optic atrophy on fundoscopy, ataxia and hypotonia. Blood tests showed significant derangement of liver function and raised lactate levels. What is the most likely diagnosis?

(a) Alpers-Huttenlocher syndrome
(b) Barth syndrome
(c) Lipofuscinosis
(d) MELAS
(e) Rasmussen's encephalitis

5 A 45-year-old woman presented with depression and was treated with amitriptyline by her general practitioner (GP). Three days later the GP was called to see her because she had become unrousable. On examination in hospital her Glasgow Coma Score was 5, respiratory rate 20 bpm and oxygen saturation 95% breathing air. Electrocardiogram (ECG) and chest X-ray were normal. Toxicology screening was negative. Which of the following statements is correct?

(a) A blood glucose of 3.2 mmol/l would explain her symptoms
(b) The hyperventilation is likely to be due to a metabolic acidosis
(c) Hyperammonaemia is diagnostic of a urea cycle disorder
(d) A plasma ammonia of 204 mol/l should be treated
(e) A normal brain computed tomography scan means she is not encephalopathic

6 A 24-year-old man presented to accident and emergency (A&E) with a history of muscle pain and anuria. Three days previously he had been cleaning out a drain. On pulling himself out of the manhole he was working in, he developed acute muscle pain. Over the next 48 hours he noticed dark urine and had now not passed any urine for the past 24 hours. Investigation revealed a creatine kinase (CK) of over 200,000 IU/l and a creatinine of 720 mol/l. Which of the following statements is correct?

(a) Haemodialysis should be commenced immediately
(b) Diagnosis will require an acute phase plasma sample
(c) Renal function is likely to return to normal
(d) Diagnosis will require a muscle biopsy
(e) He may have a lysosomal storage disorder

7 A 43-year-old man presented to A&E with a history of syncope. There was a family history of heart failure in two maternal uncles, and his sister and mother had both required a permanent pacemaker in their mid-40s. His maternal aunt developed heart failure in middle age but had not required pacing. Investigations included a resting ECG which showed 2:1 atrioventricular block. Full blood count, CK, urea and electrolytes, liver function tests and cholesterol were normal. Echocardiography revealed mild left ventricular dilatation and an ejection fraction of 50%. What is the most likely inherited disorder in this family?

(a) Hypertrophic cardiomyopathy
(b) Lamin A/C cardiomyopathy
(c) Lev-Lenegre disease
(d) Ischaemic heart disease
(e) Dystrophin cardiomyopathy

8 A 14-year-old boy presented with a history of extra beats and a single episode of syncope following a cross-country run. He had otherwise been well in the past. His paternal uncle had died of 'heart failure' at the age of 50 and his 20-year-old sister had a history of syncope and documented ventricular tachycardia (VT) requiring an implantable cardioverter defibrillator. His resting ECG shows sinus rhythm with a single LBBB ectopic beat positive in leads II,II and aVf with T wave inversion in leads V1–V3. What is the most likely cause of his symptoms?

(a) Idiopathic dilated cardiomyopathy
(b) Arrhythmogenic right ventricular cardiomyopathy
(c) Catecholaminergic polymorphic VT
(d) Hypertrophic cardiomyopathy (HOCM)
(e) Brugada syndrome

9 A 55-year-old man presented with acute left ventricular failure and high blood pressure. On examination he had the cutaneous features of neurofibromatosis 1 (NF1). If his acute problems are NF1 related, what is the most likely underlying cause?

(a) HOCM
(b) Renal artery stenosis
(c) Phaeochromocytoma
(d) Glomus tumour
(e) Gastrointestinal stromal tumour

10 A 20-year-old woman presented with walking difficulty and a history of tinnitus and difficulty in hearing in crowded rooms. Her father had died from NF. What kind of NF should be considered?

(a) NF1
(b) NF2
(c) Segmental NF1
(d) Spinal NF
(e) Watson syndrome

CME Infectious diseases SAQs

Answers to the CME SAQs published in *Clinical Medicine* October 2008

Q1	Q2	Q3	Q4	Q5	Q6	Q7	Q8	Q9	Q10
(d)	(e)	(b)	(d)	(a)	(c)	(c)	(d)	(a)	(e)