Main differences between true and false ventricular aneurysms	
<i>Pseudoaneurysm</i> rare myocardium ruptured narrow base no myocardial elements in wall high risk of rupture surgical resection essential	<i>True aneurysm</i> common myocardium locally stretched wide base (most, but not all) myocytes +/- coronary arteries in wall low risk of rupture surgery indicated if resistant symptoms

Box 3

formed without delay since these aneurysms, unlike true ventricular aneurysms, have a propensity to rupture regardless of size or symptoms.^{7,8}

Conclusions

VPA formation is an ominous complication of myocardial infarction. If diagnosed, surgical resection, with coronary artery bypass grafting

if indicated, should be performed without delay. If left untreated, a VPA is likely to rupture with an invariably fatal outcome.

Final diagnosis

Left ventricular pseudoaneurysm.

Keywords: ventricular pseudoaneurysm, myocardial infarction

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Spontaneous aortic rupture in a 22-year-old

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A previously healthy 22-year-old Caucasian man was admitted following a sudden onset of an acute abdominal and lower back pain, collapse and hypotension and was rushed to theatre with suspected acute haemorrhage of unknown origin. There was no history of any trauma, past or present and the patient was not engaged in any strenuous activity prior to experiencing the above episode. Urgent exploratory laparotomy revealed rupture of the anterior aspect of the infra-renal abdominal aorta. During repair it was found that all the vessels, including the abdominal aorta, were of very small calibre and were very friable. This caused immense problems during the anastomosis of the graft and also led to damage to the inferior vena cava during the initial dissection. Successful graft anastomosis and satisfactory haemostasis was eventually achieved and mass closure of the abdomen was carried out.

Per- and post-operatively the patient received 40 units of blood, 18 units of fresh frozen plasma and seven units of platelets. The patient developed disseminated intravascular coagulopathy postoperatively and died within 38 hours of surgery.

Post-mortem examination raised the suspicion of some 'connective tissue disorder' because the aortic wall and skin were found to be very fragile during dissection. Specimens from the aorta were sent for histopathological examinations and the opinion of a geneticist. The patient's family received genetic counselling to rule out the prevalence of disease in other close family members.

Question

What is the most likely diagnosis?

Answer

Ehlers-Danlos syndrome type IV

Discussion

In our case, the family provided certain clues which pointed to the diagnosis of the Ehlers-Danlos syndrome type IV. The patient had a tendency to bruise easily from the childhood and on one occasion developed spontaneous bruising under his chin several inches in diameter. He had a thin, wiry, build and had problems with flat feet. There was evidence of tissue paper scarring on his shins. He had a thin nose, prominent eyes and translucent skin. Examinations of histopathological specimens from the aorta of this patient confirmed the diagnosis of type IV Ehlers-Danlos syndrome. A genetic opinion was also sought and agreed with this diagnosis.

Investigation of his brother and parents failed to reveal any history suggestive of Ehlers-Danlos syndrome type IV. Both parents have declined skin biopsy for diagnostic analysis. It is entirely possible that, in our patient, a defect in the COL3A1 gene arose as a result of a new mutation. However, this is a large gene and since the amount of DNA recovered from the tissues at *post-mortem* was small, sequencing of this mutation may be impossible. Alternatively, the disorder could have been inherited in an autosomal recessive fashion.

AETIOLOGY

The Ehlers-Danlos syndromes are a heterogeneous group of connective tissue disorders (box 1). Type IV, the so-called ‘ecchymotic or acrogeric’ form of Ehlers-Danlos syndrome usually follows an autosomal dominant pattern of inheritance although rare recessive cases have been reported.¹ It is caused by mutation in type III collagen gene, COL3A1² leading to abnormal type III procollagen and diminished

levels of type III collagen. The mutations vary from family to family. As the major component of blood vessel walls are elastin and collagen type I and III, Ehlers-Danlos syndrome type IV is associated with spontaneous rupture of the major or medium sized arteries with catastrophic results.

CLINICAL FEATURES

Ehlers-Danlos syndrome may present in childhood with easy inappropriate bruising, poor wound healing and joint hyperextensibility. There are characteristic facies with prominent eyes and a thin ‘pinched’ nose.³ Patients may not be aware of their diagnosis, however, until a catastrophic, spontaneous rupture of an artery or the bowel occurs⁴ (box 2).

