# Genetics (36226)

## Self-assessment questionnaire

SAQs and answers are ONLINE for RCP Fellows and Collegiate Members

**The SAQs printed in the CME section can only be answered online to achieve external CPD credits. The closing date is 21 January 2009 (midnight GMT).**

**We recommend that answers are submitted early so that any problems can be resolved before the deadline.**

### Format

SAQs follow a best of five format in line with the MRCP(UK) Part 1 exam. Candidates are asked to choose the best answer from five possible answers. Any comments should be sent in via email only: clinicalmedicine@rcplondon.ac.uk

### The answering process

1. To access the questions, log on to the Fellows and Members area www.rcplondon.ac.uk/Members Please contact the Information Centre if you have lost or forgotten your username or password: infocentre@rcplondon.ac.uk
2. Select: **Self assessment**
3. At the top of the SAQ page select the current CME question paper
4. Answer all 10 questions in any order, by selecting the best answer
5. Click on **Submit for final marking**.

**After submitting your answers NO changes can be made.**

### The marking process

- You must submit the answers before the closing date shown at the top of the screen
- Answers will be marked automatically on the date displayed for that paper
- You can find your marks on the CME page under **My past CME papers**.

### Registering your external CPD credits

A pass mark of 80% allows you to claim two external CPD credits. Only the first seven distance-learning credits will be counted as external; the remainder can be claimed as personal credits. Credits can be recorded using the online diary system. All *Clinical Medicine* SAQs are listed under **External Approved CPD.**

**1 A 40-year-old Caucasian man presented with a three-year history of progressive drooping of both eyelids and decreased exercise tolerance. A history of other similarly affected family members was confirmed by looking at old photographs. Examination revealed bilateral ptosis and symmetrical limitation of eye movements. A Tensilon® test was negative and electromyography normal. He went on to have a muscle biopsy. What is the most likely histological finding in the muscle biopsy?**

(a) Prominent neurogenic features (muscle fibre type grouping)
(b) Eosinophilic inclusions
(c) Diffuse fibrosis
(d) Perivascular inflammatory infiltrates
(e) Mosaic defect of cytochrome C oxidase

**2 A 36-year-old Caucasian woman presented acutely with left-sided weakness, on the background of a one-year history of recurrent severe headaches and recent-onset generalised seizures. Her past history included bilateral deafness at the age of nine and diabetes at 16. Her early childhood developmental milestones had been normal. The family history revealed that a maternal aunt and uncle died at a young age with unexplained cardiomyopathy. What is the most likely diagnosis?**

(a) Kearns–Sayre syndrome
(b) Leigh syndrome
(c) Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes (MELAS)
(d) Myoclonic epilepsy with ragged red fibres
(e) Pearson syndrome

**3 A 30-year-old Caucasian man presented to his local eye unit with a four-week history of progressive visual blurring in his left eye and, since waking that morning, had noticed the same problem with his right eye. There was a family history of two maternal uncles registered blind in their early thirties. On examination, his visual acuities were reduced to counting fingers, there were dense central scotomas, but the optic discs were entirely normal. Extensive neurological investigations, including a lumbar puncture and a magnetic resonance imaging brain scan, failed to identify an underlying cause. Three months after he first presented, his visual function had not improved. What is the most likely diagnosis for his bilateral optic neuropathy?**

(a) Demyelinating retrobulbar neuritis
(b) Dominant optic atrophy
(c) Leber hereditary optic neuropathy
(d) Neuromyelitis optica (Devic's disease)
(e) Toxic optic neuropathy

**4 A three-year-old Caucasian girl presented with a three-month history of intractable complex partial seizures and jaundice, on a background of severe developmental delay. The family history revealed that a cousin had died aged six months from unexplained 'cot death'. Examination revealed**

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) |