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GRAND ROUNDS

Intercostal aneurysm causing spinal cord compression in an NF1 patient

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Abstract

Purpose The authors illustrate a case where an intercostal aneurysm was observed in a patient with type 1 neurofibromatosis.
Methods A 32-year-old man with NF1 presented with thoracic back pain. The patient's symptoms progressed to include myelopathic symptoms, including difficulty urinating, numbness in the lower extremities, and increased weakness. Imaging revealed what appeared to be a neurofibroma at the T4–T5 level and a plan to resect the mass was formulated. Upon initial limited hemilaminotomy, significant arterial blood was encountered. The patient was then taken to the interventional suite and angiography was performed, revealing a left T4 intercostal aneurysm. The aneurysm was coil-embolized with no residual filling.
Results By 6 months post-surgery, the patient had regained full strength and sensation in his lower extremities and no longer had difficulty urinating. There has been no recurrence of symptoms 3 years postoperatively.
Conclusions Intercostal artery lesions must be considered as a possible diagnosis in NF1.

Keywords Neurofibromatosis · NF1 · Intercostal artery aneurysm

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Introduction

Case presentation

A 32-year-old male with neurofibromatosis 1 (NF1) and thoracolumbar scoliosis first presented with thoracic back pain. The patient described the pain as constant and rated it as a 10 on a scale of 1–10. The pain worsened when the patient was supine and with Valsalva maneuvers, but was alleviated upon movement and standing upright. The pain wrapped around his chest wall to the anterior left side, suggesting a radicular distribution. He denied any bowel or bladder incontinence, weakness, tingling, or numbness in his legs. On examination, diffuse neurofibromas and stigmata of neurofibromatosis were observed on the patient's skin. Furthermore, the patient's thoracic region had severe coronal deformity with a large hump over the right shoulder blade. Despite this, the patient still had good clinical sagittal balance and no difficulty standing upright. The neurological exam revealed 5/5 strength in all muscle groups of the upper and lower extremities, with no clonus. Sensation in

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the patient's upper and lower extremities was intact to light touch. The patient did have hyperreflexia in his lower extremities (3+) as compared to his upper extremities (2+). No Hoffman sign was observed. Over the course of several months he developed myelopathic symptoms including difficulty urinating, numbness in his lower extremities, and increased weakness (4/5) in his right lower extremity as compared to the left.

Diagnostic imaging

Sagittal MR imaging revealed a focal soft tissue density at the T4–T5 level with significant compression of the spinal cord. CT myelography demonstrated myelographic block at this level (Fig. 1). This finding was compatible with an extramedullary neurofibroma or Schwannoma. Imaging by either modality was limited due to the patient's pronounced dextroscoliosis, which was estimated to be 120° (Fig. 2). However, after consultation with a senior neuroradiologist, it was felt that this mass represented a neurofibroma with significant spinal cord compression at the T4–T5 level.

Historical review of the condition, epidemiology, diagnosis, pathology, differential diagnosis

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is an autosomal dominant genetic disorder that affects approximately 1 in 2,600 to 1 in 3,000 individuals [1]. Although there have been few studies evaluating the long-term survival rates of patients with NF1, it is been noted that their average lifespan is reduced. The most common causes of death are cancer, myocardial infarction, cerebrovascular accidents and pneumonia [2].

The diagnostic criteria of NF1 as defined by the NIH Consensus Conference (updated in 1997) are based on the presence of specific clinical features [3]. The most common of these clinical features is the presence of "café-au-lait" spots or uniformly hyperpigmented macules which appear during the first year after birth and usually increase in number with age [4, 5]. Another notable feature is the formation of multiple peripheral neurofibromas, which are benign, heterogeneous tumors consisting of a mixture of fibroblasts, mast cells, and Schwann cells [6]. Furthermore, NF1 patients have a 8–13 % lifetime risk of developing

