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The Silent Lesion: Spinal Neurofibroma without External Stigmata: A Case Report

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Abstract

A hereditary condition called neurofibromatosis causes many nerve sheath tumors to develop at various body locations. Patients differ greatly in how it manifests clinically. A positive family history or visible skin lesions make it easy to diagnose many instances. However, some, like the current one, develop as a random mutation and manifest in an uncommon manner, with deep lesions and no outward sign. Because they can compress nerve roots and cause severe impairment, painful spinal neurofibromas are important from a clinical standpoint. We describe a male patient, age 22, who has no family history of neurofibromatosis. He had been experiencing increasing nocturnal pain in his left lower limb for six months. He used to walk for a while before the minor soreness subsided. It gradually got bad enough that he had to sleep in a sitting position instead of lying down. There was no history of neurological impairment, fever, edema, trauma, or systemic illness. The brain's MRI was normal. Contrast-enhanced MRI of the spine, however, revealed multiple intradural extramedullary enhancing neurogenic lesions, with symptomatic compression at the L1 and L5 levels. The compressive lesions were excised surgically. After surgery, the person had no more pain at all. After being checked on for a few years, he got another neurofibroma affecting the sciatic nerve, which shows that this disease gets worse over time. This case highlights the importance of understanding that neurofibromatosis can be non-discriminatory in nature (i.e., not all cases will present swelling on the outside) and that progressive leg pain occurring in a younger patient at night should not be dismissed as typical musculoskeletal pain until ruled out as well as indicating an underlying deep intervertebral spinal nerve root compression. An early MRI evaluation with prompt surgical intervention for symptomatic lesions and appropriate long-term post-operative follow-up are important components of achieving a reasonable outcome for the patient.

Keywords: Neurofibromatosis; spinal neurofibroma; nocturnal pain; nerve root compression; sporadic NF1; sciatic nerve neurofibroma; intradural extramedullary tumour.

INTRODUCTION

Neurofibroma is a benign tumour that arises from the connective tissue elements of peripheral nerves. Histologically, it is made up of a mixture of Schwann cells, perineural-like cells, fibroblasts, mast cells, and collagen stroma [1]. These lesions can present as a single solitary tumour or as multiple tumours associated with neurofibromatosis. The most common type associated with multiple neurofibromas is Neurofibromatosis Type 1 (NF1), also known as von Recklinghausen's disease [1].

NF1 is an autosomal dominant disorder. It is caused by mutations in the NF1 gene located on chromosome 17q11.2. The gene encodes a tumour suppressor protein called neurofibromin, which regulates cell proliferation via the Ras pathway. When neurofibromin function is lost, there is uncontrolled growth of nerve sheath elements and multiple neurofibromas develop over time [1,2]. Roughly half of the cases of NF1 are familial — inherited from a parent. The other nearly half are sporadic, arising from a fresh de novo mutation in the NF1 gene [2]. So a negative family history does not at all rule out NF1.

An expert international group updated the criteria for diagnosing neurofibromatosis 1 (NF1) in 2021 [3]. The updated criteria include pathogenic NF1 gene variants found in previously normal tissue and two or more choroidal lesions visible on optical coherence tomography or near-infrared reflectance imaging to diagnose NF1. The primary goal of these revisions was to shorten the amount of time it takes to make a diagnosis, particularly for atypical disease presentations [3].

Neurofibromas can occur in multiple places, such as on the skin, below the skin, around the nerve roots or the spinal cord. Superficial neurofibromas are often painless, slow-growing, soft masses. Deeper lesions can go unnoticed for years until they grow large enough to create pressure against nearby nerve roots or against the spinal cord [4,5]. Spinal neurofibromas are typically found outside of the spinal cord and may cause symptoms depending on which nerve root(s) are pressed by the tumors [5].

Several cases of classical NF1 cutaneous manifestations with family history have previously been described in the Indian medical literature [1,6,7]. However, few reports describe cases of multifocal spinal neurofibromas without epidermal manifestations or family history. We present an example of this situation, where a young adult presented with persistent and debilitating nocturnal lower limb pain; subsequent imaging revealed multiple spinal neurofibromas. While a surgical intervention may be successful in treating a patient's NF1-related tumor(s), it is essential to continue following-up with patients after surgery because they could develop additional tumors elsewhere.

Case Presentation

A male college student, 22 years old, came to our outpatient department because he had been having pain in his left lower leg for six months. The pain came on slowly at first and got worse over time. At first, the pain was not too bad, and it only hurt him when he slept at night. The person in pain said that the pain would go away on its own after getting out of bed and walking for a while.

