A life-threatening complication of Ehlers–Danlos syndrome

Introduction

Ehlers–Danlos syndrome predisposes to the development of vascular malformations, which may present as life-threatening haemorrhage. This article presents a patient with Ehlers–Danlos syndrome in whom spontaneous rupture of an unusual arteriovenous fistula occurred. This rare diagnosis can be reached through close attention to the history and examination.

Discussion

Ehlers–Danlos syndrome is attributed to the Danish dermatologist Edvard Ehlers and the French physician Henri-Alexandre Danlos. It comprises a heterogeneous group of inherited disorders arising from defective collagen synthesis and metabolism, which can be classified into at least ten types on the basis of clinical, genetic and biochemical information (Beighton et al, 1997). The manifestations of Ehlers–Danlos syndrome are many and include vascular malformations, i.e. non-athero-matous aneurysms and arteriovenous fistulae (Beighton et al, 1997).

Arteriovenous fistulae, which occur far less commonly than aneurysms, typically form spontaneously between the cavernous sinus and carotid artery (Germain and Herrera-Guzman, 2004). Arteriovenous fistula of extracranial blood vessels are quite rare; to the authors' knowledge this is the first description of an arteriovenous fistula arising from the external iliac artery. The authors suspect that the fistula arose as a consequence of the spinal surgery the patient underwent as a child. This is supported by a report of an iatrogenic hepatoportal fistula secondary to abdominal surgery (Sherry et al, 1992).

Interestingly, numerous cases of vascular malformations in Ehlers–Danlos syndrome have been established solely from clinical criteria (Oderich et al, 2005). Similarly, the diagnosis in this case was formulated based on the patient's history combined with the clinical findings of unilateral Grey–Turner's sign and an audible bruit.

Conclusions

This case serves as a reminder of the potentially fatal vascular complications of Ehlers–Danlos syndrome. Moreover, it emphasizes that history and examination form the basis of clinical diagnosis, even when confronted with an unusual complication of a rare syndrome. BJHM

Dr Sam Behjati *is Academic Clinical Fellow (Year 1) in Paediatrics, UCL Institute of Child Health, London WC1N 1EH,* **Dr Yolande Knight** *is Academic Clinical Fellow (Year 1) in Public Health Medicine, Imperial College, London,* **Dr Archana Goebells** *is Core Trainee (Year 1) in Acute Care Common Stem,Bedford Hospital NHS Trust, Bedford,* **Dr Rudi Borgstein** *is Consultant Radiologist, North Middlesex University Hospital NHS Trust, London and* **Miss Fiona Myint** *is Consultant Vascular Surgeon, North Middlesex University Hospital NHS Trust, London, and Royal Free Hampstead NHS Trust, London*

Correspondence to: Dr S Behjati

Beighton P, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ (1998) Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK). *Am J Med Genet* **77**(1): 31–7

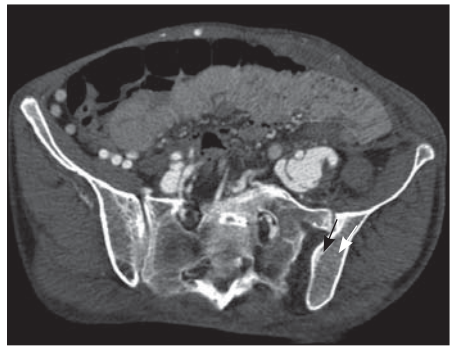
Germain DP, Herrera-Guzman Y (2004) Vascular Ehlers-Danlos syndrome. *Ann Genet* **47**(1): 1–9

Oderich GS, Panneton JM, Bower TC et al (2005) The spectrum, management and clinical outcome of Ehlers-Danlos syndrome type IV: a 30-year experience. *J Vasc Surg* **42**(1): 98–106

Sherry RM, Fisch A, Grey DP, Lubbock CA (1992) Embolization of hepatoportal fistula in a patient with Ehlers-Danlos syndrome and colon perforation. *Surgery* **111**(4): 475–8

Turner GG (1919) Local discolouration of the abdominal wall as a sign of acute pancreatitis. *Br J Surg* **7**(27): 394–5

Figure 1. Computed tomography angiogram showing the arteriovenous fistula (black arrow) arising from the left external iliac artery (white arrow).



Case Report

A 26 year-old male of Pakistani origin with Ehlers–Danlos syndrome presented to the authors' hospital complaining of sudden onset left lower abdominal pain and constipation. He had no preceding history of trauma. His past medical history included spinal surgery as a child to correct his kyphoscoliosis.

On examination, the patient appeared uncomfortable, was haemodynamically stable and afebrile. Classical features of Ehlers–Danlos syndrome were noted, including hyperelastic, translucent skin, hypermobile joints, and short stature with kyphoscoliosis. Tenderness in the left lower quadrant was elicited. Full blood count revealed leukocytosis (17.7 x 10^9cells/litre). All other blood tests were normal. With the diagnosis remaining unclear the patient was admitted for observation.

On day three of admission, the haemoglobin level dropped to 8.8 g/dl from 11.9 g/dl. Examination revealed unilateral Grey–Turner's sign, i.e. flank bruising suggestive of retroperitoneal bleeding (Turner, 1919), over which a bruit was heard. A diagnosis of acute haemorrhage from an underlying vascular malformation was suspected and confirmed by computed tomography (CT) angiogram. The CT demonstrated a haematoma in the left pelvis originating from a large arteriovenous fistula between the left external iliac artery and vein (*Figure 1*). The patient was treated by percutaneous transluminal stent insertion into the fistula. He remains well 18 months later.