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CASE REPORT

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Successful management of life-threatening spontaneous inferior thyroid artery rupture in neurofibromatosis type 1: a rare case report

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Abstract

Background Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder associated with cutaneous and vascular complications. Arterial rupture, including the rare involvement of the subclavian artery, can be life-threatening. We present a case of a ruptured inferior thyroid artery in a patient with NF1, presenting with a rapidly expanding neck hematoma.

Case presentation A 35-year-old male with a history of NF1 presented to the emergency department with sudden-onset right-sided neck swelling and pain. The swelling rapidly expanded, leading to severe dyspnea, irritability, and hoarseness, necessitating emergent intubation. After failed attempts of intubation, a surgical tracheostomy was performed. Imaging revealed active arterial extravasation at the inferior thyroid artery near the thyrocervical trunk, with a large neck hematoma extending into the mediastinum. Coil embolization was performed, resulting in a favorable outcome. Postoperative follow-up confirmed successful embolization and resolution of symptoms.

Discussion and conclusion NF1 is commonly associated with cutaneous manifestations but can also lead to vascular complications, including arterial stenosis and aneurysms, due to impaired vascular endothelial and smooth muscle cell function. Rupture of the inferior thyroid artery in NF1 is extremely rare and can present with symptoms such as hoarseness, dysphagia, and swelling, complicating initial diagnosis. Management of vascular complications in NF1 can be challenging due to the fragility of affected vessels. Endovascular interventions, such as coil embolization, offer a less invasive treatment option with promising outcomes. In this case, rapid airway management followed by angiographic embolization led to a successful resolution.

Keywords Airway, Emergency, Spontaneous artery rupture, Neurofibromatosis type 1, Case report

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Background

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen's disease, is an autosomal dominant disorder caused by mutations on chromosome 17 [1]. It is characterized by a wide range of clinical manifestations, including multiple café-au-lait spots, cutaneous neurofibromas, axillary or inguinal freckling, optic gliomas, pigmented iris hamartomas, skeletal abnormalities, Schwann cell tumors, and macrocephaly. Additional complications may include intracranial tumors, neurologic impairments, kyphoscoliosis, syringomyelia, and pheochromocytoma [1].

Arterial involvement in NF1, such as arterial aneurism or rupture, is a rare but life-threatening complication with a variety of clinical presentation, such as chest pain, epigastric pain, back pain, hemoptysis, shortness of breath, dysphagia [2]. However, Expanded Neck hematoma with compressive symptoms is rarely reported as a presentation of subclavian artery rupture [3]. The most common affected arteries include the renal arteries, carotid, vertebral, cerebral vasculature and the aorta. However, Involvement of the subclavian artery is significantly uncommon with only few cases reported in the literature [1–8].

Here, we present a rare case of a ruptured inferior thyroid artery in a patient with NF1 who presented with huge, rabidly expanded neck hematoma. The patient was successfully managed with emergent tracheostomy and coil embolization, highlighting a therapeutic approach to this unusual fatal presentation.

Case presentation

A 35-year-old man presented to the emergency department with sudden-onset swelling on the right side of his neck, accompanied by sudden pain. He reported that the swelling had not been present before and denied any history of trauma, invasive procedures, or other significant precipitating events. His medical history was significant for NF1, diagnosed by skin biopsy 10 years ago, and recurrent deep vein thromposis in the left lower limb, secondary to venous anomaly. The patient also denied experiencing palpitation, nausea, vomiting, headache or neurological symptoms. Additionally, there was no history of hypertension or diabetes.

On examination, the patient's vital signs were within normal limits, including blood pressure of 130/80, Heart rate of 96 beats/minute, and oxygen saturation of 96%. Physical examination revealed right-sided non-tender neck swelling. Skin examination showed multiple small neurofibroma-like masses across the body, especially on the abdomen, along with several café-au-lait lesions. Peripheral pulses were intact. Neurological and cardiovascular examinations were unremarkable. Laboratory findings, including hemoglobin, platelet count, coagulation profile, kidney and liver enzymes, serum electrolytes, inflammatory markers, and atrial blood gases, were within normal limits.

Initially, the neck swelling was small and associated only with localized pain. However, while in the emergency department, the swelling rapidly progressed, extending to the right side of the chest. This was accompanied by worsening pain and dyspnea, prompting an urgent CT angiography of the head and neck. During preparation for the CT scan, the swelling further expanded, leading to irritability, severe dyspnea, hoarseness, and a drop in oxygen saturation. The anesthesia team was urgently called to perform intubation. However, endotracheal intubation failed due to severe tracheal deviation and the massive neck swelling, which obscured visualization of the epiglottis and vocal cords.

