World Journal of Advanced Research and Reviews
eISSN: 2581-9615 CODEN (USA): WJARAI
Cross Ref DOI: 10.30574/wjarr
Journal homepage: https://wjarr.com/

(RESEARCH ARTICLE)

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Catastrophic rupture of an internal carotid aneurysm in a neurofibromatosis type 1 patient: A critical case study

Salma Boulman [1,*], Ghita Filali [1], Hamza Dabachi [1], Samir El Youbi [1], Hamza Naouli [1], Hamid Jiber [1], Aabdellatif Bouarhroum [1] and Nawal Hamas [2]

[1] *Vascular surgery department, Chu Hassan II, Fez, Morocco.*
[2] *Pathology department, Chu Hassan II, Fez, Morocco.*

World Journal of Advanced Research and Reviews, 2024, 22(02), 665–671

Publication history: Received on 26 March 2024; revised on 08 May 2024; accepted on 10 May 2024

Article DOI: https://doi.org/10.30574/wjarr.2024.22.2.1374

Abstract

This paper reports a critical case of catastrophic rupture of an internal carotid aneurysm in a 30-year-old female diagnosed with Neurofibromatosis Type 1 (NF1). The patient, previously operated on for scoliosis and without known history of NF1, presented to the emergency department with a rapidly expanding latero-cervical mass following a minor infection treated with antibiotics. Initial examinations revealed a jugulo-carotid fistula and extensive vascular damage characterized by a ruptured aneurysm of the left internal carotid artery. Despite emergent surgical intervention aimed at vascular control and haemostasis, the patient succumbed to complications. This case highlights the vascular fragility associated with NF1 and underscores the need for heightened awareness of its possible severe manifestations in undiagnosed individuals. The findings suggest that vascular abnormalities in NF1, while rare, pose significant risks and require careful diagnostic scrutiny to prevent fatal outcomes. This study contributes to the limited knowledge on the vascular involvements in NF1, particularly the incidence and management of carotid artery aneurysms, urging for further research and better diagnostic protocols.

Keywords: Neurofibromatosis Type 1 (NF1); Carotid Aneurysm; Jugulo-Carotid Fistula; Catastrophic Bleeding; Vascular Fragility.

1. Introduction

Neurofibromatosis type 1 (NF1; von Recklinghausen disease) is an autosomal neurocutaneous disease characterised by generalized dysplasia of the mesodermal and neuroectodermal tissues (1-2) with a incidence of approximately 1 in 3000 births (3); It is caused by an NFI mutation placed on the long arm of chromosome 17.

NF1 can involve any organ, but mainly connective and nerve tissues are affected, it classically manifests with café au lait spots, neurofibromas and iris hamartomas (4).

In NF1, vascular complications represent the second most common cause of death, after malignant peripheral nerve sheath tumor (5). However, vascular involvement is relatively uncommon in NF1, with an estimated prevalence ranging from 0.4% to 6.4% (4).

A literature review of the vascular involvement in NF1 by Oderich et al. (4) found predominantly arterial involvement, with 41% occurring in the renal artery. Cervical arterial aneurysms occur in 13%, and 3% have arteriovenous malformations, but the incidence of their coexistence is unknown. Pathogenesis, clinical spectrum, and natural history of these abnormalities are unidentified.

* Corresponding author: Salma Boulman

In this paper we report the case of a patient with no previous history of neurofibromatosis, who presented to the emergency department of our institution with a ruptured aneurysm of the internal carotid artery associated with a jugulo-carotid fistula.

2. Case report

30-year-old patient operated on for scoliosis during her childhood, not known to be a carrier of neurofibromatosis type 1.

She was admitted to our clinic for the management of a ruptured aneurysm of the left internal carotid artery associated with a carotido-jugular fistula.

The history of her illness goes back to 5 days before her admission when the patient presented an angina put under antibiotic therapy, the evolution was marked by the appearance of an expansive latero-cervical beating mass, rapidly progressive with a thrill in view motivating the patient to consult for management.

On admission the patient was conscious, hemodynamically and respiratory stable and apyretic, with the presence of scoliosis and cafe au lait spots and several nevi on the neck and trunk.

Presence of an expansive and flapping left latero-cervical mass measuring approximately 8 cm with a thrill on palpation without inflammatory signs. (Figure 1)

An Angio scanner was done in emergency, showing: (Figure 2)

- A large left laterocervical hematoma extending from the prestylial space to the homolateral low jugulocarotid level fusing laterally to the parotid level and medially infiltrating and detaching the prepharyngeal space, it is also responsible for a significant mass effect on the oropharyngeal aero-digestive tract, it measures 77*50*40 mm, this hematoma is strongly enhanced after injection of contrast medium and presents a swirl sign testifying to its hyperacute bleeding.
- A voluminous extravasation of contrast product was identified in the left internal carotid artery, which presented an aneurysmal dilatation in its cervical portion with visualization of a fistulous path with the internal jugular vein, which was opacified early.