INVASIVE PROCEDURES/SURGERY

Invasive procedures like angiograms and arterial surgery carry a very high mortality.^{5,6} In most cases it is not possible to repair the arterial defect either with graft or suture due to the extreme fragility of the arterial wall and conservative management with bed rest and close monitoring of the patient is a safer option in certain bleeding episodes. All elective surgical procedures should be performed by

Ehlers-Danlos syndrome type IV: clinical presentation

- low birth weight
- premature birth
- failure to thrive
- short stature
- congenital dislocation of the hips
- easy inappropriate bruising
- diagnostic facial features, ie, large eyes, pinched nose and thin lips
- hypermobility of the joints
- thin atrophic skin with a visible venous/capillary pattern or telangiectasia
- spontaneous arterial perforation
- spontaneous perforation of the colon

Ehlers–Danlos syndrome				
Type	Name	Inheritance	Molecular defect	Major complications
I	Gravis	Dominant	Unkown	Musculoskeletal deformities; premature rupture of fetal membranes
II	Mitis	Dominant	Unknown	Less severe than type I
III	Benign hypermobile	Dominant	Unknown	Arthritis
IV	Arterial-ecchymotic	Dominant/recessive	Type III collagen deficiency	Arterial rupture, aortic aneurysm, dissection, rupture, intestinal perforation
V	X-linked	X-linked	Lysyl oxidase defect	Musculoskeletal disorders
VI	Ocular	Recessive	Lysyl hydroxylase defect	Cornea and sclera fragility; musculoskeletal disorders
VII	Arthrochhalasis multi-plex congenita	Recessive	Deficiency of pro-collagen peptidase	Short stature; multiple joint dislocations
VIII	Peridontitis	Dominant	Unknown	Advanced generalized periodontitis
IX	Skeletal and urinary dysplasia	–	Lysyl oxidase defect	Skeletal and urinary tract abnormalities

Modified from ⁶

Box 1

Learning points

- Ehlers-Danlos syndrome type IV is caused by deficiency of type III collagen.
- spontaneous arterial and bowel rupture in a young individual should prompt urgent investigations and genetic assessment of the patient and close family members.
- trauma, pregnancy, invasive procedures and elective surgery are associated with very high morbidity and mortality.
- prenatal diagnosis and prevention of the disease is now possible

an experienced surgeon who is aware of the complications of the condition. This applies especially to varicose vein ED Grech surgery⁷ which is frequently carried out in these patients. One of the authors has personal experience of a patient with Ehlers-Danlos syndrome type IV who eventually required above knee amputation following an elective varicose vein surgery.

PROGNOSIS

With autosomal dominant inheritance there is a 50% risk of transmission to the offspring of

affected individuals. Genetic counselling and examination of other family members is therefore important. Females also have a high mortality risk, up to 25% during pregnancy.⁸ Affected individuals are recommended to modify their life style to avoid physically strenuous jobs and sports which involve body contact to lessen the chances of traumatic vascular and bowel catastrophes.

In conclusion we emphasize that spontaneous rupture of the infrarenal abdominal aorta in a young healthy, adult male should prompt urgent and thorough clinical survey, laboratory investigations and genetic counselling of the patient and close relatives to rule out Ehlers-Danlos syndrome type IV. This rare disorder should now be considered in the differential diagnosis of the haemoperitoneum in a young individual with or without previous history of connective tissue disorder.

Final diagnosis

Spontaneous rupture of an infrarenal abdominal aorta in Ehlers-Danlos syndrome type IV

Keywords: Ehlers-Danlos syndrome type IV, infrarenal aorta

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The Royal College of Surgeons

The morning session will consist of a series of 30 minute lectures on the following topics:

- the effects of boxing on the brain. New research has shown advanced brain-damage in a professional boxer aged 23
- the 'Piltown Man'. A fraudulent discovery of the 'missing link' which lasted 40 years
- forensic odontology – both victims and criminals may be identified by their teeth – even if they are false!
- an overview of pathology museums – their use for teaching and some of the unique specimens they contain

The afternoon session will be dedicated to the splendid collections housed at the Royal College of Surgeons which contain a wealth of material. The famous Hunterian Museum contains an unusual mixture of both human and animal anatomy and pathology dating back to the 18th century. Human anatomy is also represented in a separate dedicated museum as is pathology and dentistry. The College also features a fine collection of surgical instruments

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