Ruptured left external carotid artery aneurysm presenting as upper airway obstruction in von Recklinghausen’s disease

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**ABSTRACT**

**INTRODUCTION**: Neurofibromatosis type 1 (NF-1) is an autosomal dominant disorder characterized by café-au-lait macules, neurofibromas, and iris hamartomas. Carotid artery aneurysms rarely affect patients with NF-1 but may be associated with rupture. We report the first episode of a ruptured external carotid aneurysm with severe life-threatening airway obstruction in a NF-1 patient.

**PRESENTATION OF CASE**: We report a case of NF-1 32-year-old woman admitted to our department for life-threatening upper airway obstruction caused by spontaneous expanding swelling in the left sided neck. The diagnosis of ruptured aneurysm was suspected clinically and confirmed by computed tomography of the neck. The patient required tracheotomy for breathing difficulties and hemostasis was assured surgically by ligation of the external carotid artery.

**DISCUSSION**: Vascular abnormalities are rare but recognized manifestations of type NF-1. They often affect medium and large sized vessels. Carotid aneurysms are asymptomatic in most patients; they are subject to sudden rupture with potentially devastating consequences. CT angiography is the gold standard for diagnosis of aneurysmal rupture in NF-1 patients. Treatment consists on surgical ligation, resection and reconstruction or on percutaneous embolization.

**CONCLUSION**: Patients with NF-1 have a wide spectrum of vascular abnormalities. Particularly, aneurysms can be life-threatening for these patients; their surgical management must be urgent in these situations.

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1. Introduction

Neurofibromatosis type 1 (NF-1), or von Recklinghausen disease, is a rare autosomal dominant neurocutaneous genetic disorder characterized by a generalized dysplasia of mesodermal and neuroectodermal tissues [1,2]. Cardinal features of NF-1 include multiple café au lait macules, benign neurofibromas, and iris hamartomas [3]. Vascular abnormalities affect medium- and large-sized vessels and are recognized manifestations of NF-1 [1]. Stenotic lesions predominate, while aneurysms are less common [4,5]. The renal artery is the most frequent site of involvement [1,3]. Carotid artery aneurysms are rare but often present with spontaneous rupture or neurological complications [1]. Pathogenesis, clinical spectrum, and natural history of these abnormalities are unknown.

Here, we report an unusual case of von Recklinghausen disease revealed by a ruptured external carotid aneurysm causing neck swelling and life-threatening upper airway obstruction. In addition, a review of the literature is summarized.

2. Presentation of case

A 32-year-old North-African woman – without a background history of NF-1 – presented to the emergency department of our institution with sudden and severe left neck pain associated with a progressively spontaneous and painful expanding swelling in the left sided neck. She had difficulty in talking and swallowing. Her family history was negative for aneurysmal disease, neurofibromatosis, or other genetic abnormality. There was no history of surgery or medical disease. No definite trauma could be recalled despite repeated questioning.

Physical examination showed a large and tender mass of the left side of her neck from the clavicle up to the jaw causing significant airway compression and which did not display redness or pulsation (Fig. 1). There was café-au-lait spotty pigmentation of the skin that is typical in patients with von Recklinghausen's disease (Fig. 2); no benign neurofibromas were observed.

While she was tachycardic at 130/min, her blood pressure was 120/75 and her oxygen saturation was 98% on room air. A hemoglobin value of 5.2 g/dl suggested the cause of the acute
swelling to be massive bleeding. Computed tomography (CT) scan revealed a large hemorrhage in the left side of the neck with active extravasation from the external carotid artery (Fig. 3). Blood transfusion and inotropic support with noradrenaline were commenced. Angiography was not performed because not available in our institution.

The patient presented suddenly a severe breathing difficulty and was immediately taken to the operating room. After five failed orotracheal intubation attempts, an emergency tracheotomy was performed. Surgical neck exploration under general anesthesia showed a large cervical hematoma. The bleeding from the external carotid artery was identified and appeared to be a ruptured pseudo-aneurysm. We proceeded to hemostatic measures, culminating in ligation of the external carotid 10 mm up to the superior thyroid artery. The patient had no complications. Tracheotomy was removed at day 3. Follow-up at 12 months revealed no other vascular complications.

3. Discussion

Vascular abnormalities are well-recognized but rare manifestations of NF-1 and comprise of stenoses, aneurysms, pseudoaneurysms, and rupture or fistulas formation, occlusions and arteriovenous malformations, mainly of the larger blood vessels [2]. The term NF-1 vasculopathy has been coined in the medical literature to describe these vascular abnormalities [1]. The frequency of this vasculopathy is hard to define. It is reported to occur in 0.41%–6.4% of the patients [3] and to involve mainly larger blood vessels. Lin et al. [6] found a 2% prevalence of cardiovascular abnormalities among 2322 participants in the National Neurofibromatosis Foundation database.

Most patients with NF-1 vascular abnormalities are asymptomatic and have involvement of multiple vessels [7]. The most common clinical presentation is renovascular hypertension associated with renal artery stenosis in the childhood [1,3]. The renal artery is the most frequently involved, but abdominal aortic coarctation, internal carotid artery aneurysms, and cervical vertebral arteriovenous malformations have also been described [1,4]. Spontaneous ruptures from subclavian, intercostal arteries, thyrocervical, costo-cervical trunk, and peripheral arteries have been documented [2,4,5,8–10]. To the best of our knowledge, this is the first report of a rupture from the external carotid artery to be associated with neurofibromatosis. CT angiography is considered the gold standard in the diagnosis of ruptured aneurysms [8].

There are two major distinct pathologic mechanisms explaining arterial lesions associated with von Recklinghausen’s disease. The vascular anomaly may be the result of compression or infiltration of adjacent blood vessels by a neurofibroma, spindle cell proliferation, and proliferation of nerves within blood vessel walls [1,2]. Moreover, neurofibromin expression has been demonstrated in the vascular endothelial and smooth muscle cells. This suggests that deficiency in neurofibromin in NF-1 may cause alteration in the normal process of vascular maintenance and repair regulated by neurofibromin as proposed by Riccardi [11].

Survival is shorter in patients with NF-1 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. [12] and Sobata et al. [13].

The principles of treatment involve airway rescue and haemostasis, including surgical intervention and non-operative management. Treatment depends on the patient’s age and the
type and location of the lesion. Treatment of carotid aneurysms in patients with NF-1 more often requires surgical excision or reconstruction. An endovascular approach is possible using stent grafts or exclusion of the aneurysm with coils or glue; it is preferable in higher risk patients and in those with extension of the aneurysm to the distal cervical carotid artery, where operative exposure is judged to be difficult [1,7]. In some instances a combination of open surgical and endovascular methods are indicated [4]. The choice of conduit is somewhat controversial because of the potential risk of aneurysmal degeneration with vein grafts in younger patients and the risk of complications with an endovascular approach. Bertram et al. [12] concluded that, although this mode of repair is feasible but challenging because of the risk of complications, it is a better option than open repair in cases with difficult distal exposure of the involved vessels. Also, Hamasaki et al. [14] and Oderich et al. [3] judged that the endovascular stenting and coil embolization for carotid artery aneurysm associated with NF-1 were considered safe and effective.

4. Conclusion

Carotid artery aneurysms are even a rarer form of presentation for a NF-1 vasculopathy. The exact pathogenesis and natural history remains to be determined. Because these lesions are subject to sudden rupture with potentially devastating consequences as life-threatening upper airway obstruction, aneurysms require a high index of suspicion in von Recklinghausen’s disease patients.

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Author contribution

Hajar Souldi: writing the paper.
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