Figure 1 Large mass of the left side of neck of the patient and Café-au-lait spotty pigmentation of the patient's skin

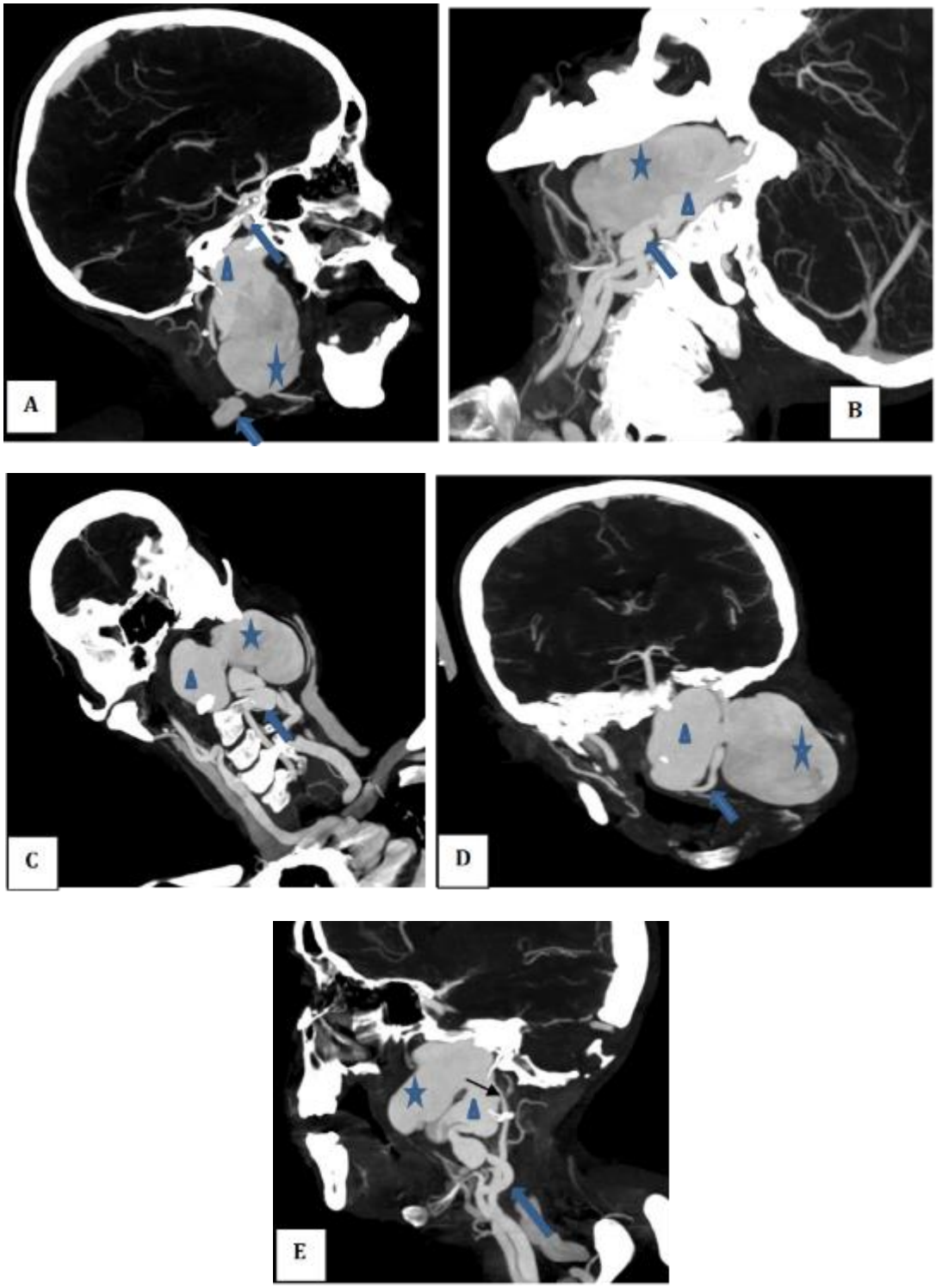

Figure 2 A. CT angiography showing: B. Left lateral cervical hematoma (star), C and D. Significant extravasation of contrast agent (triangle) from the aneurysmal internal carotid artery at its cervical segment (Blue arrow)., E. Fistula with the internal jugular vein, which is opacified early (black arrow)

A preoperative biological workup was requested with objective results (table 1)

- The patient presented a trismus and then the volume of the mass became much larger inducing a respiratory difficulty and a disorder of consciousness.
- The patient was intubated on respiratory criteria in the resuscitation room and then taken directly to the operating room for surgical exploration.

