BRITISH JOURNAL OF HOSPITAL MEDICINE

CASE REPORT

A lucky escape: popliteal aneurysm in a patient with neurofibromatosis type 1

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Introduction

Neurofibromatosis is an autosomal dominant genetic disease characterised by abnormal growth involving tissues of mesodermal and neuroectodermal origin. The disease affects multiple organs, with vascular involvement found in approximately 10% of cases (Von Recklinghausen, 1882). Association of neurofibromatosis with aneurysmal disease (particularly in peripheral arteries) has rarely been reported. This article presents a patient with neurofibromatosis type 1 and a popliteal aneurysm requiring emergency surgery.

Case report

A 42-year-old woman presented to the emergency department with a critically ischaemic left foot of 48 hours' duration. Her foot was paraesthetic, painful and she was unable to mobilise. No treatments she had attempted improved her symptoms, hence she presented to hospital.

The patient had a background history of a total abdominal hysterectomy for atypical endometrial hyperplasia and neurofibromatosis type 1, which had been diagnosed 12 years ago through genetic testing. She had no history of hypertension, hypercholesterolaemia, diabetes mellitus or tobacco use. Other than hormone replacement therapy, she was prescribed no other regular medications. She had no known allergies and was unaware of any significant family history because of a lack of contact with her biological parents. She had no prior symptoms of intermittent claudication.

On physical examination the patient was afebrile and haemodynamically stable. She had numerous café-au-lait spots on her body. Her abdomen was soft with no palpable masses or bruits. On general inspection, the circumference of her left calf was slightly larger than that of the right. The left foot was cold to touch and there were palpable femoral, popliteal and posterior tibial pulses but an absent dorsalis pedis pulse. All contralateral pulses were present. She had altered sensation at the toes.

A duplex ultrasound performed on admission excluded a deep vein thrombosis, but showed a popliteal artery aneurysm. A subsequent computed tomography angiogram (Figures 1 and 2) confirmed an aneurysm of the left popliteal artery measuring 2.7 cm containing significant clot burden. There was occlusion of the left anterior tibial artery and a distal filling defect in the peroneal artery noted (Figure 3), thought to be embolic. The posterior tibial artery remained patent. No further aneurysms at other sites were identified. She was therefore managed as having an acute, embolising popliteal artery aneurysm.

Following admission, the patient was commenced on an intravenous infusion of unfractionated heparin aiming for an activated partial thromboplastin time ratio of 2.0–2.5. A popliteal artery aneurysm repair was performed the following day through a medial approach (exclusion bypass) using ipsilateral non-reversed great saphenous vein, tunnelled anatomically from the above knee popliteal artery to the tibio-peroneal trunk. The tissue planes were particularly challenging because of the inflammatory tissue response around the vessels. On completion, no flow was identified in the posterior tibial artery so a microtibial embolectomy was performed, restoring good flow to the foot.

Postoperatively, the patient was admitted to the vascular high dependency unit and required return to theatre for four compartment lower limb fasciotomies that evening having developed acute compartment syndrome. The patient improved clinically post-surgery and the fasciotomy wounds were slowly brought together. She was discharged on postoperative day 14 with a well perfused foot, but requiring a splint for a degree of foot drop.

At a 6-week follow-up clinic appointment, her remaining fasciotomy wounds had virtually healed. A graft surveillance duplex scan showed good graft patency with triphasic waveforms within the body of the graft and no flow seen in the excluded aneurysmal segment. Her foot drop had largely recovered.

How to cite this article:
Abed H, Dattani N, Hobbs S, Wall M. A lucky escape: popliteal aneurysm in a patient with neurofibromatosis type 1. Br J Hosp Med. 2020. https://doi.org/10.12968/hmed.2020.0281

Figure 1. Reconstructed computed tomography angiogram of lower limb vasculature showing left-sided popliteal artery aneurysm (arrow 1) with patent posterior tibial vessel but distal occlusion of the peroneal and anterior tibial (arrow 2).

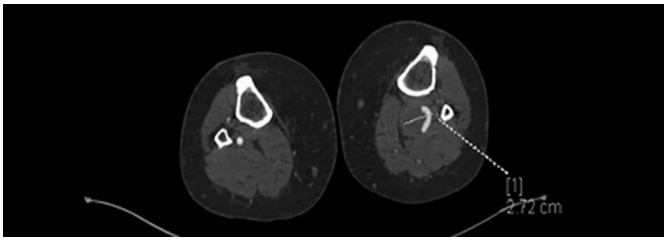

Figure 2. Transverse computed tomography angiogram image showing a left-sided popliteal aneurysm measuring 27 mm in diameter containing significant thrombus.

