TJVS Turkish Journal of Vascular Surgery

Turk J Vasc Surg. 2026;35(2):175-8
DOI: 10.9739/tjvs.2025.09.046

Available online at www.turkishjournalofvascularsurgery.org

Case Report

Red herring in superior thoracic aperture: spontaneous subclavian artery pseudo-aneurysm in a patient with neurofibromatosis type 1

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Received: September 03, 2025 Accepted: October 02, 2025 Published online: March 05, 2025

Abstract

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder with primarily cutaneous and neurological manifestations. Vascular complications are rare but potentially fatal, often overlooked due to more prominent skin findings. A 41-year-old woman with NF1 presented with left upper limb weakness, neck swelling, ptosis, and hoarseness. Initially presumed to be a neurofibroma, imaging revealed a giant subclavian artery pseudoaneurysm compressing the brachial plexus. She underwent surgical resection via trapdoor thoracotomy. Intraoperatively, subclavian artery avulsion was noted, requiring massive transfusion. Histopathology confirmed pseudoaneurysm. Recovery was uneventful, and long-term follow-up was planned. This case highlights NF1-related subclavian pseudoaneurysm caused by vascular fragility from smooth muscle dysplasia and elastica disruption. Angiography was crucial to distinguish it from neurofibroma by showing contrast extravasation and arterial connections. Surgical resection via trapdoor thoracotomy, complicated by subclavian avulsion and massive transfusion, reflects the high-risk nature of NF1 vascular surgery. Multidisciplinary care and long-term follow-up are essential due to frequent vascular recurrence.

Keywords: Neurofibromatosis Type 1, pseudoaneurysm, subclavian artery, vascular complications

INTRODUCTION

NF1 is a rare autosomal dominant disorder, affecting 1 in 3.000 to 4.000 individuals, caused by pathogenic variants in the NF1 tumour suppressor gene on chromosome 17 [1]. It is primarily characterised by multiple hyper pigmented macules (café-au-lait spots), cutaneous neurofibromas, axillary and inguinal freckling, optic nerve gliomas, and iris hamartomas. Vascular complications, though less common, can have significant consequences, affecting a range of vessels from the proximal aorta to smaller arteries, and presenting as arterial stenosis, aneurysms, or arteriovenous malformations. Aneurysms in NF1 are often asymptomatic, typically presenting only after rupture, which can be life-threatening [2].

CITATION
Danushka N, Acharige AUM, Jayasinghe R, Jeyaratnam Y, Attanayake D. Red herring in superior thoracic aperture: spontaneous subclavian artery pseudo-aneurysm in a patient with neurofibromatosis type 1. Turk J Vasc Surg. 2026;35(2):175-8.

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While the aorta, renal, mesenteric, and cerebral circulations are more commonly involved, subclavian artery involvement is rare, with only a few reported cases of dissection, aneurysm, or rupture [3]. Timely recognition of such vascular complications is crucial, as delays in diagnosis can lead to severe morbidity. Vascular lesions in NF1 are frequently overlooked because the more prominent cutaneous and neurological manifestations often dominate the clinical picture. We present a case of a subclavian artery pseudo-aneurysm, initially mistaken for a neurofibroma, focusing on its presentation and the significant surgical challenges encountered.

CASE REPORT

A 41-year-old female with NF1 and colloid goiter (Thy 2) presented with a 3-month history of progressive left upper limb weakness, pain, numbness, left eyelid drooping, hoarseness, and an enlarging left neck mass. She had no alcohol or smoking history and no other comorbidities. Examination revealed a firm, slightly mobile mass on the left neck. Neurologically, muscle strength was 3/5 at shoulder, 2/5 at elbow, 0/5 at wrist with absent reflexes; sensory loss in C5–T1 dermatomes, worst at C7; muscle wasting in left pectoralis major, deltoid, forearm, intrinsic hand muscles; and medial scapular winging. Left brachial and radial pulses were reduced. Left ptosis was noted without other Horner's signs. Figure 1 shows left upper limb wasting and the neck lump.