Over the next three to four months, things changed. The pain became more frequent. It also became more severe. Sleep was getting disturbed almost every night. Eventually, he reached a stage where he was completely unable to lie in supine or any recumbent position for sleep. He had to sleep in sitting posture to get even partial relief. By the time he reported to us, this had been going on for several weeks.

There was no history of any trauma to the limb. No fever. No visible swelling, redness, ulceration, or discharge anywhere on the body. The pain was not sudden in onset. There was no history of tingling, numbness, weakness, or sensory loss in the limb. No back pain. Bowel and bladder habits were normal. He denied any history of seizures, headache, visual disturbance, or feature suggestive of cranial nerve involvement. There was no history of weight loss, loss of appetite, or any constitutional symptoms.

His past medical history was unremarkable. No similar episode in the past. No previous surgery. He was not on any long-term medication. Neurofibromatosis, café-au-lait spots, skin tumors, or any other genetic disease had not been seen in the family. A parent and an older brother were said to be in good health. The patient did not smoke and did not drink alcohol.

The patient was awake, able to follow directions, and not feverish when a general check was done. Heart rate, blood pressure, and breathing rate were all normal. There was no icterus, pallor, cyanosis, trembling, lymphadenopathy, or swelling in the feet. In good light, the face was carefully checked. There were no café-au-lait macules found. No freckling in the armpits or groin. No neurofibromas on the skin. A look through a slit-lamp did not show any Lisch nodules. There were no other signs of neurodermatitis. A close look at the left lower leg did not reveal any unusual findings.

There was no visible swelling, no deformity, and the overlying skin appeared normal in colour and texture. There was no local rise of temperature, no redness, ulceration, or discharge. No palpable mass could be appreciated on careful palpation along the course of the sciatic nerve or in the gluteal region. Peripheral pulses — femoral, popliteal, dorsalis pedis, and posterior tibial — were well felt on both sides. Motor power was 5/5 in all major muscle groups of the lower limb. Sensory examination including light touch, pinprick, and joint position sense was within normal limits. Deep tendon reflexes (knee jerk and ankle jerk) were normally elicitable. Plantar response was flexor. Straight leg raising test was negative bilaterally at the time of initial presentation.

In view of progressive, nocturnal, and positional limb pain in a young patient — and a normal clinical examination that did not point to an obvious source — an MRI of the spine with contrast was advised to look for any deep-seated structural cause.

MRI of the lumbar spine (with contrast). Imaging revealed multiple small nodular altered signal intensity lesions of varying sizes within the lumbar spinal canal. The lesions were located within the thecal sac, in close relation to the nerve roots. They were hypointense to cerebrospinal fluid on T2-weighted images. On post-contrast images, they showed intense enhancement. The largest of these lesions was at the L5 level and measured roughly 10 × 11 mm. Lesions at the L1 and L5 levels were producing significant mass effect, with compression of the traversing nerve roots — which explained the

patient's left lower limb pain. The L5 vertebra appeared sacralized. Mild disc desiccative changes were seen at multiple levels. The lower dorsal cord and conus medullaris appeared normal.

MRI of the cervico-dorsal spine (with contrast). Whole-spine screening was done given the multifocal nature suspected on lumbar imaging. This revealed multiple small nodular enhancing lesions of varying sizes within the cervical and dorsal spinal canal, again located within the thecal sac in relation to the nerve roots. Lesions were noted at C5–C6 level on the left side, C6–C7 level on the right side, D5–D6 level on the right side, and D10 level on the left side. Similar small enhancing nodular lesions were also seen at the cervicomedullary junction and the C1 level. The vertebral bodies showed normal marrow signal intensity. There was no evidence of discitis, osteomyelitis, or paraspinal collection. No major disc herniation or protrusion was identified. The cervical and upper dorsal cord otherwise appeared normal.

MRI brain (with contrast). Given the multilevel spinal involvement, MRI brain with contrast was also performed — to look for any intracranial lesion that may sometimes occur in NF1 (for example, optic pathway glioma or meningioma). The ventricular system, grey and white matter, brainstem, cerebellar hemispheres, bilateral eighth cranial nerves, and the cerebellopontine angle regions appeared normal. There was no evidence of intracerebral or extracerebral haematoma, no space-occupying lesion, no midline shift, and no abnormal post-contrast enhancement. The MRI brain was reported as essentially normal.

Putting together the clinical picture and the imaging findings, a diagnosis of multiple spinal intradural extramedullary neurogenic tumours with nerve root compression was made. Considering the multiplicity of lesions at different spinal levels in a young patient with no family history, the possibility of sporadic neurofibromatosis-associated multiple spinal neurofibromas was considered the most likely explanation, and clinicopathological correlation was advised.