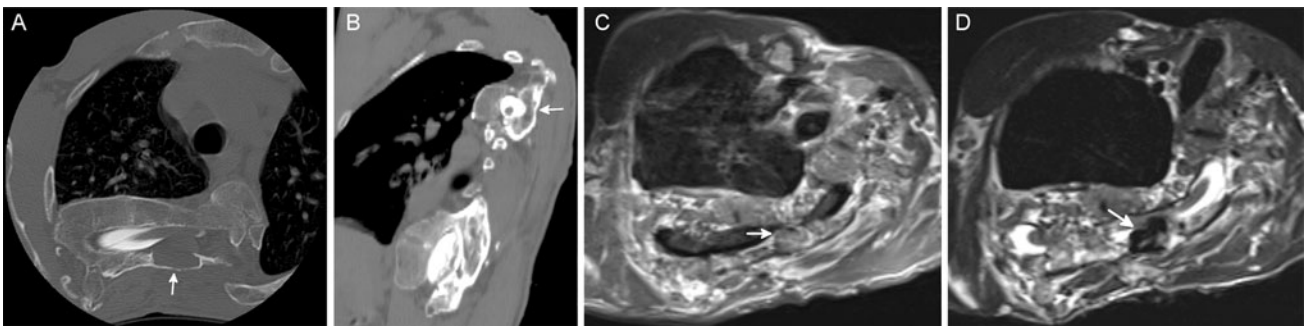

Fig. 1 Pre-operative images depicting the lesion. *Arrows* point to the lesion. **a** Axial CT myelogram, **b** sagittal CT myelogram, **c** T1-weighted axial MRI, **d** T2-weighted axial MRI

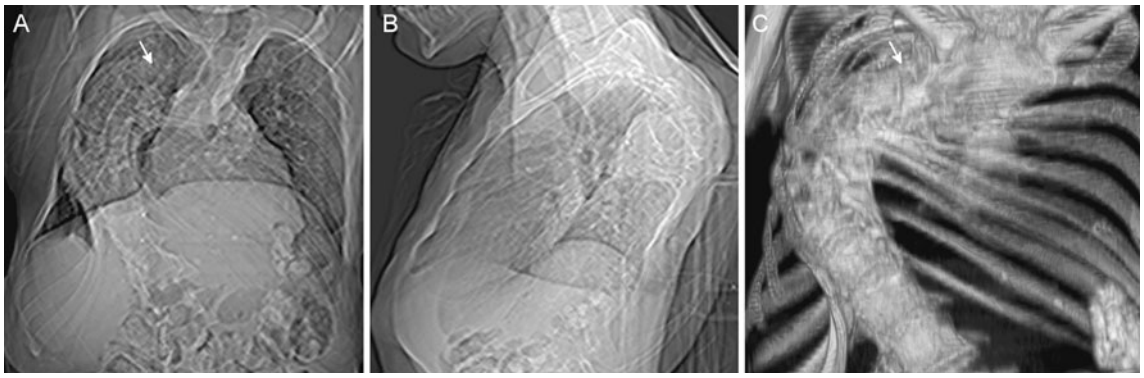

Fig. 2 Pre-operative images depicting the patient's severe scoliosis. *Arrows* point to the location of the lesion, which was located within the horizontal spine segment cranial to the apex of the major curve. **a** AP film, **b** sagittal film, **c** three-dimensional reconstruction of the CT scan depicting major and proximal curves of the scoliosis

malignant peripheral nerve sheath tumors, which often develop from pre-existing neurofibromas [7]. Additional clinical features include the occurrence of optic gliomas, two or more lisch nodules (iris hamartomas), freckling in the axillary regions or having a distinctive bony lesion such as sphenoid dysplasia [3].

Neurological abnormalities are generally the primary focus when treating NF1 patients. Neurological symptoms can range from cognitive deficits, specific learning disabilities, seizures, macrocephaly and peripheral neuropathy [8–11]. Although NF1's effects on the nervous system are generally the principal concern, the vascular system can also be compromised. Adults with NF1 have an increased incidence of aneurysms, arterial stenosis, and hypertension, as determined in postmortem studies, yet clinically appreciable vascular lesions are rarely observed in individuals with NF1 [12]. Unfortunately, these lesions generally only become apparent after rupturing.

Several reports of intercostal artery lesions in NF1 patients have been documented [13]. Aneurysms in most of these cases were discovered only after the onset of severe back pain or chest pain following a spontaneous hemothorax event [13–16]. In rare instances, intercostal artery lesions have resulted in a progressive neurological deficit that preceded discovery of the lesion.

In one previously reported case, an intercostal arteriovenous malformation (AVM) caused congestive myelopathy in a patient with NF1. This patient experienced diminished gait and urinary function, along with decreased bilateral sensation in the lower extremities. These neurological deficits resulted from the intercostal artery directly draining into the medullary vein, which caused venous hypertension of the intradural venous plexus of the spinal cord and subsequent impingement on the spinal cord. The lesion was treated with a hemilaminectomy to directly visualize the lesion, followed by clipping and electrical coagulation of the draining point, thereby preventing blood flow into the medullary vein [17].

