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Case Report

Unveiling the uncommon: Giant internal jugular vein aneurysm in a neurofibromatosis patient: A case report ☆

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ABSTRACT

Neurofibromatosis Type 1 (NF1) is a rare autosomal dominant disorder that has a wide array of clinical manifestations. NF1 Vasculopathies constitute 0.4% to 6.4% of the findings and they often develop in the arterial circulation while venous involvement is rare. We present a case of a 73-year-old male with NF1 with an incidental finding of right neck swelling for 2 months. Different radiological modalities were performed, identifying the lesion as an internal jugular vein aneurysm. The patient was managed conservatively as he was asymptomatic in relation to the swelling. NF1 venous vasculopathies are rare but they have detrimental consequences such as rupture and severe hemorrhage in view of the fragility of the aneurysmal wall and the infiltration of the neurofibroma into the vessel. Hence, high clinical suspicion and selective imaging and follow-up is advisable for physicians.

Introduction

Neurofibromatosis Type 1 (NF1), alternatively referred to as Recklinghausen disease, is a rare autosomal dominant disorder that is caused by the mutation of the NF-1 gene which is located on the long arm of chromosome 17 (17q11.2) [1–6]. Clinically, the manifestations of this disease can range from multiple benign skin neurofibromas, café au lait skin spots and iris hamartomas to freckling of the axilla or inguinal areas and optic gliomas [1,2,7]. Vasculopathies constitute 0.4% to 6.4% of the findings in NF1 and are considered the second most common cause of mortality, following malignant peripheral nerve sheath tumors [1–4,6]. NF1 vasculopathies often develop in the arterial circulation and predominantly affect the renal arteries, while involvement of the venous system is only found on rare occasions [1,2,4]. Since the first documented case of a venous aneurysm in a NF1 patient in 1996, there has been a handful other similar studies reported in literature, all of which involved the internal jugular vein [1,2,4,5,8].

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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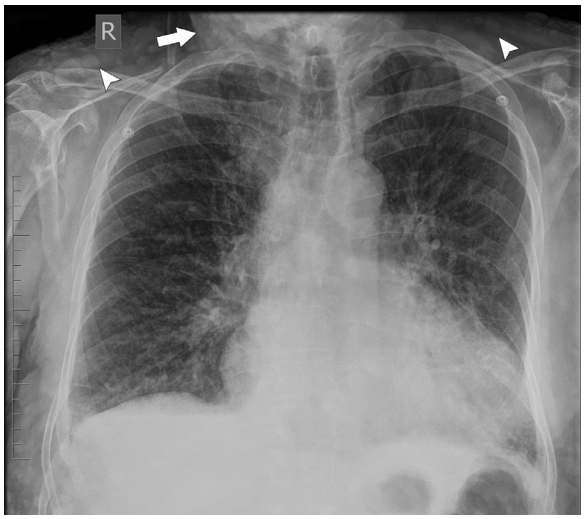

Fig. 1 – Frontal plain chest radiograph shows a faint opacity on the right side of the lower neck (arrow) and multiple rounded skin lesions (arrowheads). It also shows cardiomegaly and prominent pulmonary vasculature.

Case report

This is a case of a 73-year-old male with Neurofibromatosis 1 who presented to the emergency department (ER) with a 2-day history of shortness of breath and nonproductive cough. Following the initial assessment, the patient was found to have tachycardia with a heart rate > 140 beats/minutes, subsequently, an ECG done at the ER confirmed the presence of atrial fibrillation with fast rate and thus a stat dose of 5 mg Verapamil intravenous was given as initial management. The patient was admitted under the cardiology department with an impression of a newly diagnosed atrial fibrillation and managed with rate control medication and anticoagulant agents, furthermore, upon extensive examination it was noted that he had a swelling on the right side of his neck.

The swelling was noticed 2 months prior by the patient, however, it was not associated with pain or symptoms of dysphagia. He also mentioned the slight decrease in its size over the months and that it would disappear upon walking and sitting. Physical examination revealed multiple neurofibromatosis skin lesions all over the face and body and a diffuse swelling in the anterior neck triangle. The swelling was not tender or erythematous and it would disappear when pressed. His vitals were stable post management of the tachycardia, and his blood tests and coagulation profile were all within normal range.