- In dorsal decubitus under general anesthesia, a log was placed under the shoulders with flexion and contralateral rotation of the head.

After disinfection with betadine and conventional draping, a pre-sterno-cleido mastoid incision was made, enlarged to the mastoid, and the left common carotid artery was dissected and checked first, then the left internal jugular vein was dissected and checked. Presence of active bleeding prompting clamping of the left common carotid artery after general heparinization.

2.1. On exploration

- Flattening of a false aneurysm of the left internal carotid artery with evacuation of hematoma, during the repair the wall of the left internal carotid artery was friable hence the need for bipolar ligation to ensure hemostasis.
- Evacuation of a huge hematoma reaching the mandibular angle above and the cervical spine behind.
- A breach in the left internal jugular vein was repaired with separate 5/0 prolene stitches.

During the procedure, samples of the artery and the hematoma were taken for pathological, mycological and bacteriological studies. (Table 2,3) (Figure 3).

- After an abundant washing and a careful haemostasis, we fermented plane by plane on a suction redon drain.
- The patient was sent to the intensive care unit for further treatment.
- The postoperative course was marked by a cardiorespiratory arrest on 3 occasions, which was resuscitated, and the patient died.
- A nevus biopsy and a blood sample for genetic study were done to confirm the diagnosis of NF1.

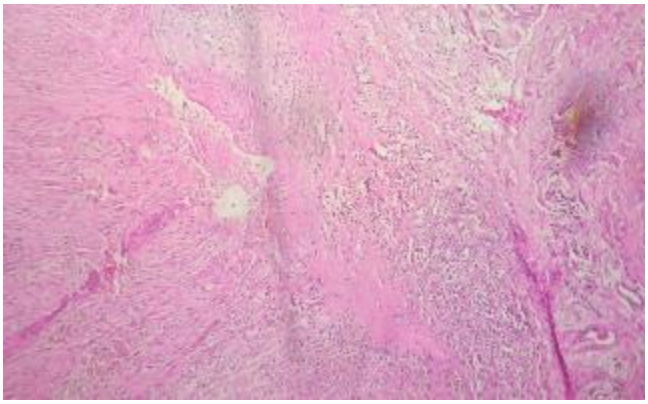

Figure 3 The results of the pathological study of the aneurysmal internal carotid artery revealed non-specific inflammatory residues.

Table 1 Results of the biological assessment

Laboratory Examination	**Mean**
Haemoglobin (12.50 – 15.50)	12 .30 g/dL
white blood cells (4.00 K – 10.00 K)	20.57 K/µL
Neutrophil (2.00 K – 7.00 K)	20.57 K/ µL: 91%
Platelet (150K – 400K)	160 K/ µL
Urea (0.17 – 0.43)	0.45 g/l
Creatinine (6.60 – 10.90)	5 mg/L
alkaline reserve (21.00 – 31.00)	17 mmol/L
C-Reactive Protein (0-5)	30 mg/L

Table 2 Mycological examination

exam direct	negative
culture	negative

Table 3 Cytobacteriological examination

cellular response	low
GRAM staining	No visible germs
bacteriological identification	negative

3. Discussion

NF is the most common single gene disorder of the nervous system; it was first described by von Recklinghausen in 1882 (6).

Types I and II von Recklinghausen's disease are the foremost common kinds of this malady. Depending on the location of the gene, type I (chromosome 17q11.2) can be differentiated from type II (chromosome 22q12).

Heterogeneity is the clinical hallmark of neurofibromatosis, and at least seven varieties of the disease have been described, of which type 1 is the most common (7)

NF1 is transmitted as autosomally dominant and represents an abnormal development of neuroectodermal tissue. As many as 50% of cases occur sporadically in patients without a family history of the disease (8).

Patients with NF-1 may have cutaneous neurofibromas, café-au-lait spots, freckling and hyperpigmentation in the trunk, axillary or inguinal areas, which are clues to the diagnosis.

Other features include Lisch nodules (hamartoma of the iris), kyphoscoliosis, dysplasia of the sphenoid, bowing and pseudarthrosis of the tibia, and an increased incidence of neoplasia, including pheochromocytoma, acoustic neuroma, meningioma, neurofibrosarcoma, astrocytoma, glioblastoma, and malignant schwannoma (8).

