

CME Genetics SAQs (89268): Self-assessment questionnaire

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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. Any comments should be sent in via email only: clinicalmedicine@rcplondon.ac.uk

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.

The answering process

- 1 Go to www.rcplondon.ac.uk/SAQ
- 2 Log on using your usual RCP username and password
- 3 Select the relevant CME question paper
- 4 Answer all 10 questions by selecting the best answer from the options provided
- 5 Once you have answered all the questions, click on Submit

Registering your external CPD credits

Carrying out this activity allows you to claim two external CPD credits. These will be automatically transferred to your CPD diary, where you can review the activity and claim your points.

- 1 A 45-year-old woman presents to the endocrine department with a 6-month history of episodic anxiety, sweating and palpitations. She has also been complaining of intermittent right flank pain for the last few weeks. Her father died of clear cell renal carcinoma at the age of 42 years.

Investigations:

blood pressure 85/105 mmHg
 urinary metanephrines 4.21 $\mu\text{mol}/24\text{ h}$ (0.00–1.20)
 MIBG scan 2.6 cm right tracer-avid adrenal mass

ultrasound scan of abdomen likely cyst in head of pancreas measuring 46 mm, with three small simple cysts measuring 7 mm, 8 mm and 6.5 mm in tail of pancreas. Normal size and appearance of kidneys, small 4 mm mid/upper pole cortical cyst on the right side, no hydronephrosis or obvious calculi seen

What is the most likely diagnosis?

- (a) sporadic pheochromocytoma
 - (b) multiple endocrine neoplasia type 2
 - (c) von Hippel–Lindau syndrome
 - (d) *SHDB*-associated pheochromocytoma–paraganglioma syndrome
 - (e) renal cell carcinoma with adrenal metastases
- 2 A 37-year-old man presents to the endocrine department with a lump in the neck and episodes of diarrhoea and flushing. Past medical history includes excision of a left-sided pheochromocytoma aged 35 years.

Investigations:

calcitonin level 20 ng/l (<12 ng/l)

What other hormone level may be elevated in this condition?

- (a) prolactin
 - (b) gastrin
 - (c) parathyroid hormone
 - (d) cortisol
 - (e) oestrogen
- 3 A 56-year-old woman presents to the oncology department with a 2-month history of abdominal swelling, weight loss and change in bowel habit. Investigations reveal a large ovarian mass which is surgically excised. Histology identifies a serous adenocarcinoma of the ovary. There is no family history of breast or ovarian cancer.
- #### What is the approximate likelihood of identifying a mutation in the high-risk breast–ovarian cancer genes *BRCA1* and *BRCA2* in her?

- (a) <1%
- (b) 1–5%
- (c) 5–10%
- (d) 10–20%
- (e) >20%

- 4 A 35-year-old man was referred to the outpatient clinic with a 3-month history of change in bowel habit. Since the onset of symptoms, he had lost 2 kg in weight. In the last week he had noticed blood in his stools. His mother had died at the age of 43 years from mucinous ovarian cancer, and his maternal aunt has just been diagnosed with endometrial cancer at the age of 63 years.

Investigations:

colonoscopy showed a mass arising from the distal sigmoid colon

An abnormality in which gene is most likely to be responsible for his condition?

- (a) *MLH1*
- (b) *BRCA1*
- (c) *APC*
- (d) *TP53*
- (e) *MEN1*

- 5 A 21-year-old female student was found to have incidental glycosuria following a urine dipstick for a urinary tract infection. An oral glucose tolerance test was undertaken. Fasting plasma glucose level was 5.7 mmol/l. Two-hour plasma glucose concentration was 10.6 mmol/l. On questioning, she mentioned that her 25-year-old brother, father and paternal grandfather are diabetic. Genetic testing identified a mutation in the *HNF1A* gene. What would be the most appropriate initial management of her diabetes?