After three failed attempts at orotracheal intubation, a surgical airway was considered. However, identifying the cricothyroid membrane was impossible due to the severely swollen neck. An emergency tracheotomy was performed instead. A horizontal incision was made approximately one fingerbreadth above the suprasternal notch. The surrounding tissues were dissected to identify the thyroid gland, and the thyroid isthmus was divided to expose the trachea. The second tracheal ring was incised, and a 6.0 mm tracheostomy tube was placed into the trachea. The patient was subsequently placed on a ventilator with low settings.

After securing the airway and stabilize the patient, CT angiography performed, regealing active arterial extravasation at the anatomical site of the thyroidal arteries near the thyrocervical trunk, with pooling of contrast during the venous phase. A large right-sided neck hematoma extending into the mediastinum was noted, resulting in significant tracheal deviation and compression (Fig. 1).

Given the patient's clinical condition and imaging findings, he underwent an urgent embolization. Through left femoral arterial access, digital subtraction angiography identified active extravasation from the right thyrocervical trunk. Coil embolization was performed using six coils to occlude the affected artery and prevent further hemorrhage. Immediate post-procedure imaging confirmed successful hemostasis with no further contrast extravasation. Following the successful embolization, the patient was admitted to the intensive care unit for close monitoring.

Post-operative head and neck CT angiography confirm successful emblization (Fig. 2). A follow-up neck ultrasound showed a large, hypoechoic, heterogenous, non-vascularized soft tissue structure with internal thick septations and scattered small cystic components, consistent with an organized hematoma (measuring of $6 \times 2.4 \times 6.2$ cm), which was not suitable for therapeutic drainage and complete evacuation.

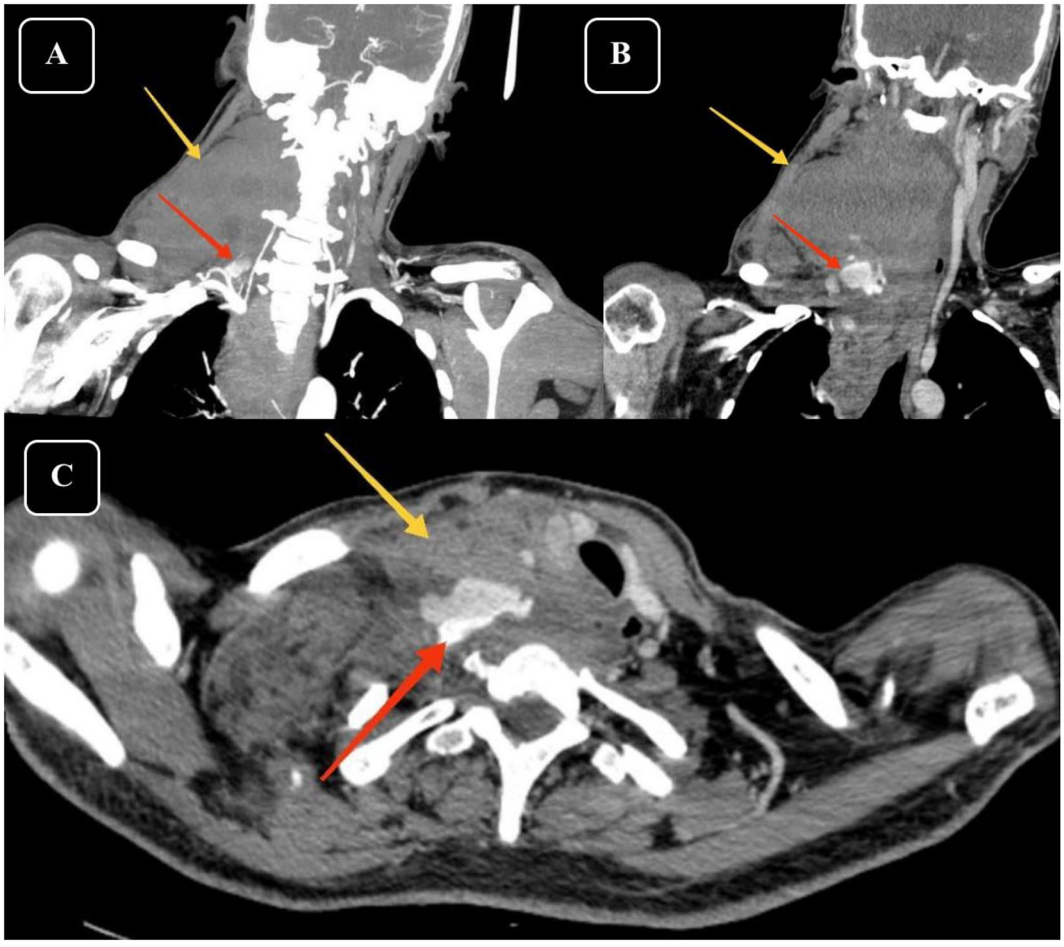

Fig. 1 **A, B**, and **C**) Contrast enhanced CT showing active extravasation of contrast at thyrocervical trunk (red arrow) with adjacent large hematoma (yellow arrow) that is causing tracheal deviation to the left side