A plain chest radiograph was initially requested as part of the workup. The lungs and heart were normal however a faint opacity on the right side of the neck was seen corresponding to the clinically noted neck swelling, as well as multiple rounded skin lesions, in accordance with the patient's history of neurofibromatosis (Fig. 1).

Ultrasonography of the neck was requested and showed a well-defined, large subcutaneous cystic lesion deep to the right sternocleidomastoid muscle, showing fine mobile internal echoes swirling within the lumen of the lesion indicating slow flowing blood (Fig. 2). Color doppler did not reveal any significant vascularity, most likely owing to the slow flow.

A contrast-enhanced computed tomography scan (CECT) of the neck was later performed and demonstrated a prominent right internal jugular vein with a diameter of about 2.1 cm, and a large focal outpouching along the right lateral wall of the vein measuring about 8.4 × 8.0 × 7.0 cm, showing contrast pooling within the lesion, similar to the contrast in the internal jugular vein. Features indicative of a large saccular right internal jugular vein aneurysm (Figs. 3 and 4). Anteriorly, there was consistent hypodensity along the wall, most likely representing partial thrombosis (Fig. 4). Rest of the main internal jugular vein was patent showing normal enhancement.

The ipsilateral right common carotid, internal and external carotid arteries were separate from the lesion showing no definite communication and appearing displaced medially due

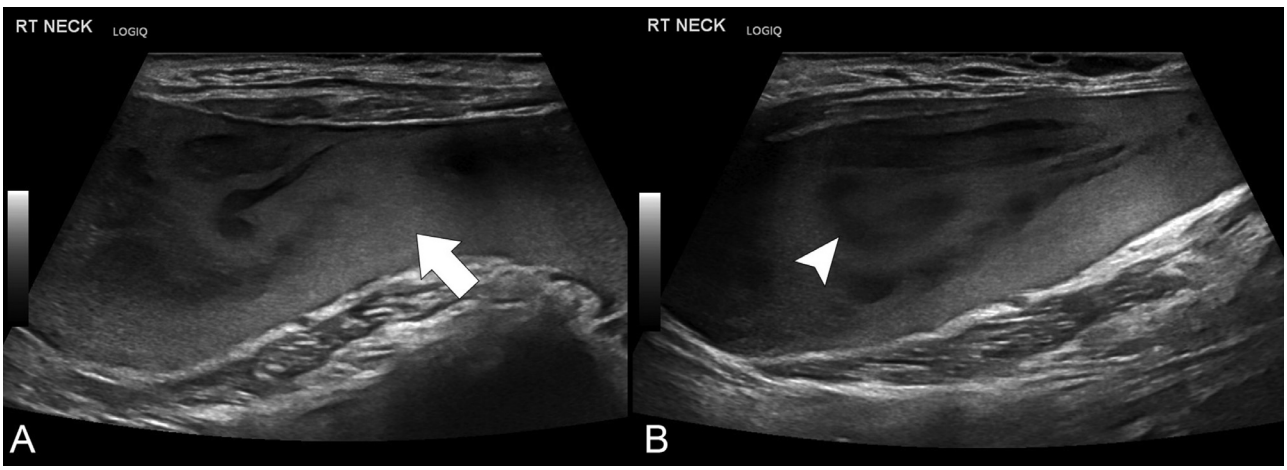

Fig. 2 – Greyscale ultrasound images of the right side of the neck showing (A) well-defined cystic lesion with fine internal echoes (arrow); (B) and curvilinear swirling mobile hypoechogenicities (arrowhead).

