

Management of neurofibromatosis spinal deformity, a case report and review of the literature

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ABSTRACT

Patients with NF-1 present musculoskeletal abnormalities with scoliosis being the most common the management of those disorders are demanding. In the present case report it is presented the operative technique that applied for the correction of the scoliotic deformity of a 12 year old patient suffering from NF1.

KEY WORDS: Neurofibromatosis; Spinal Deformity – operative treatment

Introduction

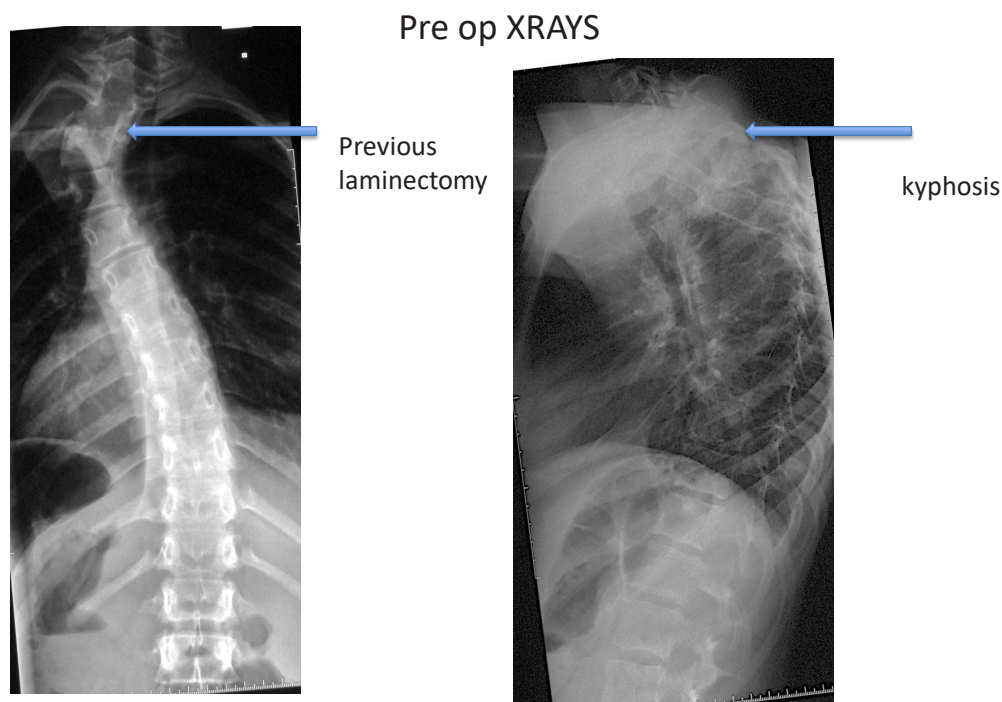
Neurofibromatosis consists a multisystemic, autosomal dominant genetic disorder defined as a spectrum of multifaceted diseases involving neuroectoderm, mesoderm, and endoderm. The German pathologist Virchow was the first one who introduced the clinical features of the disease in several family members in 1847 [1]. However, 35 years later von Recklinghausen, who was Virchow's student, described the histological characteristics of neurofibromatosis [2].

There are five types of neurofibromatosis that can be presented. These types are neurofibromatosis type 1(NF-1), neurofibromatosis type 2 (NF-2), segmental neurofibromatosis, Legius syndrome

and schwannomatosis [3]. The NF-1 is the most common one affecting approximately over two million people around the world. It is the most likely form to be presented with orthopaedic manifestations. The diagnosis of NF-1 is based on the clinical signs of the patient. These include (1) six or more café-au-lait macules more than 5mm in greatest diameter in prepubertal individuals and more than 15mm in postpubertal individuals, (2) two or more neurofibromas of any type or more than one plexiform neurofibroma, (3) freckling in the axillary or inguinal regions, (4) two or more Lisch nodules, (5) optic glioma, (6) a distinctive osseous lesion and (7) a first degree relative with NF-1. The diagnosis is established when at least two of these

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criteria are fulfilled. NF-2 despite the fact that is not associated with primary skeletal disorders, it can be presented with multiple paraspinal and intraspinal tumors. Segmental neurofibromatosis is similar to NF-1 but it involves a single body segment. Patients with Legius syndrome have mild symptoms of NF-1 and schwannomatosis, consists a separate form of neurofibromatosis with multiple schwannomas all over the body.

