Anterior spinal artery syndrome from type A aortic dissection in a patient with Marfan syndrome due to a novel fibrillin mutation

K Cheng[1], M Perenyei[2], R Sayeed[3]

Abstract

Type A aortic dissection is a rare but important cardiac surgical emergency. Few reports exist in the literature describing anterior spinal artery syndrome as a presenting feature.

We report a case of anterior spinal artery syndrome due to aortic dissection in a patient with Marfan syndrome caused by a novel fibrillin mutation. A 53-year old female presented with chest pain and sudden-onset paralysis. Neurological examination revealed normal upper limb examination, reduced lower limb power and reflexes but normal sensation. CT scanning revealed type A acute aortic dissection which was treated with emergent cardiac surgical repair. At clinic follow up 3 years later, signs of Marfan syndrome were opportunistically noted and genetic testing revealed a novel mutation in the *Fibrillin 1* gene.

This case emphasises the importance of a good initial clinical assessment, including thorough neurological examination, as well as a low threshold of clinical suspicion for an aortic dissection in such a constellation of symptoms. The importance of family history should also be emphasised given the coincidental diagnosis of Marfan syndrome in a first-degree relative. Furthermore, this case illustrates the classical signs of anterior spinal artery syndrome.

Keywords: anterior spinal artery syndrome, aortic dissection, cardiac surgery, chest pain, fibrillin, Marfan syndrome

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Correspondence to:
K Cheng
National Heart and Lung Institute
Royal Brompton Hospital
Sydney Street
London SW3 6NP
UK

Email:
kevin.cheng@doctors.org.uk

Background

This case illustrates an unusual complication of a rare but important condition that may present to the Emergency Department, Acute General Medicine or Cardiology; it highlights the importance of a detailed clinical assessment because of the unusual association of chest pain and sudden-onset paralysis and the patient was not the index case in her family. We also report a new mutation in the *Fibrillin 1* (*FBN1*) gene responsible for Marfan syndrome.

Acute aortic dissection has an incidence of 5–30 per million per year and accounts for 1 in 10,000 hospital admissions. Treatment depends on the Stanford classification: type A dissections involving the ascending aorta/aortic arch require primary surgical intervention whereas type B dissections involving the descending aorta may be treated medically at the first instance. Although rare, it is associated with 80% mortality over the first two weeks. Aortic dissection commonly presents with severe chest pain that may radiate to the back although painless dissections have been reported.[1–5] While up to a third of acute aortic dissections present with neurological symptoms, a small minority present with spinal cord ischaemia: anterior spinal artery syndrome is a rare presentation – only 2–8% present with paraplegia with or without sensory loss.[6,7]

Case presentation

Clinical history and examination

A 53-year-old female was admitted complaining of sudden-onset sharp, central chest pain radiating to her back. Breathless and vomiting, she had collapsed without loss of consciousness; simultaneously she developed numbness and weakness in both lower limbs. She had been an otherwise fit non-smoker with no risk factors for ischaemic heart disease. She had a history of pulmonary tuberculosis, bilateral hip replacements and lower back pain. Her son had undergone recent cardiac surgery.

On examination, she was alert, cold and pale. She was bradycardic (40 bpm regular), hypotensive (80/40 mmHg)

[1]Honorary Clinical Research Fellow, Royal Brompton Hospital, London; [2]Core trainee, [3]Consultant Cardiothoracic Surgeon, John Radcliffe Hospital, Oxford, UK

