Endovascular anatomical reconstruction of the failed open aortic repair for non-aneurysmal rupture of the infrarenal aorta in Neurofibromatosis Type 1

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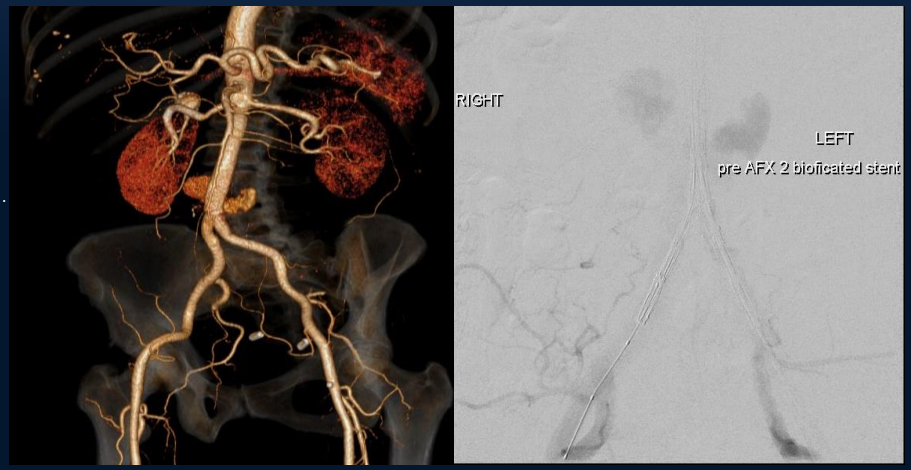
Introduction

The autosomal dominant genetic disorder Neurofibromatosis type 1 with an incidence of 1 in 3000 is due to the mutation of NF1 gene which causes a reduction or loss of the function of the protein Neurofibromin found in many tissues. This causes a spectrum of clinical manifestations, dependent on the particular pathogenic NF1 mutation, including connective tissue abnormalities. Rare cases of arterial dissection with life threatening haemorrhage have been observed.
Presented is the case of a failed open surgical repair for a non-anuerysmal rupture of the infrarenal aorta in a patient with a prior diagnosis of neurofibromatosis. Successful repair was, however, achieved with implantation of an AFX® Unibody stent to resolve a symptomatic pseudoaneurysm at aortic bifurcation at the distal aortic graft anastomosis.

Case Presentation

A 60yo female with a diagnosis of NF1 presented to our institution with sudden onset back and abdominal pain with haemodynamic instability. She was found on CT imaging to have a non-aneurysmal rupture of the infrarenal aorta (13mm) with a small associated dissection requiring emergency open repair. A 10mm Dacron tube graft was used for reconstruction. This was technically challenging due the soft nature of the aortic tissue. Haemostasis was achieved on the third attempt at the proximal anastomosis with pledglets and an external dacron cuff required. Recovery was as expected for open aortic repair with discharge from hospital occurring 18 days following repair. Ultrasound imaging performed 10 weeks post-operatively in association with lower back pain revealed a large pseudoaneurysm (4 cm²) at the distal anastomosis. The patient was promptly re-admitted. Endovascular repair was employed given the expected hostility of the abdomen 10 weeks post open repair and the known fragility of the aortic tissue. The small calibre of the native aorta determined the use of the AFX® unibody graft. This had a low profile design suitable for the narrow vasculature and achieved the desired anatomical reconstruction of the aortic bifurcation excluding the pseudoaneurysm. The patient was discharged 48hrs post operatively and no further issues or intervention have subsequently occurred



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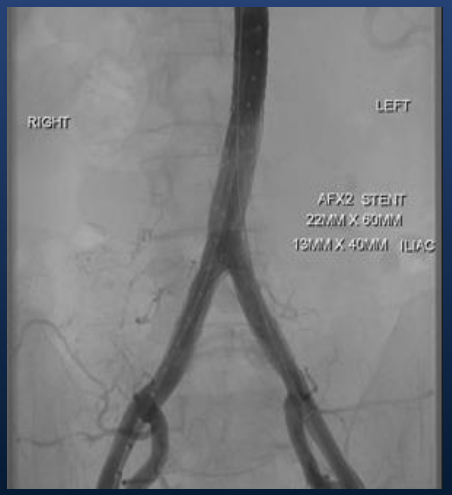
Discussion

Neurofibromatosis type 1, also known as Recklingshausen disease, is the most common form of NF. It is an autosomal dominant genetic disorder affecting 1 in 3000 individuals. Neural tumours, Classical café-au-lait macules, freckling and Lisch nodules are all hallmarks of the condition (1). Mutations of the NF1 gene located on chromosome 17 cause a reduction or loss of the function of the protein Neurofibromin found in many tissues (2, 3).

The spectrum of clinical manifestations is dependent on the particular pathogenic NF1 mutation. Connective tissue abnormalities including vasculopathies are a recognised cause of morbidity and mortality in NF1 (4). The exact cause of these remains poorly understood. There are many documented arterial abnormalities postulated as causality for the observed vasculopathies, including neurofibromas within the arterial wall. No isolated histological characteristic that encompasses all vasculopathies observed in NF1 has been documented (4). Of the observed vasculopathies occlusive and aneurysmal disease affecting the renal and abdominal aorta are the most common with mesenteric and peripheral involvement less common (5). Less well documented are cases of dissection with life threatening haemorrhage (6).
Oderich et al in their review of vasculopathies observed in NF1 noted no cases of dissection and documented no increased fragility of tissues with any of their observed aneurysmal abdominal aortic cases. They did note increased tissue fragility in two patients with non-aortic aneurysms (4). They found no increased risk of complication with angiographic procedures in NF1 patients and recommend consideration of an endovascular approach for aneurysmal repair particularly in the older patient (4).



An open approach was employed by our institution due to the haemodynamic instability of the patient; timely access to the angiographic suite was not feasible. Additionally uncertain causality of the rupture and desire for durable repair in a younger patient determined an open approach for repair. This was technically challenging due to the fragile nature of the tissue. When subsequent repair of the large symptomatic pseudoaneurysm at the distal anastomosis was required an endovascular approach was employed. This was due to the known fragility of the tissues and hostility of the abdomen post open repair.

A straight 10mm Dacron tube graft was used for the initial open repair. Given the close proximity of the distal anastomosis to the aortic bifurcation, this had to be reconstructed in order to achieve exclusion of the pseudoaneursym. Considerations that guided treatment were the narrow calibre of the aorta and iliofemoral vessels. CERAB and traditional EVAR are less favourable repair options in this context due to the risk of flow competition between limbs and loss of future cross-over approach which was felt to be significant in the setting of a concomitant asymptomatic right renal artery aneurysm. The AFX® unibody stent was chosen as this uses relatively low profile access (17F) and low profile design for an aortic device. Its construction as a unibody covered stent scaffold allowed anatomic reconstruction of the aortic bifurcation with exclusion of the pseudoaneurysm whilst maintaining equal limb perfusion and future cross-over approach.

Conclusion

Aortic dissection is an observed cause of morbidity and mortality in the spectrum of clinical manifestations of NF1. Tissue fragility should be considered in the repair approach for vasculopathies requiring treatment in NF1. Endovascular repair with the AFX® unibody stent graft achieves a successful anatomical reconstruction in a non-aneurysmal aorta when open approach is unsuccessful.