SPONTANEOUS RUPTURED OF RIGHT DEEP BRACHIAL ARTERY PSEUDOANEURYSM WITH UNDERLYING NEUROFIBROMATOSIS TYPE 1

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Abstract

Neurofibromatosis type 1 (NF-1) is known to develop vascular lesions, yet they are uncommon. If missed or ignored, it can be limb-threatening. When a ruptured pseudoaneurysm is clinically identified, rapid surgical intervention is required. We discuss a case of NF-1 that involved a spontaneous rupture of the right deep brachial artery. A 41-year-old woman reported sudden right arm swelling and intensifying pain. She was hemodynamically stable; however, her right arm was enlarged and tense on examination. A radiological test revealed a deep brachial artery haemorrhage. The patient was promptly referred to a vascular surgeon and had endovascular intervention performed. Early detection, emergency angiography, and endovascular treatment for vascular lesions should be initiated immediately.

Keywords: Neurofibromatosis, Pseudoaneurysm, Spontaneous Rupture

Introduction

There are three types of neurofibromatosis: Neurofibromatosis type I (NF-1), Neurofibromatosis type 2 (NF-2), and Schwannomatosis, of which NF-1 is the most prevalent form and one of the most common neurologic autosomal dominant diseases (1). Patients with NF-1 have a wide range of vascular anomalies, notably aortic, renal, and mesenteric aneurysms or stenoses (2). The majority of NF-1-related vascular lesions are asymptomatic. However, vasculopathy frequently manifests as a rupture of an undetected lesion, which can be devastating if disrupted (3).

Bleeding into an NF-1 lesion seldom involves the brachial artery. Bleeding might result from minor trauma or, in rare cases, spontaneous bleeding. Given such vascular fragility, endovascular treatment may be preferred for NF-1-related vasculopathy (4). In this case, we present a spontaneous right deep brachial artery pseudoaneurysm rupture in a patient with NF-1 who was successfully limb salvaged by endovascular therapy.

Case Report

A 45-year-old Malay lady presented to the Emergency Department with abrupt swelling and severe pain over the right arm, associated with hand and finger numbness, and motility impairment. She was diagnosed with NF-1 during childhood but defaulted to treatment and follow-up when she was twelve years old. This patient has no history of trauma, intravenous drug misuse, recent local or systemic illness, or arteritis. She has no risk factors for atherosclerosis and was not taking any medication. The Emergency physician attended the case, and an acute haemorrhage from a neurofibroma or vasculopathy lesion was suspected.

In the emergency department, the patient was alert and conscious with a respiration rate of 20 breaths/min, blood pressure of 138/79 mmHg, heart rate of 102 beats/min, a temperature of 37.0°C, and a pain scale of 8. A physical examination revealed swelling over the right arm without rashes or ecchymosis (Figure 1). Her right upper limb was flexed and abducted with a tense and tender arm compartment, while distal pulsation and circulation were intact. Multiple fibromas were seen on the body, while the other physical examination was unremarkable. The haemoglobin value was 12.8 g/dl.