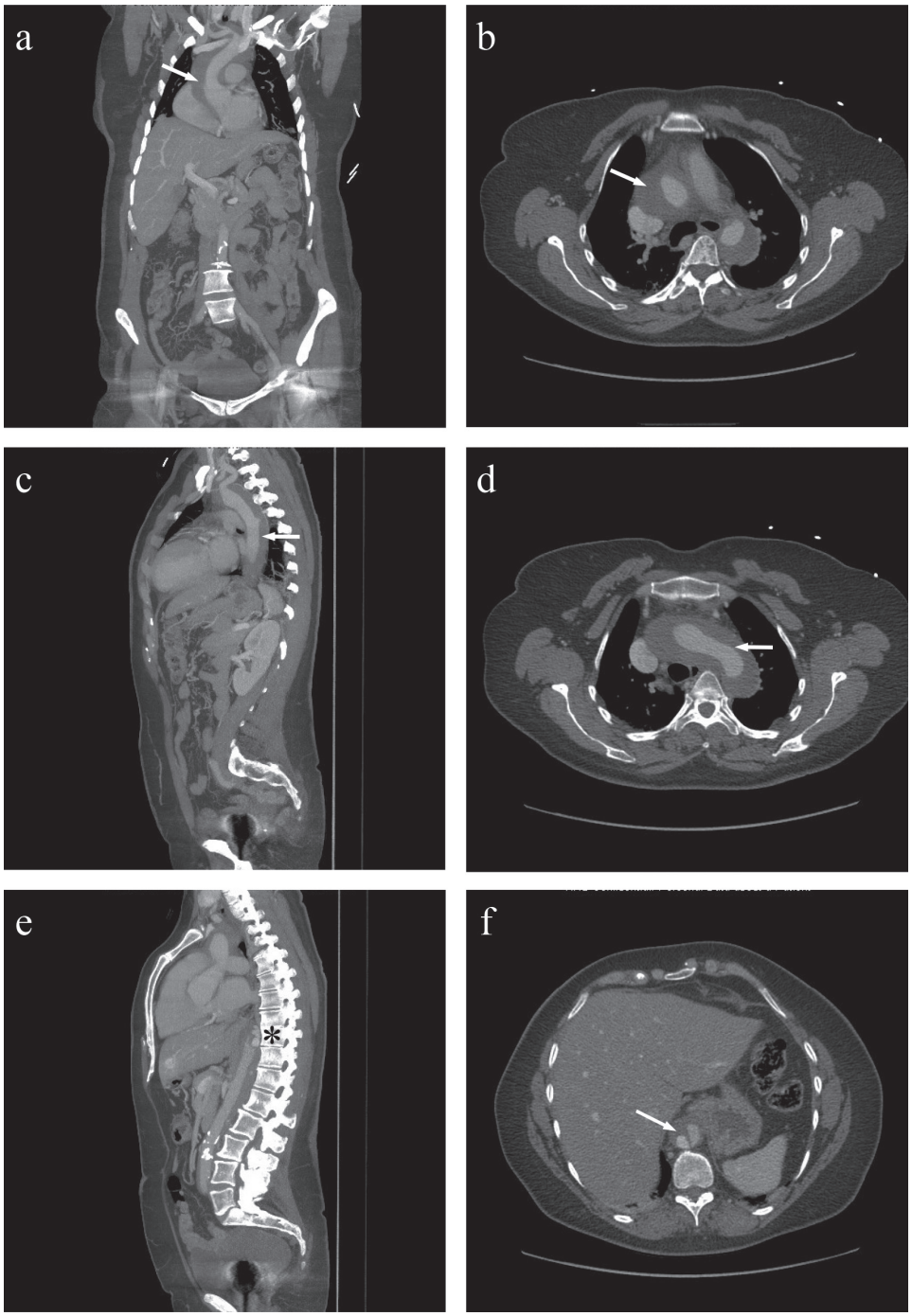

Figure 1 CT aortogram with contrast showing type A aortic dissection. a) Coronal image showing dissection in the aortic root and ascending aorta, b), c) and d) Dissection in the ascending aorta, thoracic descending aorta, and aortic arch, e) Sagittal section showing dissection in the abdominal descending aorta with blood in the false lumen (*), f) Axial section showing dissection in the abdominal descending aorta with an intimal flap (arrow)

but afebrile (35.3°C). Mildly tachypnoeic (20 breaths/min), she had an SpO_2 98% on 10 l/min oxygen through a non-rebreathing mask. Her chest was clear and she had mild epigastric tenderness but no palpable masses or abnormal pulsation. All peripheral pulses were palpable with no radio-radial delay. Neurological examination revealed normal upper limb power (5/5) but markedly reduced power (0/5) in her lower limbs. Her legs were flaccid with reduced deep tendon reflexes and absent plantar responses but sensation to light touch, pain and temperature was normal.

Investigations

Initial investigations included an arterial blood gas that showed anaemia (haemoglobin 113 g/L) and a raised lactate (1.7 mmol/L). An electrocardiogram showed sinus bradycardia with ST elevation in leads II, III, and aVF. Transthoracic echocardiography showed a dilated (4.5 cm) aortic root and mild aortic regurgitation.

Differential diagnosis

The differential diagnosis included inferior myocardial infarction and acute aortic dissection, although neither was an obvious cause of her paraplegia. Urgent contrast-enhanced CT confirmed a type A aortic dissection extending from the aortic root to the right iliac artery (Figure 1) and she was referred for emergency cardiac surgery.