After multidisciplinary discussion with the neurosurgery team, it was decided to surgically excise only the symptomatic compressive lesions at the L1 and L5 levels. The remaining non-symptomatic spinal lesions were planned for clinical and radiological surveillance, given that they were not causing any compression or symptoms at that time. Surgical excision was carried out under general anaesthesia. The intra-operative and post-operative course was uneventful.

The patient had complete relief of nocturnal limb pain in the immediate post-operative period itself. He was able to lie down and sleep comfortably from the very first post-operative night onwards — something he had not done in months. Histopathological examination of the excised lesions was consistent with neurofibroma. He was discharged in stable condition and was kept under regular follow up.

A few years later, while still on follow up, the patient came back with a slowly growing swelling in the posterior aspect of the left thigh, associated with some radicular pain. Repeat MRI of the lower limb showed a neurofibroma involving the sciatic nerve. The lesion was again managed by surgical excision, and the patient remained well thereafter.

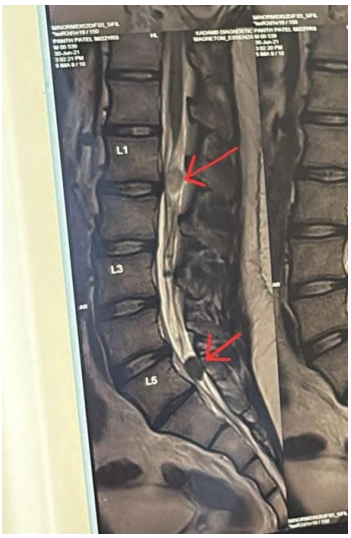

FIG-1 MRI Spinal
Upper arrow shows neurofibroma on L1 level
Lower arrow shows neurofibroma on L5 level

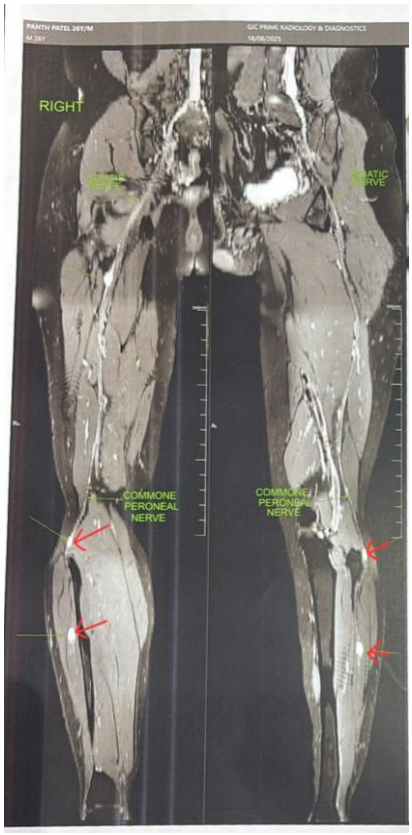

FIG – 4 MR Neurography
Right Upper arrow showing neurofibroma on common peroneal nerve
Right Lower arrow showing neurofibroma on fibular neck nerve
Left Upper arrow showing neurofibroma on common peroneal nerve
Left Lower arrow showing neurofibroma on fibular neck nerve

DISCUSSION

Our case raises a few clinically important points which we discuss below.

The first interesting point is the presentation. NF1 is classically a clinical diagnosis. Most patients are picked up on the basis of café-au-lait macules, freckling in the axillary or inguinal region, cutaneous neurofibromas, Lisch nodules, and so on [3]. In our patient, none of these classical skin findings were present. He came purely with limb pain and nothing else externally. Bagheri et al. [5] reported a 36-year-old with bilateral spinal neurofibromatosis who already had multiple café-au-lait spots, cutaneous neurofibromas, and even a large facial neurofibroma — which made the clinical diagnosis very straightforward in their case. Similarly, Sadeh and Farhat [4] reported a 50-year-old with severe high cervical cord compression from large bilateral neurofibromas, in whom cutaneous features were also present at the time of presentation.

Indian case reports too have largely described patients in whom skin findings or family history pointed early towards the diagnosis. Poswal et al. [1], from SGT Medical College in Gurugram, described a 13-year-old boy with plexiform neurofibroma along with café-au-lait macules and Lisch nodules. Deshpande et al. [6], from Datta Meghe Institute of Medical Sciences, reported a typical case where the multiple nodular cutaneous lesions were the leading clue. Choudhary et al. [7], from AIIMS, recently described a paediatric case with multiple typical features. The novelty in our case is that the patient had no skin lesion, no family history, and no other classical feature at first presentation — the deep spinal lesions were the only manifestation of the disease.