The epidemiology of spinal deformities in patients with NF-1 varies from 2% to 36% [4,5] and they consist the most frequently presented orthopaedic manifestations in these patients. The characteristic deformity tends to be a short-segmented, sharply angulated curvature that usually involves four to six vertebrae in the upper third of the thoracic spine [6]. The deformities are classified as dystrophic and non-dystrophic according to the coronal plane radiographs. The categorization is based on the coronal plane radiographs and there are certain radiographic criteria for this separation. In total there are nine criteria and if 3 of them are present then the deformity is characterized as dystrophic, otherwise it is non-dystrophic. The

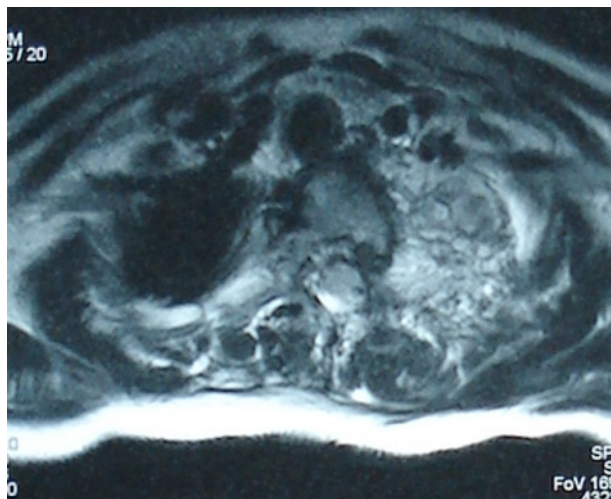
non-dystrophic curves have many similarities with the idiopathic scoliosis [7].

Here we present an interesting case report of a 12 year old patient suffering from NF-1 and was operated with a posterior fusion due to spinal deformation.

Case Presentation

A 12 year old female patients with NF 1 presented in the outpatient clinic with spinal deformation. More explicitly the patient suffered from thoracic kyphoscoliosis. The patient was previously operated with laminectomy at T3-T4 level for a plexiform neurofibroma removal. She had been operated twice for plexiform neurofibroma removal. She had a positive history from mother side and she also had cutaneous neurofibromas and typical café au lait spots.

When presented the radiographic examination revealed a kyphoscoliosis deformation of 58 degrees and high grade spondylolisthesis T3-T4. There were not/were neurological defects of the patient. In addition to the plain x-rays a 3 dimensional computed tomography (CT) examination



MRI shows the plexiform neurofibroma

was performed for better assessment of the deformity. After reviewing the x-ray and the CT operation was decided. A posterior spinal fusion from C5 to T10 was performed lateral mass screw was used in the cervical spine, Magerl' technique implemented. In the thoracic spine pedicle screws was used. In order to achieve adequate fixation and to avoid the anterior support a transvertebrae screw at the level of T3 -T4 was implanted. . The entry point of the transvertebrae screw was the usual entry point of the pedicle screw of the T4, under fluoroscopy the screw targets the body of the listhetic vertebrae. By doing this approach we destroyed the end plate in order to achieve interbody fusion. Cancellous bone allograft was used the bleeding was controlled with the use of Flo Seal.

The patient had an uneventful postoperative period and she was able to follow daily school activities after the first month.

Three years postoperatively, the patient is functional with a very good alignment with good fixation without signs of pseudarthrosis and implants failure.

