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Cervical Vertebral Arteriovenous Fistula Presenting as Progressive Spastic Quadriparesis in a Young Adult with Neurofibromatosis Type 1

Kyaw Zaw Lynn[1*], Myint Thein Naing[2] Aung Moe Myint[1], Kyaw Zaya[3], Ohnmar Saw[4] & Nwe Nwe Win[2]

[1] No. (2) Military Hospital (700-Bedded), Aung Ban, Shan State, Myanmar
[2] Defence Services Medical Academy, Migalardon, Myanmar
[3] No. (2) Defence Services General Hospital (1000-Bedded), NayPyitaw, Myanmar
[4] No. (2) Military Hospital (500-Bedded), Yangon, Myanmar

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***Corresponding author:** Kyaw Zaw Lynn
No. (2) Military Hospital (700-Bedded), Aung Ban, Shan State, Myanmar

Abstract

Neurofibromatosis type 1 (NF1) is a common autosomal dominant neurocutaneous disorder with multisystem involvement. Although cutaneous and neural manifestations are frequent, vascular complications are relatively rare and often under-recognized. Spinal arteriovenous fistulas (AVFs) are an uncommon but important vascular manifestation of NF1 and may lead to progressive myelopathy due to venous hypertension and cord compression. We report a case of a 30-year-old male soldier presenting with gradually progressive spastic quadriparesis and sensory impairment caused by a cervical vertebral arteriovenous fistula associated with NF1. Successful endovascular embolization resulted in complete obliteration of the fistula and neurological stabilization. This case highlights the importance of early suspicion, detailed neuroimaging, and timely intervention in potentially reversible causes of myelopathy.

Keywords: *Neurofibromatosis type 1, Cervical vertebral arteriovenous fistula, Spinal vascular malformation, Spastic quadriparesis, Endovascular embolization*

Introduction

Neurofibromatosis type 1 (NF1) is one of the most common neurocutaneous disorders, with a worldwide prevalence of approximately 1 in 3,000 individuals. It is caused by mutations in the NF1 gene on chromosome 17, resulting in loss of neurofibromin, a tumor suppressor protein that regulates the Ras/MAPK signaling pathway.

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While dermatological and neurological manifestations are well recognized, vascular involvement in NF1 remains relatively uncommon but clinically significant. Reported vascular abnormalities include arterial stenosis, aneurysms, arteriovenous malformations, and arteriovenous fistulas. Cervical vertebral AVFs are particularly rare and may present with progressive myelopathy due to spinal cord compression and venous congestion. Early diagnosis is crucial, as delayed recognition can result in irreversible neurological deficits.

Case Presentation

Patient Information

A 30-year-old male soldier, chronic smoker and alcoholic, previously healthy, unmarried, and residing in Nan San, Myanmar, presented with progressive weakness of all four limbs for two months.

History of Presenting Illness

The weakness had an insidious onset and was gradually progressive. Initially, the patient was able to walk with support, followed by difficulty standing, and eventually became bed-bound. The weakness was symmetrical and involved both upper and lower limbs.

Associated symptoms included:

- **Sensory impairment below the T4 dermatome**
- **Sphincter disturbances**
- **Persistent neck pain**

There was no history of trauma to the head, neck, or spine. The patient denied fever, respiratory or gastrointestinal infection, recent vaccination, or swallowing difficulty. There was no family history of similar neurological illness.

Clinical Examination

General and Systemic Examination

- Conscious and oriented; GCS 15/15
- Afebrile, no pallor or jaundice
- Heart rate: 68/min, regular
- Blood pressure: 130/90 mmHg
- Respiratory rate: 20/min
- Oxygen saturation: 96% on room air
- Cardiovascular, respiratory, and abdominal examinations were unremarkable

Neurological Examination

- Higher cortical functions: Intact
- Cranial nerves: Normal
- Fundoscopy: Normal

Motor System

- No muscle wasting, atrophy, or fasciculations
- Muscle power (MRC grading):
  - Upper limbs: 2/5
  - Lower limbs: 2/5
- Tone: Increased in all four limbs
- Deep tendon reflexes: Exaggerated
- Hoffman's sign: Positive
- Bilateral ankle clonus: Present

Sensory System

- Impaired sensation below the T4 level

Gait

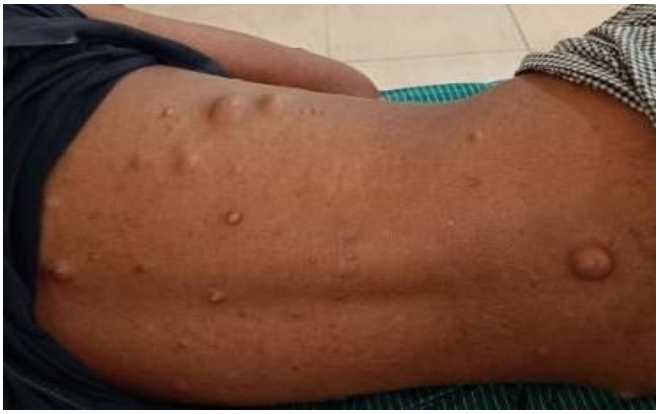
- Unable to stand without support

Cutaneous Findings

Examination revealed multiple cutaneous features consistent with Neurofibromatosis Type 1:

- More than six café-au-lait macules (>5 mm)

Multiple well-circumscribed cutaneous neurofibromas, soft to firm in consistency, showing button-hole invagination



- **Figature1**: Multiple Neurofibromas.
- Axillary freckling

Based on the **NIH diagnostic criteria**, a clinical diagnosis of NF1 was made without genetic testing.