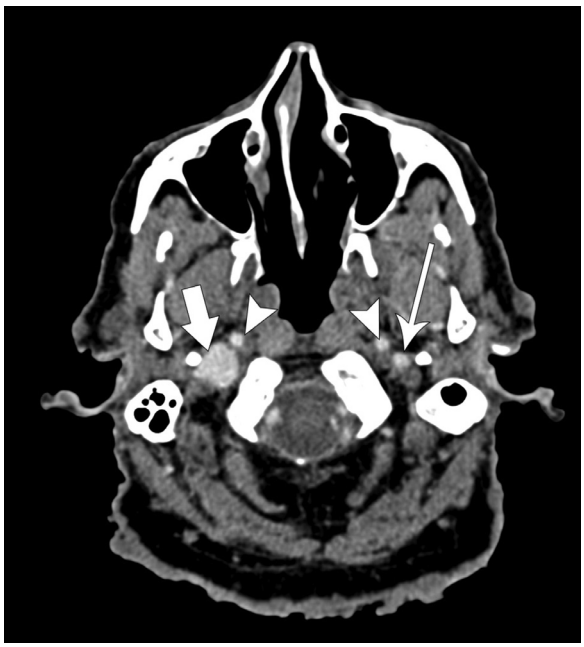

Fig. 3 – Axial contrast enhanced CT neck at the level of the skull base showing ectatic right internal jugular vein (arrow) compared to the normal caliber left internal jugular vein (long arrow). Both internal carotid arteries are also normal in size (arrowheads).

to mass effect from the enlarging lesion. They appeared to be patent. There was slight reduction in the transverse diameter of the oropharynx at the level of the lesion, but the airway remained patent. The left internal jugular vein was normal in caliber (1.1 cm) and the left common carotid and its branches were also normal and patent (Fig. 5). The multiple neurofibromatosis skin lesions were again demonstrated on the CT scan.

The case was referred to the vascular and thoracic surgery department at our facility and considering the asymptomatic state of the patient in relation to the swelling, surgical intervention was deemed non-urgent at the current time. In addition, the patient was tested positive for COVID-19 and thus he was scheduled for follow up on an outpatient bases for further management once he recovered.

Discussion

Vasculopathies are considered some of the rarest manifestation of Neurofibromatosis Type 1, afflicting around 7% of the patients with this disorder [2–4,6,9]. Due to the mutation of the NF-1 gene, production of the neurofibromin protein is altered, this may lead to cellular proliferation or differentiation causing vascular lesions [10]. In literature, neurofibromin expression has been found in vascular, endothelial, and smooth muscle cells histologically [3], this infiltration and localization of the neurofibroma within the vessel walls is speculated to be one of the bases of aneurysm formation and friability [1–5,10]. This mutation also shares partial sequence with the *ras* tumor suppressor oncogene and although it has complete penetrance, its clinical expression remains variable [1–3].

NF1 vasculopathies affect a minor subset of patients and are often asymptomatic [1,10], therefore routine screening is not regularly done and so the true frequency of this complication is ill-defined and most cases are incidental, our case included, however studies in literature have estimated a prevalence of 0.4% to 6.4% [1,3]. The most common vascular lesion in a NF1 setting were found to be arterial aneurysms, stenoses and arterial dysplasia [3]. NF1 vasculopathies frequently tar-