Discussion

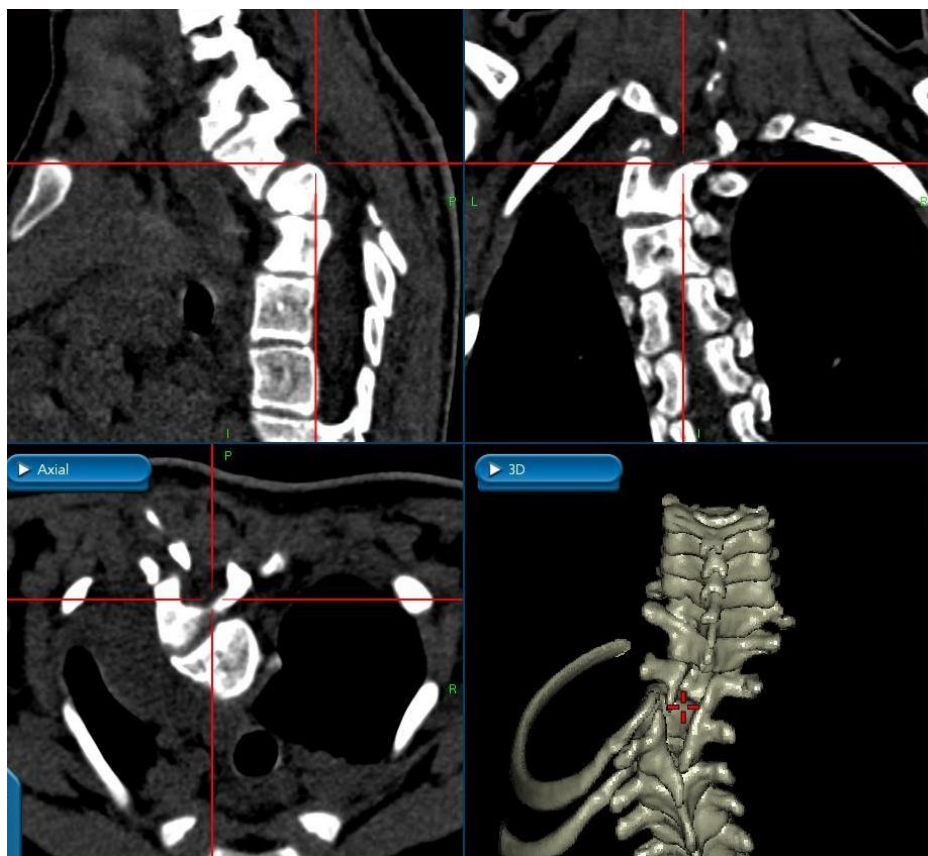
Patients with NF-1 present musculoskeletal abnormalities with scoliosis being the most common. The management of these disorders in young pa-



CT shows the deformity

tients are demanding and require experienced surgeons. Apart from the scoliosis there are also other deformities of the spine associated with NF-1 such as kyphosis, lordoscoliosis , kyphoscoliosis and spondylolisthesis.

In the present case report the 12 year old patient presented with kyphoscoliosis deformation. The definition of kyphoscoliosis is a kyphosis deformity more than 50o which accompanies the scoliotic curve. The deformation of kyphoscoliosis may present in early stages of the disease. In the present case the patient had undergone a previous operation for a neurofibroma removal. Moreover, severe kyphotic deformity is the most common cause of neurological defects even with paraplegia. An explanation for this complication is the elongation of the spinal cord and the deformation after increased



Multiplanar CT reconstruction shows the deformity

spinal flexion such as in kyphoscoliosis [8]. When the angle of the curve surpasses the 50°, the anterior approach for release and fusion is recommended followed by posterior segmental instrumentation one or two levels above and below the end vertebrae [9,10]. In the previously described case a posterior fusion was performed without anterior stabilization. The use of a transvertebrae screw aimed to provide good fixation without anterior support. In such patients even with a combined approach the bony fusion is not always achieved. That is a fact that makes our case even more interesting. Because with the use of posterior approach only, we achieved both adequate fixation and bony fusion as well.

On the other hand, lordoscoliosis is rarer compared to kyphoscoliosis. In such cases it is also recommended anterior release and intervertebral fusion along with posterior instrumented fusion.

Spondylolisthesis is also very rare in NF-1 patients. The definition of spondylolisthesis is the pathologic forward progression of the anterior elements of the spine. In a patient with NF-1 the presence of spondylolisthesis is associated with abnormally thin and long pedicles or pars interarticularis by lumbosacral foraminal neurofibromas or dural ectasia [11].

Apart from the thoracolumbar spine deformities in patients with NF-1, there are also cervical spine deformities as well. Kyphosis is the most common deformity in cervical spine and especially in its progressive form. The posterior cervical spine fusion is recommended in such cases where instability is also present. In cases where flexible deformity is present, the use of halo preoperatively is indicated whereas for stiff cases anterior release and after that the use of halo traction and posterior fusion is a common practice [7].



3 yrs post op

The operations for deformity corrections in a patient with NF-1 do not lack complications. Non-union, vertebral column dislocation, rib protrusion and paraplegia are some of them [7]. During the operation the surgeon should take care of the haemostasis and prevent the formation of haematomas. Additionally, erosion of the laminae secondary to dural ectasia may be noted [12]. In the follow-up period, the deterioration of the curves is not rare in combination with pulmonary symptoms. The infection and thromboembolic are also common complications that the surgeon and the patient should be aware of [7].

Conclusion

In conclusion the management of spinal deformities in patients with NF-1 are challenging and require surgeon's experience and expertise in spine surgery. The principles of the corrections of each curve should be followed. The treating physician should be able to make the separation between the non-dystrophic and the dystrophic curves due to the fact that the latter ones may result in scoliosis, kyphosis and kyphoscoliosis. The postoperative management of the patient is also of high importance as well as a multidisciplinary approach in order to minimize the complications. 

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READY - MADE
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