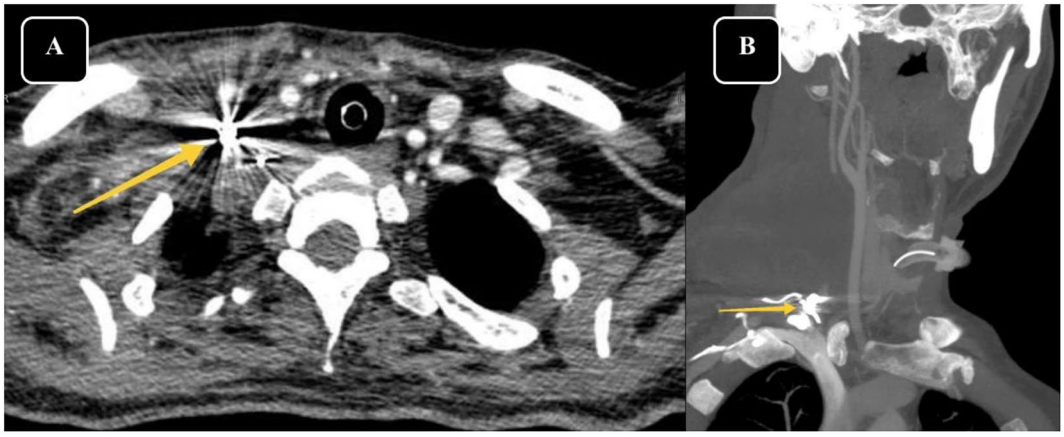

Fig. 2 **A** and **B**) Post-Angioplasy Contrast enhanced CT showing the coil (yellow arrow) within the thyrocervical trunk without adjacent active extravasation

The patient's condition improved, and he reported no pain or dyspnea. He was discharged home on post-operative day 10. At a one-month follow-up, the patient showed excellent clinical outcomes, with no recurrence of neck pain or swelling.

Discussion

NF1, first described by Von Recklinghausen in 1821, is a genetic disorder characterized by cutaneous manifestations such as café-au-lait spots, benign neurofibromas, and iris hamartomas. While these cutaneous features

are well-documented, the vascular complications associated with NF1 are less commonly Among these vascular abnormalities, arterial stenoses are more frequent than aneurysms [1–3, 9]. Renal artery stenosis is the most common vascular lesion in NF1, often presenting as unilateral stenosis and accounting for 41% of cases with vascular involvement. Other vascular abnormalities, such as aneurysms of the vertebral, carotid, and cerebral arteries, are less common and predominantly occur in females during the third decade of life.

The pathophysiology of vascular complications in NF1 is multifactorial, involving processes such as abnormal cellular proliferation, smooth muscle degeneration, impaired healing, and excessive fibrosis [10]. These changes are believed to result from the dysfunction of neurofibromin, a protein encoded by the NF1 gene, which plays a critical role in regulating vascular endothelial and smooth muscle cell functions. The loss of neurofibromin disrupts normal cellular signaling pathways, contributing to structural and functional abnormalities in the vasculature. The prevalence of vascular abnormalities in NF1 patients ranges from 0.4 to 6.4%, with most affected individuals remaining asymptomatic. These abnormalities often involve multiple vessels and are typically discovered incidentally [10, 11].

Historically, the pathology of vascular lesions in NF1 was first described by Reubi in 1944, who put them into three main categories, including intimal proliferation occluding the vascular lumen, an aneurysm formation characterized by fibrohyaline thickening and muscular fragmentation, and a nodular changeing involving spindle or epithelioid cells disrupting the vessel wall. Subsequent studies by Salyer and Salyer proposed that Schwann cell proliferation may initiate secondary degenerative changes in affected vessels [12]. Greene et al. later divided vascular lesions into two categories, which include larger vessel involvement associated with neurofibromatous or ganglioneuromatous tissue, and small vessel dysplasia [13].

