SHORT REPORT

Lower Limb Arterio-venous Fistula as a Late Complication of Phlebectomy in a Patient with Ehlers-Danlos Type IV

R.E. Brightwell a,*, P.J. Walker b, c

a Department of Biosurgery, Imperial College London, St Mary’s Hospital Campus, Paddington, London, UK
b Department of Surgery, University of Queensland Centre for Clinical Research, Brisbane, Queensland, Australia
c Department of Vascular Surgery, Royal Brisbane & Women’s Hospital, Brisbane, Queensland, Australia

Submitted 15 November 2010; accepted 20 June 2011
Available online 22 July 2011

KEYWORDS
Arterio-venous fistula; Ehlers-Danlos type IV; Varicose vein

Abstract  Ehlers-Danlos syndrome type IV (EDS type IV), the vascular type, results from mutations in the gene for type III procollagen (COL3A1). Affected patients are at risk for arterial, bowel, and uterine rupture. The timing, frequency and course of these events are unpredictable. We report a 50-year-old patient with previous complications of EDS type IV who presented with recurrent varicose veins that subsequent imaging identified as an arteriovenous fistula (AVF) at the site of previous phlebectomy.

Patients with EDS type IV present vascular surgeons manifold management problems. A pre-existing diagnosis of EDS type IV should alert the clinician to the risk of unusual presentations, both acutely and as complications subsequent to intervention. Once identified, appropriate investigation and follow-up of these patients by a vascular surgeon is mandated.

Crown Copyright © 2011 Published by Elsevier Ltd on behalf of European Society for Vascular Surgery. All rights reserved.

Introduction

Vascular complications of EDS type IV are protean,1 with most clinicians aware of the associated life-threatening presentations. We report a 50-year-old female with previous complications of EDS type IV who presented as an outpatient with subtle signs that subsequent imaging identified as a significant AVF.

Case Report

A 50-year-old female with genetically-proven EDS type IV was referred to our clinic by a Clinical Geneticist for
vascular surgical opinion. Sequencing of genomic DNA had shown that the patient was heterozygous for a missense non-conservative mutation c.1916G > T in the COL3A1 gene. This mutation is predicted to replace the glycine at position 639 of the triple helix domain with a valine.

The patient had a background history of uncomplicated caesarean section 21 years earlier and right saphenofemoral junction ligation, great saphenous vein stripping and multiple phlebectomies at a point shortly thereafter. Spontaneous rectal perforation complicated by peritonitis and managed with a Hartmann's procedure (subsequently reversed) occurred 2 years later. The following year the patient underwent splenectomy for spontaneous capsule rupture and 8 years thereafter she developed a spontaneous, massive rectus sheath haematoma. 2 years after that she required emergent right nephrectomy for spontaneous renal vein rupture.

The patient was asymptomatic at time of review, but was aware of some prominent veins in the right lower limb on the anterolateral aspect of her calf. She was normotensive and had a regular resting pulse rate of 76 beats per minute. Cardiac auscultation revealed dual heart sounds with a soft ejection systolic murmur. Examination of the peripheral pulses was normal throughout. It was noted that the varicose veins remained prominent even when the patient was recumbent. There was no thrill but auscultation revealed a loud 'machinery' bruit, most intense over a previous phlebectomy scar.

Suspecting an AVF (and to exclude other latent complications of EDS type IV) imaging consisting of duplex ultrasonography (DUS — Fig. 1) and CT angiography (CTA — Fig. 2) was requested. DUS demonstrated low resistance fistulous flow in the common femoral, superficial femoral and popliteal arteries with velocities ranging from 124 to 200 cm/s. There was monophasic, antegrade flow in the anterior and posterior tibial arteries, while flow in the peroneal artery was reversed. The popliteal vein was aneurysmal. CTA demonstrated that the right common and superficial femoral arteries were larger than those on the left. In the region of the calf that was affected by 'varicose veins' a complex network of vessels was seen, with arterial-phase flow identified in the popliteal and superficial femoral veins.

Clinically there was no evidence of steal syndrome or ischaemia affecting the right lower limb. There were no clinical signs of high-output cardiac failure and her chest radiograph and echocardiogram were normal. In view of the fact that she was haemodynamically stable, that she herself was reluctant to consider intervention, and her previous history of complicated EDS type IV, a decision was made to manage this AVF conservatively.
Discussion

Any surgery in a patient with EDS type IV carries a significant complication (especially bleeding) risk with some suggesting only ‘essential’ procedures are undertaken.\(^2\) Elective varicose vein surgery in this group of patients has been complicated by severe vascular injury including femoral vein laceration, and even endoluminal procedures carry increased risk.\(^3,4\) It therefore seems prudent to manage varicose veins in patients with known EDS type IV conservatively if at all possible.

Acquired AVF near the level of the knee is well represented in the literature and the majority of cases result from vascular trauma or penetrating injury.\(^5\) often associated with orthopaedic procedures such as joint replacement.\(^6\) AVF complicating endovenous laser treatment of truncal varicose veins has also been reported.\(^7\) Also spontaneous arteriovenous fistula formation in EDS type IV patients has been previously reported\(^8\) so it would appear natural that that AVF could complicate varicose vein surgery in these patients. However, the authors believe this is the first case of AVF complicating standard varicose vein surgery in a patient with EDS type IV.

EDS type IV, the vascular type, results from mutations in the gene for type III procollagen (COL3A1). Clinical diagnosis is established from at least two of four major clinical criteria: thin, translucent skin; arterial, intestinal, or uterine fragility or rupture; extensive bruising; and characteristic facies. Genetic testing confirms the clinical suspicion. Pepin et al. have written an exhaustive paper on the clinical and genetic features of EDS type IV.\(^9\)

Vascular assessment should be performed in all cases and include clinical examination, carotid and abdominal DUS in asymptomatic patients, and non-invasive imaging of chest and abdomen if incidental findings are detected. Due to the fragility of vessels in affected individuals, a well-accepted strategy for the management of vascular complications of EDS type IV is a conservative approach (as we adopted), with surgical or endovascular treatment reserved for patients with imminent or frank life-threatening haemorrhage.\(^1\)

Funding

None.

Conflict of Interest

None.

References