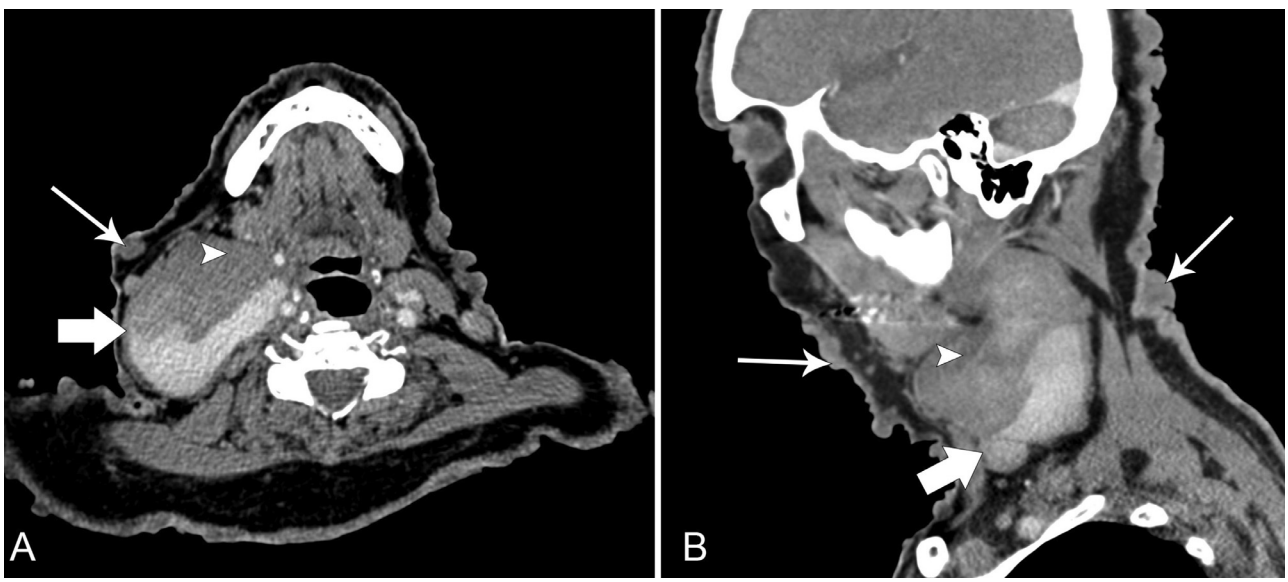

Fig. 4 – Contrast enhanced CT neck, (A) axial view showing a large right neck lesion originating from the right internal jugular vein (arrow) showing contrast layering in the dependent part and partial thrombosis anteriorly (arrowhead). (B) Parasagittal view showing the location of the large aneurysm and the numerous skin lesions (long arrow).

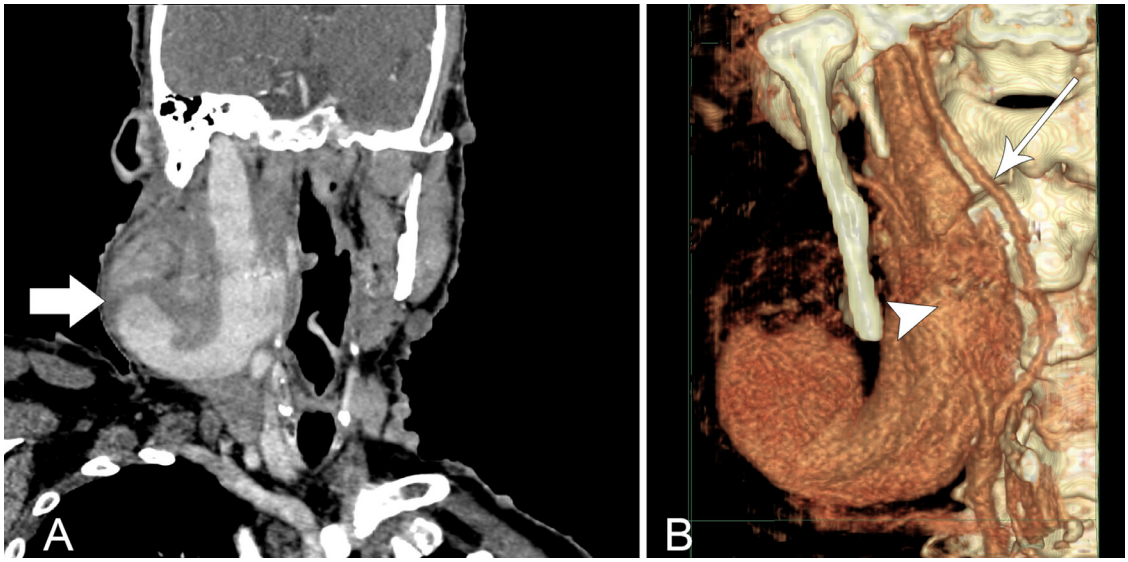

Fig. 5 – Contrast enhanced CT neck, (A) coronal view showing the large right internal jugular vein aneurysm (arrow) showing contrast enhancement. (B) 3D reformats anterior projection showing the extent of the aneurysm and its relationship with the ectatic internal jugular vein (arrowhead). Note the normal right internal carotid artery (long arrow).

Table 1 – Characteristics of internal jugular vein aneurysms in neurofibromatosis type 1 in literature [8].