- (a) diet
- (b) insulin
- (c) metformin
- (d) a sulfonylurea
- (e) an alpha-glucosidase inhibitor

- 6 A 17-year-old male presented with a 2-month history of progressive polyuria and polydipsia. Urine dipstick revealed glycosuria and haematuria. Fasting plasma glucose level was 8.9 mmol/l. A glucose tolerance test (OGTT) showed a 2-hour plasma glucose level of 12.3 mmol/l.

Na	141 mmol/l
Albumin	38 g/l
K	3.7 mmol/l
alkaline phosphatase (ALP)	130 U/l
urea	10 mmol/l
alanine transaminase (ALT)	89 IU/l
creatinine	145 µmol/l
Gamma-glutamyl transpeptidase (γ-GT)	73 U/l
bilirubin	17 µmol/l
ultrasound scan of abdomen	multiple renal cysts

What is the most likely inheritance pattern of this patient's diabetes?

- (a) multifactorial
 - (b) mitochondrial
 - (c) autosomal dominant
 - (d) autosomal recessive
 - (e) X-linked recessive
- 7 A 31-year-old woman presented at 20 weeks of pregnancy after the detection of multiple cardiac lesions consistent with cardiac rhabdomyomas on the fetal anomaly scan. On examination she had multiple facial angiofibromas in the nasolabial area and four irregular hypopigmented patches over her trunk and abdomen.

What is the leading cause of early death in this condition?

- (a) central nervous system tumours, eg giant cell astrocytomas/cortical tubers/subependymal nodules
- (b) renal cell carcinoma
- (c) angiomyolipoma
- (d) peripheral nerve sheath tumours
- (e) lymphangioleiomyomatosis

- 8 A 19-year-old woman presented with a 6-month history of exertional dyspnoea. Computed tomography (CT) scan of the thorax revealed a coarse reticulonodular pattern in the lung bases, with pulmonary cysts characteristic of lymphangioleiomyomatosis. Renal ultrasound revealed angiomyolipomas and genetic testing confirmed a mutation in the *TSC1* gene. She opted for enrolment in a clinical trial looking at sirolimus treatment in patients with tuberous sclerosis.

Which of the following is NOT a recognised feature of treatment with sirolimus?

- (a) interaction with grapefruit juice
- (b) hyperlipidaemia
- (c) thrombocytopenia
- (d) interstitial pneumonia
- (e) interaction with oral contraceptives

- 9 A 24-year-old man presented to the chronic pain department with chronic joint pain in the small joints of the hand, shoulders, hips and knees. He gave a history of multiple joint dislocations from childhood, easy bruising and hypermobility. Past medical history included a diagnosis of irritable bowel syndrome. On examination his Beighton score was 6/9 (see www.nhs.uk/Conditions/Joint-hypermobility/Pages/Diagnosis.aspx), with passive dorsiflexion of both fifth fingers beyond 90°, passive apposition of each thumb to the flexor surface of the forearm and hyperextension of each elbow beyond 10°. He had soft skin with normal extensibility and fragility.

How would you confirm the diagnosis?

- (a) skin biopsy with electron microscopy of collagen fibres
- (b) genetic testing of *COL5A1*
- (c) genetic testing of *COL5A2*
- (d) clinical examination
- (e) genetic testing of *COL3A1*

- 10 A 36-year-old man presented with an inguinal hernia. He gave a history of poor wound healing. On examination he had extensive skin hyperextensibility, with multiple atrophic scars. He had joint hypermobility, with a Beighton score of 5/9 (see www.nhs.uk/Conditions/Joint-hypermobility/Pages/Diagnosis.aspx).

Which of the following would you NOT expect to see on examination/investigation?

- (a) 'cauliflower' deformity of collagen fibrils on electron microscopy
- (b) mutation in the *COL5A1* gene
- (c) mitral valve prolapse on echocardiogram
- (d) abnormalities of type III procollagen on biochemical testing
- (e) multiple bruises

CME Palliative medicine SAQs

Answers to the CME SAQs published in *Clinical Medicine* June 2014

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