Figure 1. Patient image with arrow pointing towards the neck lump (lesion)

Ultrasound confirmed multinodular goiter. Neck and chest MRI showed a 7.5x4.5x5 cm oval mass with cystic and solid parts from C6 to T1, compressing and displacing the left brachial plexus. Differential diagnoses were tuberculoma, neurofibroma, and vascular malformation. Repeat Magnetic Resonance Imaging (MRI) and Computed Tomography (CT) angiography revealed peripheral heterogeneous arterial phase enhancement, a fistulous connection to the mid-left subclavian artery, central blood pooling, and an enlarged tortuous proximal left subclavian artery. The left common carotid was displaced anteriorly; the left subclavian artery was compressed and displaced posteriorly. Contrast extravasation indicated active bleeding. Digital subtraction angiography (DSA) confirmed a pseudoaneurysm from the second part of the left subclavian artery with proximal left vertebral artery compression.

Figure 2 & 3 show the pre-operative CT angiographic images.

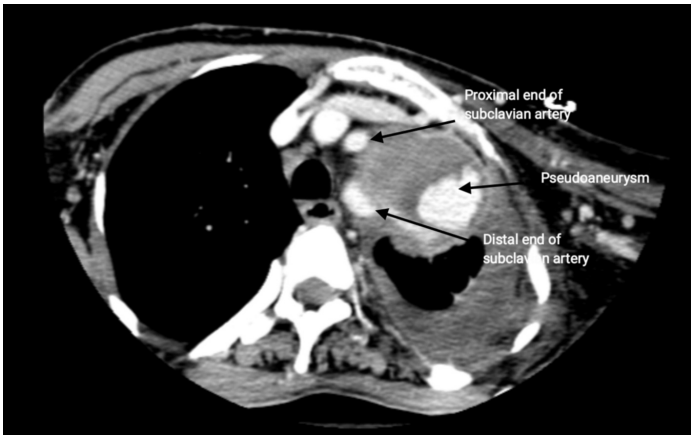

Figure 2. Contrast-enhanced CT axial image showing the pseudoaneurysm proximal and the distal end of the left subclavian artery

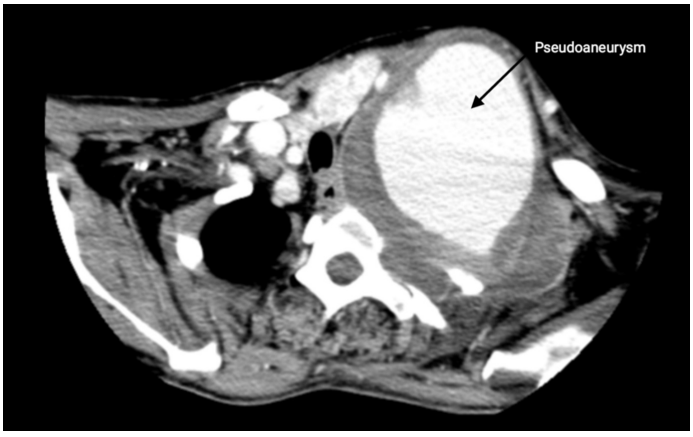

Figure 3. Contrast-enhanced CT axial image demonstrating the pseudoaneurysm

A multidisciplinary team consisting of neurosurgeons, vascular surgeons, cardiothoracic surgeons, and interventional radiologists decided on surgical intervention. The patient underwent a trapdoor thoracotomy. Intraoperatively, a large

heterogeneous mass was identified, originating from the root of the neck and extending into the left thorax. While gaining the proximal control, the left subclavian artery avulsion was noted from its origin. Upon repairing the subclavian root, the mass was totally resected and removed. The procedure was complicated by massive transfusion. Figure 4 shows the intraoperative image of pseudoaneurysm.

The patient stayed in the intensive care unit (ICU) for five days, during which her recovery was uneventful. Histopathological examination of the excised mass revealed no evidence of malignancy, consistent with the contents of a pseudoaneurysm. Long-term follow-up was planned to monitor for recurrence and manage potential complications.

