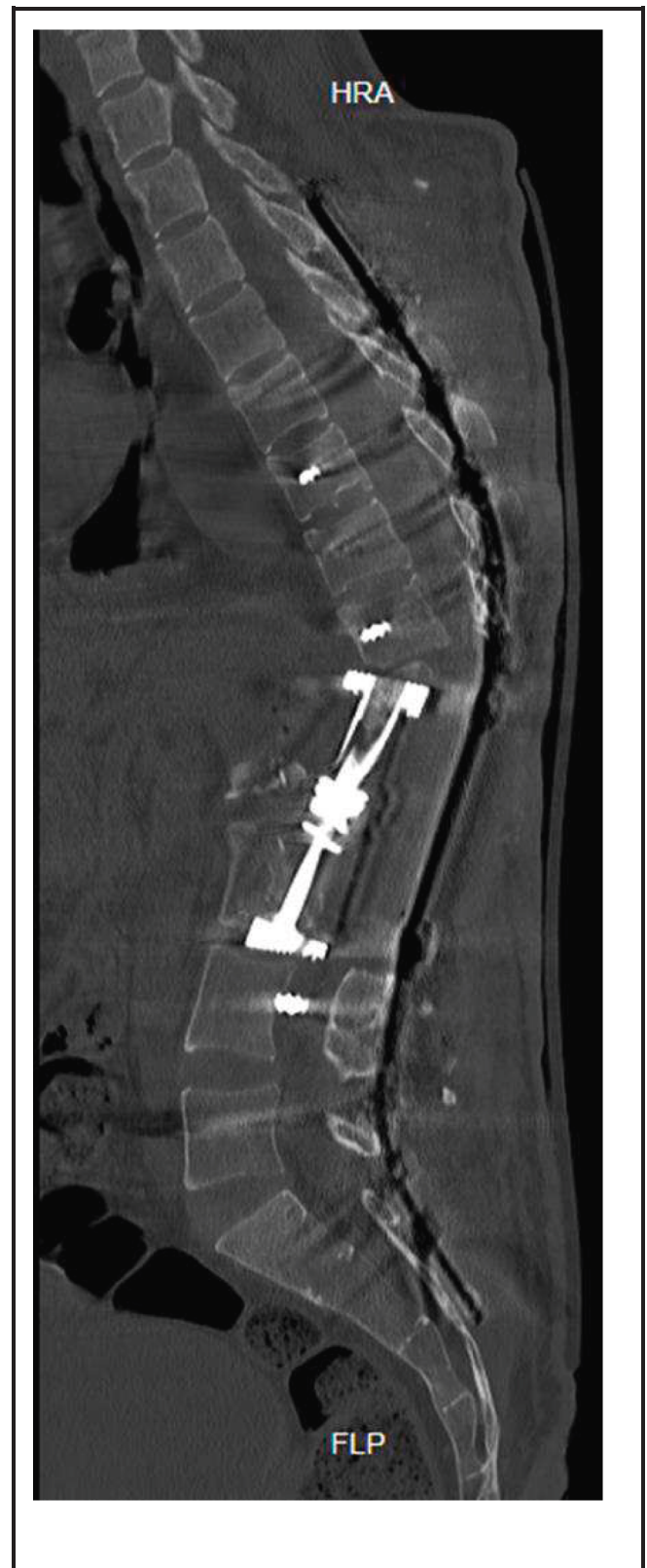


Fig. 5. Sagittal view of postoperative CT scan showing arthrodesis material, corpectomies, and expandable spine cage.