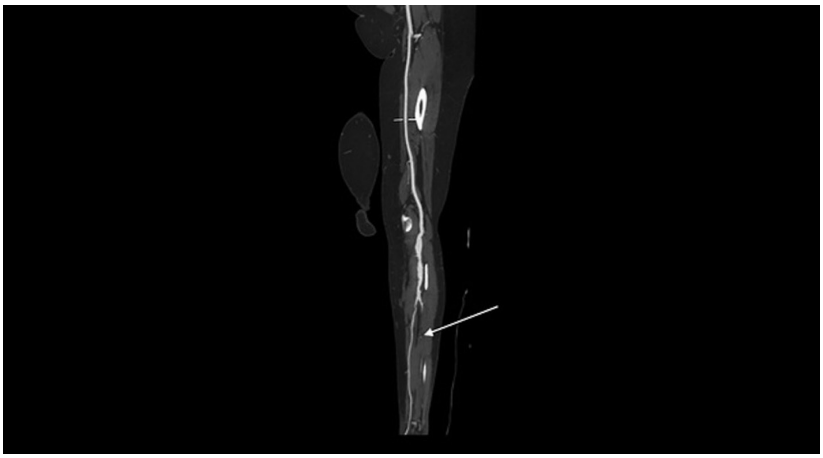

Figure 3. Sagittal computed tomography angiogram image showing occlusion of the anterior tibial artery (arrow) on the left.

Discussion

Neurofibromatosis is an autosomal dominant disease characterised by abnormal growth involving tissues of mesodermal and neuroectodermal origin. It was first described by Von Recklinghausen (1882), with the most common sub-type being neurofibromatosis type 1. Vasculopathy in neurofibromatosis type 1 is well described in the literature, but is rarely encountered clinically.

In a literature review, Bueno et al (2005) identified six cases of lower limb vasculopathy associated with neurofibromatosis, of which four were aneurysmal. Of the cases published, the diagnoses of aneurysmal disease were made either because of sudden growth, rupture or an incidental finding. To the best of the authors' knowledge, this is the first case reporting a popliteal artery aneurysm in a patient with neurofibromatosis type 1 presenting with

Learning points

- It is important to be aware of the presence of an association between neurofibromatosis type 1 and peripheral arterial disease, in particular aneurysmal disease.
- Peripheral arterial disease should be considered in younger patients with a background history of neurofibromatosis type 1, with or without the presence of predisposing comorbidities.
- Surgical treatment is recommended in cases of symptomatic or complicated lower limb aneurysmal disease.
- Operative strategies may differ in patients with neurofibromatosis, with the specific procedure decided on a case-by-case basis.

symptoms of an acutely ischaemic limb. A combination of the symptomatology, absence of dorsalis pedis pulse and demographics supported an embolic phenomenon.

Operative strategies may differ in patients with neurofibromatosis type 1, with options including ligation, aneurysm exclusion and arterial reconstruction with a bypass graft (Young et al, 2001). On review of the literature, there is minimal evidence supporting one management option over another. One case of a ruptured brachial artery aneurysm proposed that diffuse infiltration of neurofibromatous tissue pertained poorer quality vessel wall and hence precluded ligation and bypass grafting procedures (Saitoh and Matsuda, 1998). An aneurysm exclusion bypass was deemed the most suitable for the case described.

The association of neurofibromatosis type 1 and peripheral arterial disease, in particular aneurysmal disease, is not common and this case highlights the importance of its recognition. Clinicians need to be suspicious when faced with acute leg pain in patients with a background of neurofibromatosis. Surgical treatment is recommended in cases of symptomatic or complicated lower limb aneurysmal disease, with the specific procedure decided on a case-by-case basis. Consideration should be given to performing arterial imaging in patients with a history of neurofibromatosis type 1.

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References

Bueno A, Acín F, Rodríguez JM et al. Ruptured popliteal aneurysm resulting from neurofibromatosis. Vasc Endovascular Surg. 2005;39(5):449–455. https://doi.org/10.1177/153857440503900511

Saitoh S, Matsuda S. Aneurysm of the major vessels in neurofibromatosis. Arch Orthop Trauma Surg. 1998;117(1–2):110–113. https://doi.org/10.1007/BF00703457

Von Recklinghausen F. Ueber Die Multiplen Fibrome Der Haut and Ihre Beziehung zu Den Multiplen Neuromen. Berlin: A. Hirschwald; 1882:138

Young LP, Stanley A, Menzoian JO. An anterior tibial artery aneurysm in a patient with neurofibromatosis. J Vasc Surg. 2001;33(5):1114–1117. https://doi.org/10.1067/mva.2001.112704