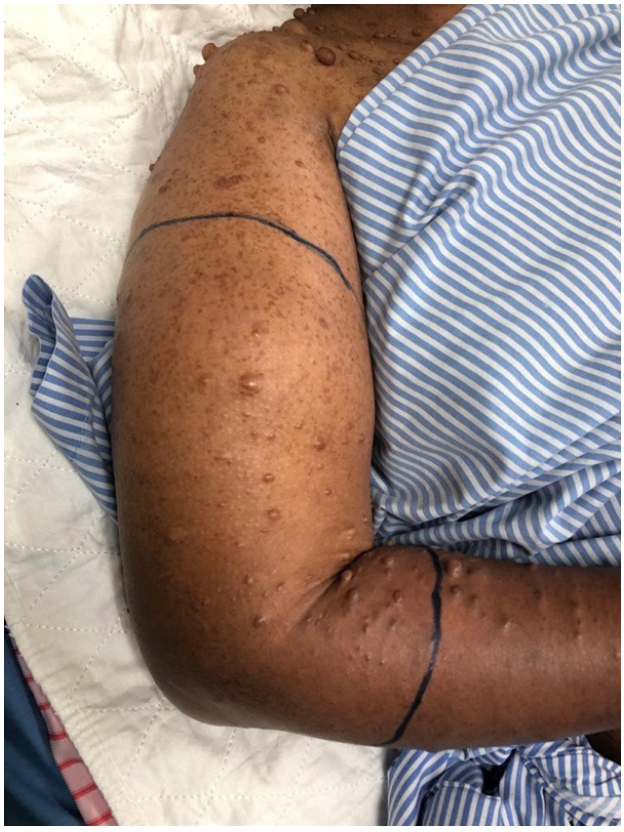


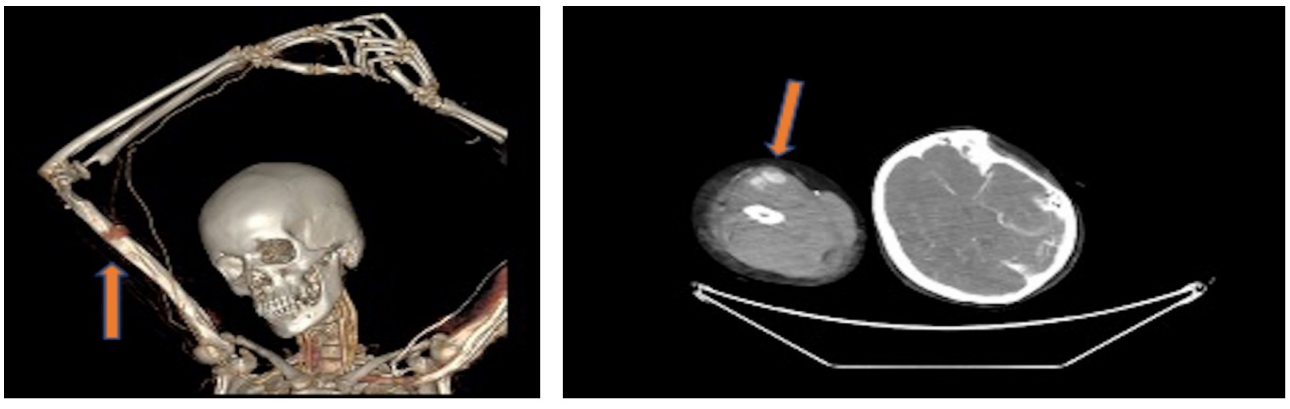
Figure 1: Physical examination revealed swelling over the right arm, with multiple neurofibromas seen.

A bedside scan done by the Emergency physician showed an intact brachial artery but revealed an area of hypoechogenic without Doppler colour flow concerning a fluid collection. Computed tomography angiography (CTA) was conducted. The CTA images showed a pseudoaneurysm arising from the deep brachial artery at the distal third of the right arm and a massive haematoma involving the anterior and posterior compartments of the right arm (Figure 2).

The case was immediately referred to the vascular team in Hospital Kuala Lumpur due to the lack of expertise at the local hospital. An emergent endovascular angiography was performed the next day. The right deep brachial artery (arteria profunda brachii) was identified intraoperatively as the major feeder of the pseudoaneurysm. The artery branching from the axillary artery was successfully embolised with a metallic coil. She had a smooth recovery with outpatient follow-ups under the surgical team. After the swelling had subsided, she was referred to the orthopaedic team as an outpatient for right upper limb neurology assessment.