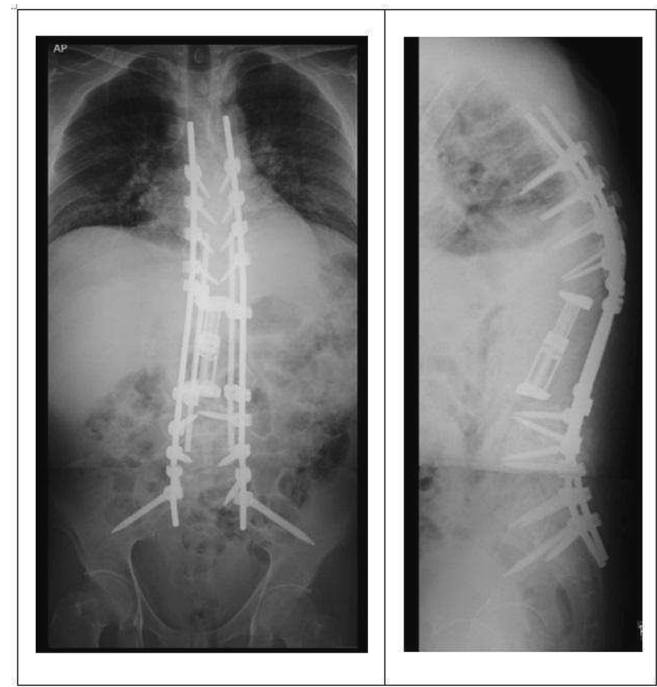


Fig. 6. Postoperative radiographs show evidence of improvement in coronal and sagittal balance, as well as the proper placement of the arthrodesis material.

pseudoarthrosis or fusion failure. In both non-dystrophic and AIS, careful surgical planning and correction of sagittal alignment are imperative to ensure long-term outcomes [11]. Dystrophic scoliosis, on the other hand, is less common but much more aggressive and resistant to surgical management. It is characterized by multiple morphological changes, such as spinal cord dural ectasia, wedging of the vertebral body, and vertebral scalloping, among others.

Due to the high risk of progression, even after surgical treatment, dystrophic curves must be treated aggressively. Therefore, surgical management is recommended as soon as possible [4,12]. Dystrophic scoliotic curves measuring less than 20 degrees should be closely monitored every six months to detect any progression and allow for timely surgical management. Curvatures between 20 and 40 degrees, with less than 50 degrees of kyphosis, may be treated with posterior spinal arthrodesis using segmental fixation [1,2,7].

The patient in this case report presents most of the criteria for dysplastic scoliosis outlined in Table 1 [7,12]. One of the most challenging aspects of this type of deformity is the risk of progression even after fusion. Therefore, an extensive arthrodesis supported by a double bar was performed as an aggressive treatment. The surgical correction achieved has been maintained for ten months of follow-up, as evidenced by the control radiographs (Fig. 6). However, this patient will require continued medical check-ups for years due to the strong tendency of dystrophic scoliosis to progress even after spinal fusion [7,12].

Based on this experience, an aggressive surgical approach, such as extensive arthrodesis supported by a double bar and ensuring solid 360° spinal instrumentation when VCR involves four or more vertebral levels, can significantly improve the results. The decision to perform multilevel VCR should be considered when the spinal deformity, regardless of its etiology, presents a short acute angle kyphosis with significant sagittal imbalance.

Additionally, due to the high probability of CSF fistulas resulting from dural sac compromise, aggressive and early management is necessary. Finally, the management of dead space resulting from the resection of the multiple vertebral levels should be considered, such as utilizing the turnover pedicled latissimus dorsi muscle flap for

Table 1
Typical dysplastic changes evident on radiographs in patients with Neurofibromatosis type 1 .

Table 1: Typical dysplastic changes evident on radiographs in patients with Neurofibromatosis type 1. Adapted from [4,7]	Items present in the patient
Vertebral scalloping associated with dural ectasy	Yes
Transverse process spindling	Yes
Vertebral wedging	Yes
Paravertebral soft tissue mass	Yes
Short curve (five vertebral levels) with severe apical rotation	Yes
Sagittal plane deformities: angular apical thoracic kyphosis	Yes
Intervertebral foraminal enlargement	No
Widened interpedicular distances	No
Dysplastic pedicles	Yes

Adapted from [4,7]

reconstruction [13].

6. Conclusion

The management of severe spinal deformities associated with NF1 presents a surgical challenge. Available treatment options, including staged surgical decompression and reconstruction, are necessary, distinguishing between dystrophic and non-dystrophic scoliosis is crucial for providing appropriate treatment.

Severe spine deformities require an aggressive surgical approach with solid spinal instrumentation. It is recommended to take into account secondary issues related to the surgical intervention of these pathologies, such as dural sac compromise and management of dead space.

This surgical strategy combined with comprehensive rehabilitation can significantly improve the sagittal balance and patient's quality of life. Nonetheless, every patient's condition is unique, and careful consideration of all available treatment options is necessary to develop an individualized management plan.

Ethical considerations

This case involves a patient who has reached the age of majority and has preserved cognitive function. The patient is fully aware of the nature, benefits, and risks of the research, as well as the academic interest of publishing his medical history. Informed consent procedures were carried out by the directives of the International Medical Ethics Committee.