Our patient presented café-au-lait spots, freckling and hyperpigmentation in the trunk, cutaneous neurofibroma, kyphoscoliosis and no family history of neurofibromatosis.

Neurofibromatosis associated with vascular disorders is generally known as a vascular neurofibromatosis (9).

A study, on English literature spanning from 1957 to 2005 found that there were 237 individuals, with NF I showing 320 vascular abnormalities. The common lesions were identified in the renal artery representing 41% of the cases. These lesions were often unilateral in 68% of occurrences and tended to be more stenotic than aneurysmal. Additionally, abnormalities in the carotid cerebral arteries were detected in 19% of the patients mainly appearing as aneurysms. These issues typically arose during their thirties. Were more prevalent, among women (72%). Furthermore, cases involving coarctation or aneurysms which sometimes extended to the renal and mesenteric arteries accounted for around 12% of the total reported cases. (4)

Spontaneous ruptures from subclavian, intercostal arteries, thyrocervical, costo-cervical trunk, and peripheral arteries have been documented (2,10-14). The risk of rupture in NF1 patients with extracranial cerebrovascular manifestations is unknown, but it is thought to be higher than from aneurysms of other causes. (4,15). CT angiography is considered the gold standard in the diagnosis of ruptured aneurysms (12).

The heightened risk of rupture in NF1-affected arteries is attributed to their increased fragility, as described in various reports. This fragility stems from pathophysiological changes and degeneration within the arterial structure, specifically in the elastic lamina and the medial layer. (4,15) These arterial wall alterations are believed to be a consequence of

medial cystic degeneration and the disruption of the tunica elastica, which originates from smooth muscle. Additionally, these changes are linked to the presence of antibodies against desmin and muscle-specific actin. (4)

Two primary pathological mechanisms have been identified to explain arterial lesions in von Recklinghausen's disease. One mechanism involves the compression or infiltration of nearby blood vessels by neurofibromas, spindle cell proliferation, and nerve growth within the vessel walls. (1,2) Additionally, neurofibromin, which is typically found in vascular endothelial and smooth muscle cells, has been noted to be deficient in NF-I. This deficiency might disrupt the usual vascular maintenance and repair processes that are controlled by neurofibromin, as suggested by Riccardi. (16) Individuals with NF-I typically have a reduced lifespan when compared to the general population. In those with NF-1, the most frequent cause of death is due to cancers, especially tumors originating from connective and soft tissus. In younger patients, specifically those under 40, the second most common causes of death are vascular diseases and hypertension. (4) Carotid aneurysms are particularly dangerous for NF-1 patients, often leading to spontaneous ruptures and bleeding. It has been noted that pregnancy significantly increases the risk of such vascular complications, as highlighted in studies by Bertram et al. and Sobata et al.

Survival is shorter in patients with NF-I compared with the general population. The most common cause of death in patients with NF-1 is malignancy often from connective and soft-tissue neoplasms; however, in patients younger than 40, vascular disease and hypertension are the second leading causes of death [3]. Carotid aneurysms in patients with NF-1 are often associated with spontaneous rupture and bleeding, with pregnancy reported as a strong predisposing factor as reported by Bertram et al. [17] and Sobata et al. [18].

The treatment approaches for these conditions focus on securing the airway and achieving hemostasis, which may involve both surgical and non-surgical strategies. The choice of treatment is influenced by factors such as the patient's age, as well as the type and location of the lesion. In cases of carotid aneurysms in patients with NF-1, treatment typically necessitates surgical removal and subsequent reconstruction of the affected area.

In our situation, the aneurysm was positioned high above the bifurcation, and the delicacy of the area, further weakened by a surrounding hematoma, made it impossible to proceed with surgical reconstruction.

An endovascular technique can be utilized, employing stent grafts or sealing off the aneurysm with coils or glue. This approach is often favored for higher risk patients and for those whose aneurysms extend to the distal cervical carotid artery, where surgical access is considered challenging. (1) In certain cases, a mix of traditional surgery and endovascular methods may be appropriate. (10)

4. Conclusion

In summary, individuals with NF1 may exhibit a range of vascular complications. Among these, carotid artery aneurysms are a particularly uncommon manifestation of NF-1 vasculopathy. The invasion of the vessel wall by a neurofibroma can lead to significant weakening of both the aneurysmal wall and the adjacent tissues, posing a high risk of severe bleeding during surgical procedures. Moreover, these lesions are prone to abrupt ruptures, which can lead to catastrophic outcomes, including life-threatening obstruction of the upper airway. The authors believe that recognizing the potential for associated vascular lesions in patients with neurofibromatosis could help avoid misdiagnosis and its potentially fatal outcomes in the future.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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