Differential Diagnosis

- Cervical compressive myelopathy
- Spinal arteriovenous malformation / fistula
- Intradural extramedullary tumor (meningioma, neurofibroma)
- Prolapsed intervertebral disc
- Syringomyelia
- Demyelinating disease

Investigations

Laboratory Studies

- ESR, CRP, autoimmune screening: Within normal limits
- Viral serology (HBV, HCV, HIV) and VDRL: Non-reactive
- Metabolic profile: Normal

Systemic Screening

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- Chest X-ray: Normal
- Ultrasonography of abdomen: Normal
- Cardiac evaluation: Normal
- Ophthalmological assessment: No Lisch nodules or retinal hamartomas

Neuroimaging Findings
MRI Cervical Spine (08-08-2024)
Revealed a left cervical vertebral arteriovenous fistula with intra- and extraspinal extension, causing significant spinal cord compression at the C4–C5 level with associated flow voids.

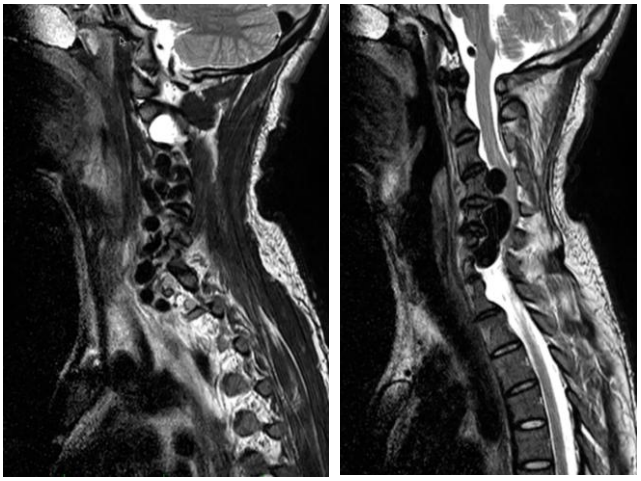

Figure 2: MRI cervical spine demonstrating dilated vascular channels and cord compression at C4–C5.

CECT Head and Neck (26-08-2024)
Confirmed the presence of a **left vertebral AVF** with abnormal arteriovenous communication extending into the spinal canal.

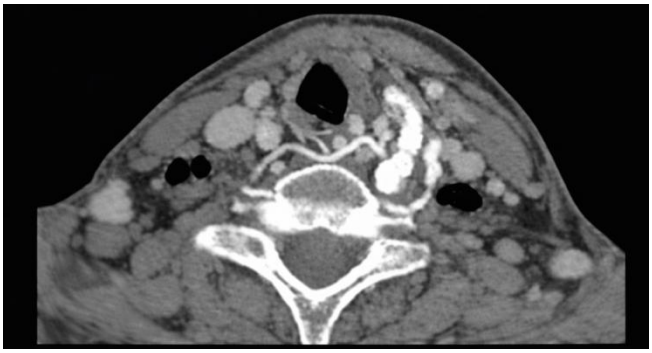

Figure 3: Contrast-enhanced CT showing abnormal enhancement of vertebral artery and venous structures.

Treatment
Endovascular Management
The patient underwent staged **endovascular embolization**:

- **First session (30-08-2024):**
  - Embolization of the AVF using a histoacryl glue–lipiodol mixture
  - Major feeder identified as the deep cervical artery
  - Post-procedure angiogram showed significant reduction in shunt flow
- **Second session (27-09-2024):**
  - Embolization of residual feeders including the left vertebral artery
  - Post-embolization angiogram demonstrated near-complete occlusion

Follow-up Imaging
CECT (18-10-2024):

- Complete obliteration of the left vertebral AVF

Embolized material seen within the vertebral artery, fistula, and vertebral vein

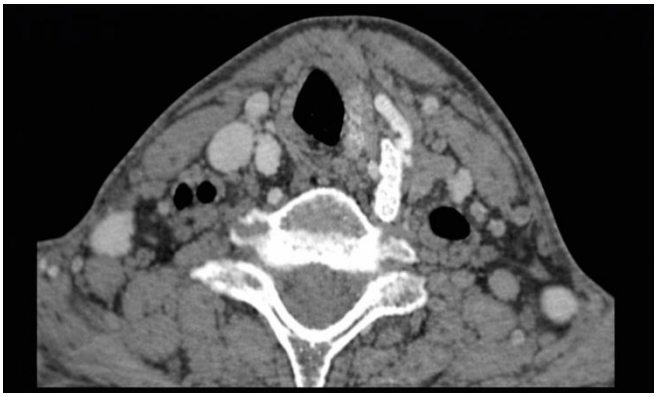

Figure 4: Post-embolization CT showing complete resolution of the AVF.

Discussion

Vascular abnormalities in NF1 result from dysplasia of vascular smooth muscle and endothelial cells due to loss of neurofibromin. Persistent activation of the Ras/MAPK pathway leads to vessel wall fragility and abnormal arteriovenous communications. Spinal AVFs produce neurological deficits through venous hypertension, spinal cord congestion, and ischemia rather than direct arterial insufficiency. The cervical region is particularly vulnerable due to its rich vascular supply and high hemodynamic flow.
Endovascular embolization has emerged as the preferred treatment modality, offering high success rates with low recurrence when complete occlusion is achieved.

Prognosis
Early diagnosis and complete embolization are associated with favorable outcomes. Most patients demonstrate significant neurological improvement, particularly in motor function, while recurrence rates remain low following definitive treatment.

Conclusion

This case underscores the importance of considering spinal vascular lesions in young patients with progressive myelopathy, particularly in the presence of cutaneous features of NF1. Prompt neuroimaging and timely endovascular intervention can result in excellent neurological outcomes and prevent permanent disability.

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