Another important idea is the link between NF1 and family history. People can be diagnosed with NF1 if they have a family history of it. About half of those people will get NF1 from their parents (familial cases), and the other half will have a new mutation that happened on its own (de novo), which happens randomly in a group. A group from PGIMER Chandigarh and SGPGIMS Lucknow, led by Srivastava et al. [2], recently found proof that NF1 cases that happen naturally in the population may be found earlier than NF1 cases that are passed down through families. The patient did not have any family members with NF1 at the time of the initial evaluation, which is one reason why NF1 was not considered as a possible diagnosis. However, it is important to remember that a patient who does not have

any family members with NF1 is still not automatically diagnosed with NF1.

Third, the patient had pain that got worse at night, changed positions, and happened at night. The pain was so bad that the patient couldn't lie down to sleep and had to spend most of the night sitting up. For the young patient, severe limb pain at night that gets worse when sitting down and gets better when walking is a strong sign that an intradural mass is pressing on a nerve root. These people are sometimes told they have sciatica or general joint pain. Gokkus et al. [8] reported a case of a malignant peripheral nerve sheath tumor in the sciatic nerve in a person with NF1 who first came in with sciatica, which was the wrong diagnosis that slowed down diagnosis and treatment. In these situations, waiting too long to diagnose and treat can be very bad if there was malignant change.

The fourth point relates to the role of imaging. MRI with contrast is the investigation of choice for spinal neurofibroma [4,5]. It defines the lesion well, characterizes the signal pattern, shows the typical post-contrast enhancement, and demonstrates the relation of the lesion to the spinal cord and nerve roots. In our patient, MRI was the single most useful investigation that nailed the diagnosis. Whole spine imaging — not just imaging limited to the symptomatic region — is critical in such cases because of the multifocal nature of the disease. Had we limited the MRI to only the lumbar spine on the basis of leg pain, the cervical and dorsal lesions would have gone unnoticed and the diagnosis would have remained incomplete.

The fifth point is the surgical strategy. Surgical excision is indicated in spinal neurofibromas when there is evidence of nerve compression and associated clinical symptoms [4]. Not every spinal lesion needs to be excised in a patient with multifocal disease. Careful tracking is needed for parts of a disease that don't show any symptoms like this. The L1 and L5 tumors in our patient were the only ones that showed compression on imaging and were causing symptoms, so we decided to only remove them. The smaller tumors that didn't cause any symptoms were left alone because they would need to be looked into more further later. The patient's pain went away completely and right away after the lesion was removed, which shows that the targeting approach worked.

The last and sixth thing we want to discuss is that people who have NF1 may develop new neurofibromas in other places over time. A person can get a new symptomatic neurofibroma in another part of their body even after having their symptomatic tumors safely removed [1]. This patient developed a neurofibroma on the sciatic nerve about two years after the first surgery on the spine. A case report of a child with an isolated giant neurofibroma of the sciatic nerve was recently published by Sultana and Sailaja at AIIMS Bibinagar. Neurofibromas affecting the sciatic nerve have only been seen in a few cases, as this case shows. Thawabtah et al. [9] also reported a young adult who had a giant neurofibroma on the sciatic nerve that was isolated. When you put these reports together with our own, they show that NF1 gets worse over time and stays with a person for their whole life. Surgery rarely stops the disease from progressing. [10]

Long-term clinical and radiological follow up is essential. Malignant transformation into a malignant peripheral nerve sheath tumour (MPNST), though rare, should always be kept in mind, especially if a previously stable lesion starts becoming painful, grows rapidly, or develops a neurological deficit [8].

A few limitations of our report must be acknowledged. Genetic testing for the NF1 mutation was not performed in our patient. Confirmation at the molecular level would have added strength to the diagnosis, though it was not strictly necessary given the imaging and clinical picture. Long-term surveillance imaging at fixed intervals was also not done; the second tumour was diagnosed only when the patient developed new symptoms. In future, we plan to follow such patients with periodic clinical review and imaging, even in the absence of fresh symptoms.

CONCLUSION

Neurofibromatosis can occur sporadically and may present without any visible skin lesion or family history. Progressive nocturnal pain in the lower limb of a young patient — particularly if it worsens on lying down and improves on walking - should make the clinician think of a deep-seated spinal pathology, even when the routine clinical examination is normal.

MRI with contrast, covering the entire spine, is the key investigation in such cases. Surgical excision of symptomatic compressive lesions gives excellent and immediate pain relief. But the story does not end with one surgery. New lesions can develop at other sites in the same patient at any point of time, as happened in our case with the later development of a sciatic nerve neurofibroma. Long-term follow up is therefore not optional.

Awareness of such atypical presentations of neurofibromatosis among general practitioners, orthopaedic surgeons, and primary care physicians is important - so that nocturnal limb pain in a young patient is taken seriously and not dismissed as a nonspecific musculoskeletal symptom.

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