In our case, the patient presented with severe neck pain, accompanied by expanded right-sided neck swelling. The initial differential diagnosis included abscess, hematoma, lymphadenopathy, and angioedema. Given the rapid expansion and associated airway compromise, vascular injury was highly suspected prompting urgent imaging which revealed active arterial extravasation at the anatomical site of the thyroidal arteries near the thyrocervical trunk. Rupture of the ITA in NF1 is exceedingly rare with significant clinical outcomes. Such ruptures can manifest with various symptoms such as hoarseness, dysphagia, and swelling [14]. Which may overlap with differential diagnoses like allergic reactions, strokes, or infections, complicating initial assessment.

One previously reported case described a ruptured internal thoracic artery pseudoaneurysm presenting with hemothorax. Management involved urgent thoracic aortography to localize the lesion, followed by endovascular embolization. However, the fragility of arterial walls in NF1 posed a challenge for vessel reconstruction, making embolization essential to prevent rebleeding [8]. Similarly, another case involved a subclavian artery pseudoaneurysm managed with endovascular stenting combined with coil embolization, avoiding the risks associated with open surgery in NF1 patients. Postoperative imaging confirmed complete aneurysm occlusion and preserved blood flow [2]. Seow et al. presented a case of a 46-year-old female with neurofibromatosis type 1 disease presented with left neck pain, a growing supraclavicular mass, and progressive dyspnea. Imaging revealed a ruptured left subclavian artery aneurysm and massive hemothorax. Despite securing the airway with emergency tracheostomy and draining the blood, the patient died from pulseless electrical activity before surgery [3].

Vascular complications management in NF1 is very challenging due to arterial fragility [4]. In cases of arterial rupture, three primary treatment modalities have been described in the literatures: conservative management, surgical intervention, and endovascular approaches. Conservative treatment may involve blood transfusions, fluid replacement, and thoracic drainage. Surgical management, including thoracotomy, ligation, repair, and grafting has been used in cases of ruptured aneurysms. However, surgical repair is often complex due to the fragility of the vessels in NF1 patients, which may lead to complications like ischemia or gangrene of the upper extremities. So sometimes, grafting to restore distal blood flow is necessary.

Endovascular techniques, including stent placement and coil embolization, have emerged as less invasive alternatives. These methods aim to prevent rebleeding while reestablishing peripheral blood flow. They have been particularly successful in cases of carotid artery pseudoaneurysms, highlighting their potential utility in other vascular lesions associated with NF1 [5]. Open surgical repair is often complicated by the fragility of NF1-associated vascular lesions, leading to difficulty in achieving secure anastomosis and increased risk of intraoperative bleeding. Failed primary suturing attempts can lead to catastrophic hemorrhage. Similarly, in endovascular treatment, arterial fragility may predispose to dissection, aneurysm expansion, or stent-graft failure over time.

There is no universal consensus on routine screening for vascular abnormalities in NF1. However, given the risk of spontaneous rupture, screening with Doppler ultrasound, CT angiography, or MR angiography may be beneficial for high-risk patients, especially those with a history of hypertension or unexplained vascular symptoms. A surveillance protocol similar to that used for

vascular Ehlers-Danlos syndrome may be considered for NF1 patients with prior vascular complications [15].

In our patient, the intervention was challenging because of the need to stabilize the patient because of the presence of a large neck swelling causing airway deviation. We initially attempted direct laryngoscopy, followed by video laryngoscopy and fiber-optic intubation, all of which failed due to severe tracheal deviation and compression. Supraglottic airway devices were considered but deemed ineffective in maintaining a secure airway given the patient's condition. Tracheostomy was performed as a last resort to establish a definitive airway.

Conclusion

This rare presentation highlights the importance of considering NF1-related vascular complications in patients with unexplained vascular symptoms or hemorrhage, especially when systemic or cutaneous signs of NF1 are present. In addition to focus on the significant of rabid management of such life-threatening complications.

Abbreviations

NF 1	Neurofibromatosis type 1
CT	computed tomography

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None.

Author contributions

H.A, Y.A, and M.M have participated in writing the manuscript. H.A, M.A, and M.A have participated in review and editing the manuscript. M.B and M.A have participated in imaging interpretation. H.A and M.B and supervised the conduct of the study. All authors read and approved the final manuscript.

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Data availability

No datasets were generated or analysed during the current study.

Declarations

Ethical approval

Not applicable. As it's a case report, it is exempted from ethical approval by local institution responding on the case.

Consent for publication

Written informed consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Competing interests

The authors declare no competing interests.

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