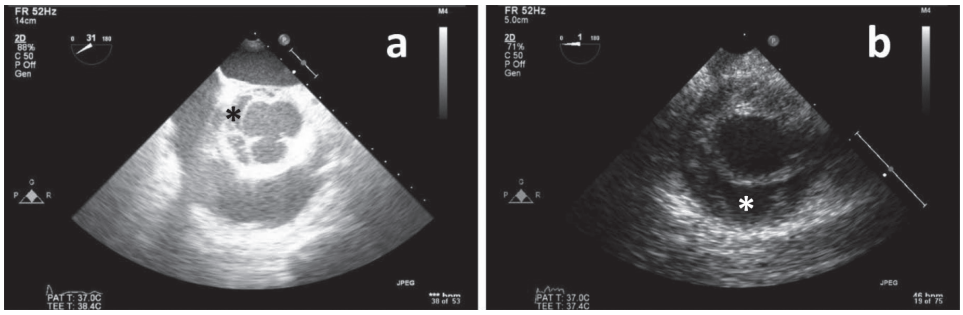

Figure 2 Peri-operative transoesophageal echocardiography. Type A aortic dissection affecting (a) the aortic root and (b) continuing into the descending aorta. Dissection denoted by *

Treatment and outcome

Peri-operative transoesophageal echocardiography showed a Type A aortic dissection involving the aortic root and extending around the arch into the descending aorta with normal left ventricular function (Figure 2). She arrested following anaesthetic induction and required immediate sternotomy to release an incipient tamponade caused by rupture of the dissection. Surgical repair involved an interposition graft to replace the ascending aorta, re-suspension of the native aortic valve and a long saphenous vein bypass graft to the distal right coronary artery for suspected malperfusion.

Admitted to the intensive care unit postoperatively, examination of her lower limbs following extubation 5 days after the event revealed normal tone but reduced power both proximally (1/5) and distally (2/5). There was altered sensation to light touch bilaterally, knee jerk reflexes were present bilaterally and ankle jerk reflexes absent with plantar reflexes equivocal. A sensory level at T8 was found. Preserved dorsal column function (proprioception, light touch, and vibration sense) led to a diagnosis of anterior spinal artery syndrome secondary to occlusion of the artery of Adamkiewicz owing to type A aortic dissection extending into the abdominal aorta.

At clinic follow up three years after surgery, it was incidentally noted that the patient had skin striae and hyperextensile carpal, interphalangeal and metacarpophalangeal joints, raising the clinical suspicion of Marfan syndrome. She was not tall nor did she have a high-arched palate, but she was significantly short-sighted, had an arm span wider than her height, mild pectus carinatum, and had suffered a prolapsed lumbar disc a few weeks before her presentation with aortic dissection. Her son had an aortic valve replacement following infective endocarditis and had been diagnosed with Marfan syndrome after genetic testing. She was found to harbour the same heterozygous mutation as her son, a nonsense mutation c.3031G>T in exon 24 of the *FBN1* gene. At her latest 7-year follow up, transthoracic echocardiography showed mild to moderate aortic regurgitation and good left ventricular function. Magnetic resonance aortography showed stable postoperative appearances with small false aneurysms adjacent to the proximal and distal interposition graft margins and a mild bulge of the proximal descending aorta. She remains wheelchair-bound but otherwise well.

Conclusion

This case serves to remind clinicians that sudden-onset chest pain may suggest a diagnosis of aortic dissection and such a unifying vascular event may be responsible for an acute presentation of paraplegia. Furthermore, this case illustrates a basic understanding of the vascular supply to the spinal cord to explain the constellation of symptoms presenting in anterior spinal artery syndrome. We are aware of only one other such case in the literature.[8]

Acute aortic dissection extending into the descending thoracic aorta may cause malperfusion or occlusion of the segmental arteries supplying the spinal cord, including the artery of Adamkiewicz that provides a reinforcing supply to the anterior spinal artery, typically arising between T6 to L3 on the left side of the aorta.[1] Compromise of the artery of Adamkiewicz results in ischaemia of the anterior regions of the cord (the anterior spinal artery supplies the anterior two-thirds) while the dorsal columns are spared due to their supply from the posterior spinal artery. Lesions at T4–6 are particularly common because this is a 'watershed' territory.[1,9,10] In contrast, the sacral spinal cord is relatively resistant to ischaemia because of an extensive supply from the lumbar, iliolumbar, lateral and median sacral arteries anastomosing with the posterior spinal arteries; continence may thus be preserved in anterior spinal artery syndrome.[11]

The organisation of the intrinsic blood supply of the cord – the central branches and pial arterial plexus – further explains the clinical findings in anterior spinal artery syndrome. Occlusion of the central branches, which arise from the anterior spinal artery, results in infarction of the grey and adjacent white matter – consequently, vibration sense and proprioception are spared, and patients present with paraparesis, areflexia, and loss of pain and temperature sensation.[11] Interestingly, as in our patient, the spinothalamic tracts may be spared due to the pial arterial plexus, which arises from circumferential branches from both the anterior and posterior spinal arteries – anastomotic branches may thus preserve perfusion despite infarction of the anterior spinal artery.[7,11]

Finally, this case emphasises the importance of a thorough family history given that the patient was not the index case in her family. Subsequent genetic testing revealed a nonsense mutation (c.3031G>T) in exon 24 of the *FBN1* gene. A search of the Universal Mutation Database (UMD-*FBN1*) for nonsense mutations in the *FBN1* gene did not reveal previous reports of this mutation.[12]

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