Ref.	Age/Sex	Side	Size (cm)	Treatment	Thrombosed
Nopajoroonsri and Lurie (1996) [5]	62 M	Left	10 × 5 × 4.5	Resection	Yes
Oderich et al. [1]	73 M	Right	"Giant"	Resection	Yes
Belcastro et al. [2]	60 F	Left	12 × 12 × 10	Resection	Yes
Hiraki et al. [4]	60 M	Left	5.5 × 5 × 2	Resection	Yes
Delvecchio et al. [8]	63 F	Left	6.9 × 5.8 × 5.4	Resection	Yes
Present case	73 M	Right	8.4 × 8.0 × 7.0	Conservative	Yes

* M: Male; F: Female.

get large arteries and are often extra-parenchymal in nature [7]; a study by Oderich et al. revealed the renal artery (41%) to be the most affected by this rare disorder, the lesions presenting unilaterally and are characteristically stenotic. While other sites also include the head and neck vessels (19%) and the abdominal aorta (12%) [1,2]. In contrast, involvement of the venous system is rare and could be due to tumor growth, trauma or even occur spontaneously [4]. Thus far, there have been 5 other cases, in addition to our presented case, all of which described internal jugular veins aneurysms formation [1,2,4,5,8].

As summarized by Delvichio et al. in Table 1 [6], the patients with reported internal jugular vein aneurysms fell into the range of 60-73 years of age, with the gender, location and size of the swelling varying between the documented cases [1,2,4–6]. Furthermore, all of the aneurysms were found to be thrombosed following investigations and the main complaint was an enlarged swelling that required prompt surgical intervention [1,2,4,5,8], in contrast to our patient who presented to the ER with a completely different complaint.

The radiological modalities available for non-invasive diagnosis of jugular vein aneurysms include ultrasonography with color Doppler, CECT, CT angiography and magnetic resonance angiography [11,12]. Ultrasonography with Doppler is considered the first-line tool as it offers information about the extent of swelling as well as its relation to other neck structures. It can also differentiate cystic and solid lesions and differentiate vascular from non-vascular lesions [11–13]. Findings such as a flat wave on duplex sonogram can indicate that the swelling originates from a venous site, furthermore, performing the ultrasound before and after Valsalva maneuver can provide a more dynamic evaluation of the size of the swelling [8,11,13]. Our ultrasound findings showed a well-defined cystic lesion in the right side of the neck with curvilinear swirling mobile hypoechogenicities, but due to slow flow within the lesion, color doppler did not show any significant vascularity.

CECT also demonstrates a high diagnostic power in assessing neck masses, it provides insight regarding the deeper structures of the neck anatomy and may rule out any mechanical compression causes [11,13]. Additionally, it can detect thrombus within the vessel wall as in our case where a partial thrombosis was noted along the anterior aspect of the saccular right internal jugular vein aneurysm.

Of the documented cases of internal jugular vein aneurysms that underwent surgery, histopathology reports

post operation revealed reduction and disruption of elastic fibers in the tunica media layer [1,4], as well as NF1 localization within the vessel walls, these findings are consistent throughout the case studies and may explain the excessive fragility of the aneurysmal wall and the subsequent severe bleeding and difficulty to maintain homeostasis during the procedures [1,2,4,5,8].

As for the management of NF1 vasculopathies, factors such as the patient's age, type of vascular lesion, location, and size should be taken into account. A conservative approach is advisable considering most vasculopathies are clinically insignificant and self-limiting in nature [8], however surgical intervention is indicated when there is risk of thrombosis, rupture, worsening of symptoms or those who desire cosmetic treatment [8,9]. It is also important to note, that due to the fragile state of the aneurysmal wall, severe surgical complications such as rupture of the aneurysm sac and major bleeding are a possibility and thus management is determined case by case [2,4,5]. Our patient was asymptomatic regarding the swelling for months and so surgical intervention was not recommended at the time of his admission for the cardiac issues.

Conclusion

In summary, internal jugular vein aneurysms are considered some of the rarest NF1 vasculopathies that have a high rate of complications due to the neurofibroma infiltration into the vessel wall, this could be further complicated by thrombosis of the aneurysms which can lead to detrimental consequences [2,4,5]. And although routine vascular assessment cannot be recommended for all patients with NF1, high clinical suspicion and selective imaging and follow-up is advisable for clinicians [1].

Patient consent

The authors confirm that written informed consent has been obtained from the patient and that they have given approval for this information to be published in this case report and accompanying images.

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