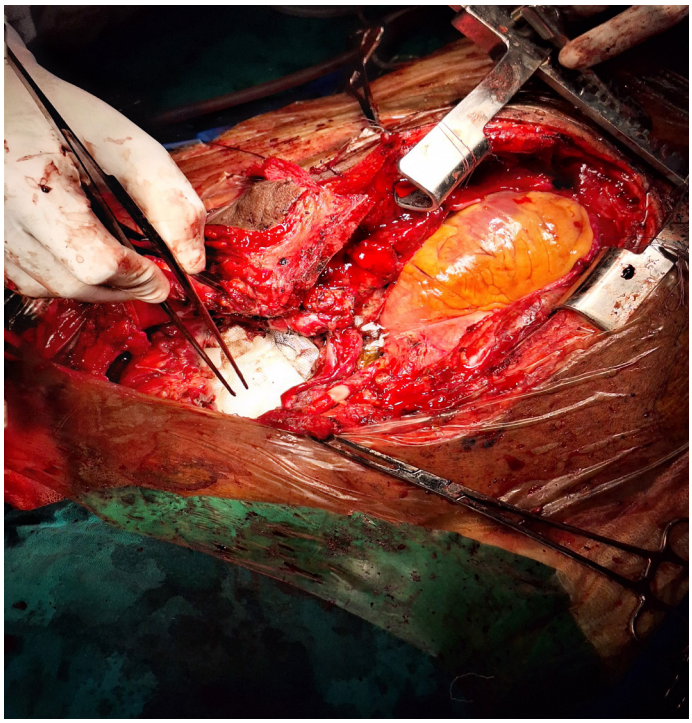

Figure 4. Intraoperative image showing after the thoracotomy

DISCUSSION

NF1 is an autosomal dominant disorder caused by mutations in the NF1 tumor suppressor gene, leading to multisystem involvement including cutaneous neurofibromas, café-au-lait spots, and vascular complications [2,4]. Vascular issues in NF1 include arterial stenosis, aneurysms, pseudoaneurysms, and arteriovenous malformations, often affecting large vessels like the subclavian artery [4,5]. These lesions arise from vessel wall fragility due to smooth muscle dysplasia, tunica elastica disruption, and fibromuscular dysplasia, causing spontaneous arterial wall disruption and pseudoaneurysm formation without trauma [1,4,5].

The patient's symptoms indicated a large subclavian pseudoaneurysm causing compressive and ischemic effects. Progressive left upper limb weakness (C5–T1), sensory loss, and muscle wasting reflected brachial plexus compression [6,7]. Medial scapular winging suggested long thoracic nerve injury. Reduced left brachial and radial pulses and a non-pulsatile neck mass indicated subclavian artery compression from the pseudoaneurysm. Left eyelid ptosis and hoarseness pointed to sympathetic chain and recurrent laryngeal nerve involvement from mass effect [7,8].

The role of imaging is paramount in diagnosing such vascular complications. MRI and CT angiography revealed a large mass with cystic and solid components, compressing and displacing the subclavian artery, and showing evidence of contrast extravasation-hallmarks of a pseudoaneurysm. DSA confirmed the presence of a pseudoaneurysm arising from the second part of the subclavian artery and demonstrated non-visualization of the proximal vertebral artery due to compression. These imaging modalities are essential for distinguishing vascular lesions from infectious or neoplastic processes in NF1 patients and for preoperative planning [2,9].

The diagnostic challenge between neurofibroma and pseudoaneurysm in NF1 arises from their similar clinical and imaging features. Both can appear as soft-tissue masses with cystic and solid parts, compressing or displacing nearby structures on MRI and CT [10,11]. Neurofibromas are common and heterogeneous, while pseudoaneurysms mimic them but show vascular signs like contrast extravasation or arterial connections on angiography [5,10]. CT/MR angiography and DSA are essential to identify pseudoaneurysm-specific features, aiding differentiation from neurofibromas [11].

Treatment of subclavian artery pseudoaneurysm in the context of NF1 presents significant challenges, primarily due to the vascular fragility associated with NF1-related arterial dysplasia. This inherent weakness of the vessel wall greatly increases the risk of intraoperative hemorrhage, as seen in this case [9].

Anatomically, the close proximity of the pseudoaneurysm to critical neurovascular structures, including the brachial plexus, subclavian artery and vein, complicated the surgical dissection and exposure. A trapdoor thoracotomy was necessary to achieve adequate access and control, highlighting the complexity of operative management in such cases [12].

Surgical ligation or repair is often required in the context of active bleeding or rupture, as was the case with our patient [1,2]. Endovascular techniques, including stent placement or coil embolisation, offer minimally invasive alternatives, especially in high-risk surgical candidates [3,8]. Medical management, such as blood pressure control and close imaging surveillance, may be appropriate for small, asymptomatic lesions [3].