Differentiating neurofibromas from vascular lesions can be difficult. For instance, while neurofibromas within the neural foramen can have a characteristic dumbbell shape, an aneurysm in a similar location may mimic the shape of a neurofibroma. Furthermore, both can result in erosion of vertebral bodies or other bony structures. MRI can differentiate between the two lesions. On T1- and T2-weighted images, neurofibromas present as a hyperintense region, while aneurysms/AVMs often have a markedly diminished signal pattern, or flow voids. The void exists because stimulated protons present in a blood vessel are displaced by non-stimulated protons during the course of normal blood flow. This results in a significantly decreased emitted signal [18]. However, vascular lesions may not present with a characteristic flow void. This may be due to the fact that blood flow through the vascular lesion undergoes various disturbances resulting in turbulence instead of laminar unidirectional flow [19]. This would prevent excited protons from escaping the region and would result in a hyperintense region that would mimic a soft tissue lesion. Another possibility is that the vascular structure may have been partially thrombosed. At least one report has demonstrated a mischaracterization of a mass in an NF1 patient [20].

In addition to neurofibromas, malignant peripheral nerve sheath tumors, and vascular lesions, the differential diagnosis of the presented patient must include etiologies related to the unique spinal deformity present in this patient. As mentioned earlier, the patient had a pronounced scoliotic curvature that significantly altered his anatomy. However, spinal cord compression caused by etiologies including a dysplastic rib causing spinal canal impingement, rotary dislocation of the spine at focuses of severe angulation, or stenosis at the apex of the coronal curves or kyphosis was ruled out after careful review of imaging studies by a senior neuroradiologist and a senior orthopedic deformity surgeon.

Rationale for treatment and evidence-based literature

A previous diagnosis of neurofibromatosis can bias a clinician to think of neurofibromas when presented with a lesion of the spine. Given the fact that the patient developed progressive myelopathy, a plan was made to resect the apparent soft tissue mass and decompress the spinal cord through a limited T4-5 laminectomy. The goals of this treatment were to prevent progression of the patient's myelopathy and potentially reverse the deficits that he had developed.

Procedure (surgery/intervention)

After standard prone positioning and dissection of the T4-5 lamina, a neuroforamen-like area was encountered on the left side. This resulted in arterial-like bleeding, indicating the presence of significant vascularity. After controlling the bleeding, additional exposure over the apparent soft tissue mass was obtained. A limited hemilaminotomy was performed with Kerrison rongeurs and the inside of the spinal canal was identified. At this point, a membrane suggesting a vascular abnormality was clearly visualized. After partially decompressing the spinal cord, the vascular abnormality was isolated from the spinal cord with a piece of titanium mesh and the area of significant bleeding was packed with gelfoam and cotton balls. It was difficult to identify the true nature of the lesion, as attempts to characterize it through

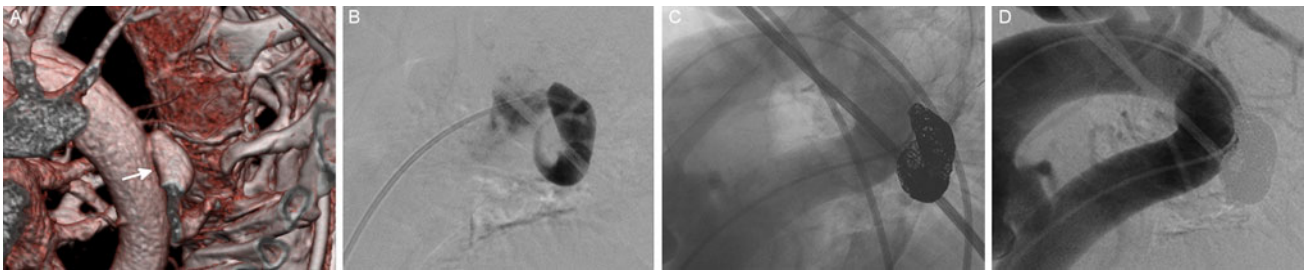

Fig. 3 Angiography performed after initial unroofing of the lamina was attempted. The intercostal aneurysm is seen clearly. **a** 3D CT Angiogram. The *arrow* points to the aneurysm, **b** angiogram of the aneurysm before coiling, **c** angiogram of the aneurysm after coiling, **d** digital subtraction angiogram of the aneurysm after coiling

surgical exploration resulted in torrential bleeding. Only a small hemilaminotomy was performed during initial exposure. Given that minimal exposure was performed and that bilateral facet joints were preserved, it was determined that the spine retained sufficient stability to not require instrumented stabilization. Had there been a need for more extensive exposure to completely resect the mass, we would have considered a short segment fusion.

