EJVES Extra (2011) **22**, e22—e23

SHORT REPORT

Ruptured Left Subclavian Artery Aneurysm in a 41-Year-Old Woman with Neurofibromatosis Type 1

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Submitted 4 May 2011; accepted 9 June 2011

KEYWORDS
Neurofibromatosis type 1;
Artery aneurysm;
Subclavian artery;
Endovascular treatment

Abstract *Introduction:* Intrinsic lesions of the arterial wall are important manifestations of Neurofibromatosis type 1.
Report: A 41-year-old woman with Neurofibromatosis type 1, suffering sudden onset of upper back as well as left shoulder and upper chest pain is addressed to our hospital. The contrast-enhanced thoracic computed tomogram demonstrated a huge hematoma due to ruptured left subclavian artery aneurysm treated with endovascular therapy.
Discussion: A ruptured left subclavian artery is an uncommon but life threatening manifestation in Neurofibromatosis type 1.

Introduction

Neurofibromatosis type 1 (NF-1) or von Recklinghausen's disease has a wide variability of clinical expression affecting skin, eyes, bones and nervous and vascular systems. Subclavian artery aneurysm, described in this work, is an uncommon manifestation of this kind of disease and may be fatal if not promptly recognised.

Case report

A 41-year-old woman suffering sudden onset of upper back as well as left-shoulder and upper-chest pain was addressed to our hospital. At the emergency room, a bulge in the supraclavicular area was evident and, on physical examination, she was tachycardic, tachypnoeic and dyspnoeic. Her heart rhythm was tachycardic but regular. The

DOI of original article: 10.1016/j.ejvs.2011.06.046.
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1533-3167
doi:10.1016/j.ejvsextra.2011.06.003

contrast-enhanced thoracic computed tomogram (CT) demonstrated a huge haematoma in the same zone. Admission haemoglobin level and haematocrit were 8.8 g dl^{-1} and 26.3%, respectively. As an emergency, under local anaesthesia, using Seldinger technique and the left-femoral artery approach, a selective 5F catheter was advanced into the left subclavian artery, and the arteriogram demonstrated a 2.1-cm ruptured left subclavian artery aneurysm 1.5 cm after the origin of the vertebral artery. The bleeding was successfully stopped with a 10 × 40 mm *Fluency plus* stent graft.

The patient was discharged from the hospital on day 7, without any sequelae. The planned follow-up included 1-month CT angiography and Doppler ultrasound assessment, 6-month Doppler ultrasound assessment and then yearly CT angiography, with extended assessment of the aorta and its main branches.

Past medical history was significant for neurofibromatosis, manifested by dermal and spinal neurofibromas, café-au-lait spots and kyphoscoliosis of the cervical and thoracic spines. There was no history of injury or operation in the left supraclavicular region, clavicle fracture, or left subclavian vein catheter-insertion attempt. There was no neurologic deficit.

Discussion

NF-1 or von Recklinghausen's disease is one of the most common genetic diseases, affecting 1/3000—1/4000 individuals. It has a wide variability of clinical expression affecting skin, eyes, bones and nervous and vascular systems. The most common vascular abnormality in patients with NF-1 is renal artery stenosis.[1,2]

NF-1, or von Recklinghausen's disease, is caused by an abnormality of chromosome 17 and has an incidence of 1 in 2500—3300 live births. Patients with NF-1 may have: cutaneous neurofibromas, café-au-lait spots, freckling and hyperpigmentation in the axillary or inguinal areas, pigmented iris hamartomas, optic gliomas, distinctive osseous abnormalities, Schwann cell tumours, intracranial tumours, neurologic impairment, kyphoscoliosis, macrocephaly, syringomyelia and pheochromocytoma. Most patients with NF-I have asymptomatic abnormalities of multiple vessels. The renal artery is involved in most cases, but abdominal aortic coarctation, carotid artery aneurysms and cervical vertebral arteriovenous malformations are other common manifestations.

The pathogenesis of the arterial-wall lesions remains unclear. The pathological changes may cause fragility of the vessel and may result in aneurysms and/or rupture. Likely arterial abnormalities in these patients occur by a process of cellular proliferation and fibrosis, involving neurofibromin expression in the vascular endothelial and smooth muscle cells.[3,4]

From the literature review, carotid, vertebral and cerebral aneurysms occur in the third decade of life and are more frequent in women (72%). Cervical vertebral arteriovenous malformations are a classic presentation and may be the late result of contained ruptured vertebral artery aneurysms. Spontaneous subclavian artery rupture, upper- and lower-extremity aneurysms and arteriovenous malformations have been documented.[5]

Clinically significant arterial lesions are relatively uncommon, but their consequences can be extremely dangerous. If an arterial abnormality is detected on an exhaustive first evaluation of an NF-1 patient, radiological noninvasive imaging of the head, chest and abdomen appears indicated because of the multiplicity and dangerousness of the vascular lesions. Endovascular treatment may be a successful option to deal with this condition, as previously reported in the literature.[4]

Conflict of Interest

None declared.

Funding

None declared.

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