Discussion

Neurofibromatosis type 1 (NF-1), NF-2, and Schwannomatosis are a set of autosomal dominant illnesses that predominantly affects the neurological system. NF-1, in particular, affects a range of organ systems (1). NF-1 is



(a) (b)

Figure 2: (a) 3D Computed tomography angiography (CTA) of the right upper limb. **(b)** Axial view of CTA of right upper limb. A multilobulated saccular outpouching is suggestive of a pseudoaneurysm arising from the deep brachial artery at the level distal 1/3rd of the right arm. It measures approximately 1.4 cm (AP) x 2.2 cm (W) x 2.1 cm (L). The neck measures about 0.5 cm. A huge heterogeneous enhancing soft tissue lesion is seen involving the anterior and posterior compartments of the right arm, suggestive of hematoma. It measures approximately 10.6 cm (AP) x 8.6 cm (W) x 14.0 cm (CC).

the most prevalent form of NF, commonly known as Von Recklinghausen's illness, which affects roughly 1 in 3,500 people globally and is caused by mutations on chromosome 17 in the NF-1 gene (5). Hallmark cutaneous findings include cafe´-au-lait spots, dermal neurofibromas, axillary freckles, and some plexiform neurofibromas (5, 6).

Despite significant improvements in diagnosis and treatment, people with NF-1 still have a life expectancy of 8 to 15 years lower than that of the general population. Malignant development in neurofibromas is the most frequent cause of death, followed by vascular complications (7). The first description of NF-1's vascular manifestations,

including occlusion, stenosis, aneurysm, and ectasia, was made in 1945 (8). The majority of NF-1 vasculopathy patients are asymptomatic, with multiple affected vessels. The frequency is unknown, but it is rare (9).

Numerous studies have shown that a neurofibromin deficiency is likely to be the pathogenesis of NF-1-associated vasculopathy. The tumour suppressor gene NF-1 encodes neurofibromin, a large and multifunctional protein that ensures normal vascular and nervous system development (10). Mutation of the gene function results in poor homeostasis, vascular inflammation, and endothelial and smooth muscle cell proliferation, especially after vessel injury, which causes distorted vessels (11, 12). Meanwhile, neighbouring neurofibromas can invade or compress vascular walls (12, 13). Over time, vascular stenosis, occlusion, aneurysm, pseudoaneurysm, rupture, or fistula might occur.

Aneurysms have been documented to rupture spontaneously at diverse places. The bleeding might be severe and lethal without immediate reorganisation and haemostatic therapy. The most common site of vasculopathy is the renal artery, followed by the cerebrovascular circuit and subclavian/vertebral vasculature. Vasculopathy of extremity vessels has been reported but is relatively rare (14).

As vasculopathy is increasingly recognized, the diagnosis is identified early in this case—the clinical manifestation of abrupted limb swelling with intense pain caused by massive haemorrhage of the rupture pseudoaneurysm. Duplex ultrasound is the primary assessment tool in the emergency department for screening aneurysms or pseudoaneurysms. CTA is rapid, readily available, and provides a high spatial resolution of the brachial artery, which assists the treatment plan. It can also mark the presence of neurofibromas.

Due to the extreme fragility of the arterial wall and the difficulties in obtaining haemostasis, the potential for significant intraoperative bleeding, surgical repair, or vessel reconstruction of the ruptured aneurysm is challenging and risky (15, 16). From the literature review, only two cases successfully underwent brachial artery reconstruction (17, 18). Despite the significant mortality and morbidity, most failed operations have resorted to salvaging techniques such as amputation, resection, and ligation.

Endovascular treatment can be the initial selection because it is a less invasive and readily accessible procedure. According to a systematic review, even in haemodynamically unstable patients, endovascular treatment of NF-associated aneurysms is safe and efficacious (19). Surgical treatment such as reconstruction with a vein graft is suggested for traumatic brachial artery injury in a patient with NF-1 rather than embolisation (20). In this case, endovascular embolisation with a metallic coil for the spontaneous rupture of the right deep brachial artery was successful without complication.

Due to their low occurrence, it is debatable whether frequent screening for vascular lesions is necessary for all asymptomatic NF-1 patients. However, multiple aneurysms have been found in NF-1 patients. Therefore, patients treated for vascular incidents should undergo vascular imaging (21).

Conclusion

Vascular complication in neurofibromatosis is rare and might be clinically silent but can manifest later as limb or life-threatening. Early identification of vasculopathy, followed by emergent angiography and endovascular therapy, is life-saving.

Competing interest

The authors declare that they have no competing interests.

Financial Support

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Ethical Clearance

Verbal informed consent was obtained from the patient in this case report.

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