After controlling the bleeding, a decision was made to evaluate the lesion via angiography, as the significant bleeding prevented adequate surgical treatment. The patient was then transported to the neurointerventional suite, where formal angiography was performed. Angiography revealed an intercostal aneurysm that was subsequently coil-embolized (Fig. 3).

Given the complex vascular anatomy of the spinal cord, making a decision solely based on intraoperative visualization is poorly qualified, since this aneurysm could have arisen from an arteriovenous malformation (AVM) or dural-based arteriovenous fistula (AVF). As such, it is important to understand the relationship of vascular structures in the operative field, when making decisions regarding intraoperative treatment versus endovascular treatment. As this particular aneurysm was fed by a segmental artery, simple trapping of the aneurysm was a less favored option given the possibility of infarcting a portion of the spinal cord at that level. If this lesion were to have arisen from an underlying AVM or AVF, simple trapping of the aneurysm would have been even less favorable as the risk of infracting the spinal cord is even greater. As such, treatment consisted of coil embolization, as opposed to other techniques including clipping and electrical coagulation, to treat the vascular abnormality.

It is interesting to note that there exists a previous case where a patient with NF1 was similarly misdiagnosed with a neurofibroma. After gaining surgical access to the lesion, it became clear that the mass was in fact a lumbar epidural arteriovenous fistula. The fistula was ultimately dissected from adjacent structures and removed in its entirety [19]. As our specific case was less structurally complicated, such measures were not required.

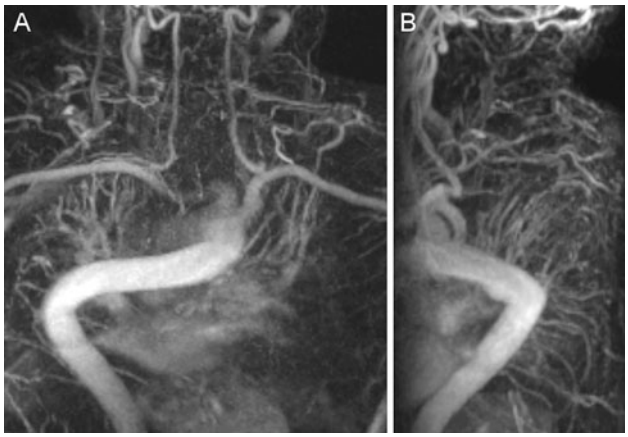

Fig. 4 Post-operative MR angiography demonstrated occlusion of the aneurysm and regular blood flow **a** Coronal view and **b** sagittal view

The relative rarity of vascular lesions in NF1 and lack of clinical findings that indicated vascular involvement resulted in a misdiagnosis in this presented case. The progressive neurological deficit experienced by the patient, in addition to his underlying NF1 and the imaging appearance, seemed to point directly to a peripheral neurofibroma that was impinging on the spinal cord. The presence of a neurofibroma was supported by the MRI, which showed an enhancing soft tissue density. Another complicating aspect was the presence of severe scoliosis, which prevented clear visualization of the lesion.

Outcome, follow-up

Post-operative angiography demonstrated normal laminar flow with complete occlusion of the aneurysm (Fig. 4). The patient's post-operative course is notable for developing a delayed surgical wound infection. Two and a half months after the procedure, the wound was explored and debrided. However, in the weeks following his surgery, the patient's symptoms did resolve. His numbness and urination difficulty resolved, with no reported limb weakness. Successful

symptom resolution indicated that the primary cause of neurologic compromise was not due to the patient's structural spinal abnormalities, which were unchanged after surgery. Given the patient's significant scoliosis, we have routinely followed him to monitor for progression of his symptoms and deformity for 3 years postoperatively.

Although the intercostal aneurysm has not continued to be a problem, the patient went on to develop a distinct malignant peripheral nerve sheath tumor 3 years after the procedure that resulted in sacral region pain, lower extremity radicular pain, and bowel and bladder dysfunction. This lesion has been subsequently resected with lumbosacral-pelvic stabilization.

Conflict of interest Received research support from Eli Lilly.

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