Prognosis in NF1 patients with vascular involvement depends on early recognition and timely intervention [4,9]. Regular follow-up with appropriate imaging is essential to detect recurrence or new vascular complications [3,4].

CONCLUSION

NF1 is a complex disorder with multi-system involvement, including rare but serious vascular complications like subclavian artery pseudoaneurysms due to vessel fragility. Early recognition and effective management are vital for improving outcomes. This case highlights the importance of both multidisciplinary surgical collaboration and long-term care, including monitoring, rehabilitation, and coordinated management to address ongoing risks and prevent complications.

Highlight key points

- NF1 can cause rare but serious vascular complications due to vessel wall fragility.
- Subclavian artery pseudoaneurysm in NF1 may mimic neurofibroma clinically and radiologically.
- Angiographic imaging is crucial to identify vascular lesions and differentiate them from tumors.
- Surgical resection is high-risk due to vascular fragility and proximity to critical structures.

Patient Consent for Publication: Informed consent was obtained, and procedures followed institutional ethical standards.

Data Sharing Statement: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Author Contributions: All authors contributed equally to the article.

Conflict of Interest: The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

Funding: The authors received no financial support for the research and/or authorship of this article.

REFERENCES

1. Sakamoto S, Sumida M, Takeshita S, Shibukawa M, Kiura Y, Okazaki T. et al. Ruptured Subclavian Artery Pseudo-Aneurysm Associated With Neurofibromatosis type 1. Acta Neurochir. 2009;151:1163–6.
2. Negreira KE, Lichtenberger JP, Allais B, Alhaddad A, Bernetich M, Jain V. Subclavian artery branch pseudoaneurysm rupture with massive hemothorax in a patient with neurofibromatosis type 1. Chest. 2020;157:e103-5.
3. Oderich GS, Sullivan TM, Bower TC, Miller DV, Babovic-Vuksanovic D, Macedo TA, et al. Vascular abnormalities in patients with neurofibromatosis syndrome type I: Clinical spectrum, management, and results. J Vasc Surg. 2007;46:475-84.e1.
4. Sheerin UM, Holmes P, Group LNR, Childs L, Roy A, Ferner RE. Neurovascular complications in adults with neurofibromatosis type 1: a national referral center experience. Am J Med Genet A. 2022;188:3009-15.
5. Iwama T, Mizutani K, Kubo H, Katsumata M, Akiyama T, Toda M. A case of neurofibromatosis type 1 diagnosed after idiopathic rupture of superficial temporal artery pseudoaneurysm requiring endovascular treatment. NMC Case Rep J. 2023;10:125-30.
6. Alsayed A, Kamal D. Subclavian artery pseudoaneurysm with arteriovenous fistula following trauma: a case report. Bahrain Medical Bulletin. 2024;46:2166-8.
7. Wojtyś ME, Skórka P, Kordykiewicz D, Falkowski A, Jakubowska-Grzeszyk J, Wójcik J, et al. Post-traumatic left subclavian artery pseudoaneurysm secondary to clavicular fracture: a case report and literature review. Biomedicines. 2025;13:187.
8. Francis D, Kumar M, Singh M, Okafor TL, Reddy MMRK, Inban P, et al. Endovascular management of traumatic pseudoaneurysm of left subclavian artery: a case report. Radiol Case Rep. 2023;18:4066-70.
9. Lee J, Kim Y. Life-threatening brachial artery hemorrhage and a lethal outcome in patients with neurofibromatosis type 1: two case reports and a review of the literature. J Int Med Res. 2021;49:03000605211025344.
10. Fortman BJ, Kuszyk BS, Urban BA, Fishman EK. Neurofibromatosis type 1: a diagnostic mimicker at ct. Radiographics. 2001;21:601-12.
11. Kobayashi T, Iinuma S, Hashiguchi H. Radial artery pseudoaneurysm with cutaneous manifestations in a patient with neurofibromatosis type 1. Cureus. 2025;17.
12. Jaldin RG, Bertanha M, Sobreira ML, Braz LG, Freitas CCMd, Yoshida WB, et al. Treatment of an iatrogenic subclavian artery pseudoaneurysm near vertebral artery branch: endovascular approach or open surgery?. J Vasc Bras. 2013;12:237-42.