disturbances in calcium metabolism nor do we have proof of impaired mucopolysaccharide metabolism to provide some aetiological clues for the basal ganglia calcification. Thus, we must await a more definitive explanation before drawing any conclusions regarding the pathogenesis of basal ganglia calcification in Down’s syndrome.


Final diagnosis

Bilateral basal ganglia calcification in Down’s syndrome.

Keywords: Down’s syndrome; calcification; basal ganglia

A man with purple toes

Syed Zaman, Jagdish Mallya, Matthew Thomas

A 79-year-old man was admitted for urgent angiography because of pain and discolouration of his toes. Six months prior to admission he was commenced on amiodarone and warfarin for paroxysmal atrial fibrillation. He was known to have hypercholesterolaemia, ischaemic heart disease and congestive cardiac failure. He was an ex-smoker. His other medications were enalapril, frusemide and cholestyramine.

Examination revealed purple discoloration of his toes with evidence of digital infarction (figures 1 and 2), blood pressure was 140/80 mmHg and no murmurs or bruits were audible. Lower limb pulses were all palpable and Doppler pressure studies suggested small vessel occlusion without significant large vessel disease.

Investigations revealed: haemoglobin 12.2 g/dl, platelets $337 \times 10^9$/l, erythrocyte sedimentation rate (ESR) 36 mm/h, white blood cells $6.9 \times 10^9$/l (neutrophils 4.5, lymphocytes 1.5, monocytes 0.5, eosinophils 0.1, basophils 0.1). Na 141 mmol/l, K 4.7 mmol/l, urea 12.1 mmol/l, creatinine 169 mmol/l, cholesterol 3.6 mmol/l, auto-immune profile and ANCA negative. Electrocardiogram showed sinus rhythm, left ventricular hypertrophy, anteroseptal Q waves. Echocardiogram showed impaired left ventricular function. Abdominal ultrasound showed a 4-cm abdominal aortic aneurysm.
Answers

Question 1
Cholesterol crystal embolism (CCE). The risk factors for CCE are listed in box 1.

Risk factors for CCE
- atherosclerotic cardiovascular disease
- hypertension
- angiography/angioplasty
- anticoagulant/thrombolytic therapy
- vascular surgery
- male sex
- age greater than 50 years
- smoking history
- hypercholesterolaemia
- diabetes mellitus
- cardiopulmonary resuscitation

Box 1

The presence of pedal pulses with digital infarction suggests small vessel disease. CCE is a well-recognised but probably under-diagnosed disorder where cholesterol crystals embolise from atherosclerotic plaques, occluding arterioles with an external diameter ranging from 55 to 900 μm, thus causing irreversible organ dysfunction.

The reported incidence of CCE is variable; in a recent study of the Dutch population CCE was reported with an average frequency of 6.2 cases per million population per year.1 Mayo and Swartz, in their review of in-patient nephrology consultations, suggest an incidence of at least one case every two weeks.2 Both papers suggest that the condition is probably under-diagnosed.

The typical patient with CCE is elderly, male and has a history of hypertension and atherosclerotic disease. The more risk factors present, the more likely the diagnosis of CCE and the presence of four or more risk factors should be taken as presumptive evidence of CCE.2

The condition can occur spontaneously but often there is a history of mechanical disruption of thrombus overlying atheroma, eg, following angiography, angioplasty or vascular surgery, thus exposing the plaque to the general circulation. Thrombolysis and anticoagulation, by destabilising atheromatous plaques, are recognised precipitants and with the increasing use of these drugs clinicians should exercise vigilance for the development of CCE.

Question 2
Many organs can be affected (box 2) thus the clinical features (box 3) depend on the site(s) of embolisation. The commonest clinical presentations are cutaneous features, renal failure (50% of cases) and accelerated hypertension. Nonspecific presentation can occur with fever, myalgia or weight loss and can be confused with other systemic disorders such as infective endocarditis and vasculitis.

Cutaneous features occur in up to 35% of patients and tend to be bilateral. Our patient presented with ‘purple toes’, a complication particularly associated with warfarin therapy.4 The only opportunity to visualise cholesterol emboli is on fundoscopy (Hollenhorst plaques); their presence is an important marker for the development of cerebral infarction.4

Laboratory abnormalities associated with CCE are listed in box 4. Eosinophilia occurs often, 39–100% in some series.4

Organ involvement in CCE (% occurrence) 4
- kidney (73)
- spleen (42)
- bowel (28)
- pancreas (20)
- skin/muscle (19)
- adrenal (12)
- heart (12)
- brain (12)
- liver (8)

Box 2

Clinical features of CCE
- fever
- myalgia
- weight loss
- lower extremity pain
- gastrointestinal bleeding
- cutaneous manifestations: livedo reticularis, purpura, cyanosis, ‘purple toes’, cutaneous necrosis
- accelerated hypertension
- Hollenhorst plaques
- vascular bruits
- encephalopathy/altered mental state
- renal impairment
- pancreatitis

Box 3

Laboratory features of CCE
- ESR greater than 30 mm/h
- eosinophilia/eosinophiluria
- thrombocytopenia
- raised amylase
- elevated creatine kinase
- abnormal liver function tests
- elevated serum urea and creatinine
- hypocomplementaemia

Box 4
QUESTION 3
If at all possible, the diagnosis should be confirmed histologically by tissue biopsy of skin or kidney to reveal the pathognomonic needle-shaped clefs in arterioles. Once established, invasive procedures known to precipitate CCE should be avoided. Anticoagulation and thrombolysis are also contraindicated. Associated risk factors such as hypertension, hypercholesterolaemia and smoking should be managed aggressively. Renal replacement therapy may be required.

Once "pre-mortem" diagnosis is made (less than 40% of cases) prognosis is poor. Mortality is high, with death most commonly due to ischaemic heart disease, cerebrovascular disease and renal failure, while 10.8% of patients die of the multiple cholesterol emboli syndrome. 1

Heightened awareness of CCE will lead to early diagnosis, thus avoiding precipitating factors. Unnecessary immunosuppressant therapy will also be avoided since CCE can mimic the clinical presentation of systemic vasculitis. 2

Our patient did not undergo angiography, nor tissue biopsy and his condition deteriorated rapidly despite stopping warfarin. There was progressive renal failure and a step-wise decline in cognitive function secondary to vascular dementia (brain computed tomography scan revealed multiple small ischaemic areas). He died four months after presentation. The presence of more than four risk factors together with the clinical features makes the diagnosis of CCE very likely.

Final diagnosis

Cholesterol crystal embolism.

Keywords: cholesterol crystal embolism; purple toes; renal failure; anticoagulation

Blunt abdominal trauma

D S Bhandarkar, T R Raju, L R Jenkinson

A 25-year-old man was brought to the casualty department complaining of severe upper abdominal pain after being crushed between a tractor and a wall. His pulse was 100 beats/min and blood pressure 150/80 mmHg. Abdominal examination revealed a 10 cm by 5 cm, oval, tender epigastric swelling with bruising of the overlying skin. The remainder of the abdomen was soft and non-tender. Chest and abdominal X-rays, blood investigations and urinalysis were normal. A computed tomography (CT) scan was ordered to ascertain the nature of the epigastric swelling (figure).

Questions

1 What is the diagnosis?
2 What is the differential diagnosis of an abdominal wall swelling appearing after blunt trauma?

A man with purple toes.

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