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Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

References

[1] A.H. Crawford, M.G. Lykissas, E.K. Schorry, S. Gaines, V. Jain, T. Gregg, D. Viskichil, Neurofibromatosis: Etiology, commonly encountered spinal deformities, common complications and pitfalls of surgical treatment, *Spine Deform* [Internet]. 1 (1) (2012) 85–94, <https://doi.org/10.1016/j.jspd.2012.04.004>. Available from: .

[2] G. Price, M.L. Martini, J.M. Caridi, D. Lau, E.K. Oermann, S.N. Neifert, A Nationwide study characterizing the risk and outcome profiles of multilevel fusion procedures in neuromuscular scoliosis patients with neurofibromatosis Type 1, *World Neurosurg* [Internet]. (2023) 1–11. Available from: 10.1016/j.wneu.2022.12.068.

[3] S. Cai, Y. Tian, G. Qiu, J. Zhang, J. Shen, H. Zhao, Y. Zao, Neurofibromatosis Type 1 with severe dystrophic kyphosis: Surgical treatment and prognostic analysis of 27 patients, *Orthop Surg.* 12 (6) (2020) 1923–1940.

[4] A.I. Tsirikos, A. Saifuddin, M.H. Noordeen, Spinal deformity in neurofibromatosis type-1: Diagnosis and treatment, *Eur Spine J.* 14 (5) (2005) 427–439.

- [5] M. Sial, K.J. George, A Review of Spinal Lesions in Neurofibromatosis Type 1 in a Large Neurofibromatosis Type 1 Center, *World Neurosurg* [Internet]. 169 (2023) e157–e163. Available from: [10.1016/j.wneu.2022.10.100](https://doi.org/10.1016/j.wneu.2022.10.100).
- [6] A.I. Tsirikos, R. Dhokia, S. Wordie, Rotatory Dislocation of the Spine in Dystrophic Kyphoscoliosis Secondary to Neurofibromatosis Type 1, *J Cent Nerv Syst Dis*. 10 (2018), 117957351881948.
- [7] B.J. Park, S.J. Hyun, S.H. Wui, J.M. Jung, K.J. Kim, T.A. Jahng, Surgical outcomes and complications following all posterior approach for spinal deformity associated with neurofibromatosis type-1, *J Korean Neurosurg Soc*. 63 (6) (2020) 738–746.
- [8] K. Singh, D. Samartzis, H.S. An, 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 (4) (2005) 461–466.
- [9] S.N. Neifert, H.A. Khan, D.B. Kurland, N.C. Kim, K. Yohay, D. Segal, A. Samdani, S. Hwang, D. Lau, Management and surgical outcomes of dystrophic scoliosis in neurofibromatosis type 1: A systematic review, *Neurosurg Focus*. 52 (5) (2022) 1–21.
- [10] H. Koller, J. Zenner, V. Gajic, O. Meier, L. Ferraris, W. Hitzl, The impact of halo-gravity traction on curve rigidity and pulmonary function in the treatment of severe and rigid scoliosis and kyphoscoliosis: A clinical study and narrative review of the literature, *Eur Spine J*. 21 (3) (2012 Mar) 514–529, doi [10.1007/s00586-011-2046-5](https://doi.org/10.1007/s00586-011-2046-5). Epub 2011 Nov 1. PMID: 22042044; PMCID: PMC3296862.
- [11] Kato S, Zeller RD, Magana SP, Ganau M, Oshima Y, Tanaka S, Lewis SJ. Postoperative distal coronal decompensation after fusion to L3 for Adolescent Idiopathic Scoliosis is affected by Sagittal Pelvic Parameters. *Spine (Phila Pa 1976)*. 2020 Nov 1;45(21):E1416-E1420. doi [10.1097/BRS.0000000000003616](https://doi.org/10.1097/BRS.0000000000003616). PMID: 32694489.
- [12] P.T. Calvert, M.A. Edgar, P.J. Webb, Scoliosis in neurofibromatosis. The natural history with and without operation, *J Bone Jt Surg - Ser B*. 71 (2) (1989) 246–251.
- [13] S.H. Kang, Y.C. Kim, Y.J. Sur, Reverse turnover pedicled latissimus dorsi muscle flap for lower back reconstruction Two case reports, *Medicine (Baltimore)*. 97 (51) (2018 Dec) e13851, <https://doi.org/10.1097/MD.00000000000013851>. PMID: 30572555.