Brachial artery aneurysm rupture in a patient with neurofibromatosis: a case report

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ABSTRACT

Peripheral vascular manifestations of neurofibromatosis are rare but may result in fatal haemorrhaging when they rupture. Surgeons should be aware of this life-threatening condition. We report a case of 35-year-old woman with neurofibromatosis who presented with a swollen and tender mass around her right arm. Angiography revealed 2 aneurysms in the brachial artery. Surgical occlusion revealed a large amount of clotted blood within the subfascial space, and the bleeding point was identified as a pinpoint opening in the aneurysm. The brachial artery abutting the aneurysm and the surrounding soft tissues was extremely brittle and fragile, with massive oozing during dissection. The brachial artery was irreparable and was resected after ligation of the artery and surrounding soft tissues and the aneurysm. Despite an uneventful recovery, the patient died on day 4.

Key words: aneurysm; brachial artery; neurofibromatoses

INTRODUCTION

Neurofibromatoses are hereditary diseases with varied phenotypic expressions, characterised by the abnormal growth of tissues of mesodermal and neuroectodermal origin, sometimes involving osseous and central nervous systems. Multiple skin tumours and spotty pigmentation—cafe-au-lait spots—are common symptoms. Vascular manifestations of neurofibromatosis (characterised by stenosis, occlusion, aneurysm, pseudoaneurysm, and fistula) are rare but can be catastrophic. Cases with involvement of the renal, cerebrovascular and visceral arteries have been reported, but it is uncommon in major peripheral arteries. We report a rare case of a brachial artery aneurysm rupture in a patient with neurofibromatosis.

CASE REPORT

In November 2004, a 35-year-old woman presented to our hospital with acute onset of pain and swelling in her right upper arm. She was known to have
neurofibromatosis and had undergone excision of cutaneous neurofibromas on 2 occasions 10 years previously. She was lethargic and complained of intractable pain, weakness, and numbness in her right hand. She had multiple cutaneous fibromas and café-au-lait spots over her entire body. Pulses of the radial artery of the right wrist were palpable but weaker than those of the left side. Her blood pressure was 100/60 mm Hg and heart rate was 120 beats/min. Blood tests revealed a haemoglobin level of 97 g/l, a haematocrit of 30.1%, and a white cell count of 24 500/μl. A peripheral blood smear had microcytic hypochromic features with anisotrophy.

Magnetic resonance imaging of the right arm revealed a large haematoma with an arterial aneurysm. Angiography revealed 2 aneurysms in the brachial artery. The smaller one was located in the middle of the brachial artery with a narrow pedicle and the larger one was located more distally and had a broad base (Fig. 1). Right common carotid and vertebral angiographies demonstrated no further aneurysms. Transarterial embolisation was successfully performed in the small aneurysm but failed in the larger aneurysm. Surgical occlusion of the larger aneurysm revealed a large amount of clotted blood within the subfascial space, and the bleeding point was identified as a pinpoint opening in the aneurysm. Both the brachial artery abutting the aneurysm and the surrounding soft tissues was extremely brittle and fragile, with massive oozing during dissection. The brachial artery was irreparable and was resected after ligation of the artery, the surrounding soft tissues and the aneurysm. Despite recovering well postoperatively, the patient died on day 4 after losing consciousness with an immeasurably low blood pressure. No autopsy was performed but it was suspected that massive bleeding had occurred from another site, possibly proximal to the brachial artery ligation.

The characteristics of the proliferating tissue in the aneurysm and around the vessels were determined using haematoxylin and eosin staining and immunohistological staining with antibody for S-100 protein. Tissue from the aneurysm showed elastic fragmentation and loss of media smooth muscle cells, which were replaced by spindle cells in the myxoid stroma (Fig. 2a). These spindle cells stained positively for S-100 protein, suggesting neurofibroma in the vessel wall (Fig. 2b). The histological findings of vessels immediately proximal and distal to the aneurysm were very similar to those of the tissue at the aneurysm. The proliferating tissue was also positive for S-100 protein and had destroyed the arterial wall adventitia and infiltrated the smooth muscle fibres in the media (Fig. 3).

**DISCUSSION**

Neurofibromatosis is the most common single genetic disorder of the human nervous system. It is inherited as an autosomally dominant trait and is characterised by disordered growth of neuroectodermal tissue. Classic manifestations include café-au-lait spots and neurofibromas, with a wide spectrum of concurrent pathologies, including intracerebral, extracerebral and extracranial schwannomas, meningiomas, gliomas, astrocytomas, neuroblastomas, pheochromocytomas, rhabdomyosarcomas, and even leukaemia, along with hydrocephalus, macrocephaly, and bony defects.

Vascular manifestations are classified into 3 types based on vessel size: pure intimal, intimal aneurysmal, and nodular. Intimal proliferation involves breakdown of muscle and elastic layers and adventitial nodular thickening.\(^1\) Many arterial lesions are often overlapped and it is difficult to classify an individual lesion as a single ‘pure’ type.\(^2\) Proliferation of Schwann cells in the arterial walls with secondary degenerative change is the basic pathogenesis of the vascular lesions in neurofibromatosis.\(^3\) In larger vessels (aorta, carotid, and proximal renal artery), direct invasion by Schwann cells, intimal thickening, and destruction of the media and elastic tissue leads to either stenosis or aneurysm.\(^3\) In smaller vessels,
mesodermal dysplasia causes the proliferation of smooth muscle in the intima leading to stenotic lesions and, occasionally, poststenotic aneurysms, as seen in our case where tumour cells directly invaded the vessel wall and formed aneurysms without mesodermal dysplasia.

The incidence of vascular manifestations of neurofibromatosis (e.g. stenosis, occlusion, and aneurysm) has been reported to be 3.6% in 110 patients, but the number appears to have risen recently. The 3 types of vascular manifestations may coexist, with stenosis being the most frequent.

Stenosing or aneurysmal lesions in the renal arteries, causing renovascular hypertension, has been reported, as have aneurysms in the superior mesenteric, splenic, hepatic, carotid, vertebral, and coronary arteries. Involvement of the peripheral arteries is uncommon; clinical manifestations include abrupt swelling, pain, and hypovolaemic shock caused by massive haemorrhage of the ruptured aneurysm. Neurofibromatosis causes arterial weakening and haemorrhage. In larger vessels, direct tumour invasion causes tissue compression of the vasovasorum and wall weakening. In smaller vessels with a less prominent vasovasorum, fibrohyaline thickening of the intima and fragmentation of the intima and muscularis result in arterial wall weakening and rupture. The fragility of the vessel walls and hypertrophied surrounding tissue make surgical correction difficult. Although some authors have successfully re-created a vascular channel using
a bypass graft, many have failed and have adopted salvage procedures such as amputation, resection, and ligation, despite the high mortality and morbidity. Endovascular embolisation is recommended when the circulation distal to the aneurysm is available via collateral vessels. In our patient, endovascular embolisation failed to occlude the large, broad-based aneurysm, necessitating surgical occlusion by resection and ligation of the involved vessel and surrounding tissues. Regrettably, the patient died on postoperative day 4. The cause of death was not determined because no autopsy was performed. It was suspected that massive bleeding had occurred from another site, possibly proximal to the brachial artery ligation. Similar clinical and pathological findings including a huge neurofibromatous mass invading into the surrounding brachial artery have been reported. We speculated that the fatal haemorrhage was caused by rupture of a weak vessel wall secondary to direct invasion by a neurofibroma. Surgeons should be aware of this life-threatening condition.

REFERENCES