QI&CPD
Accredited Activity
CATEGORY 2

Each of the questions or incomplete statements below is followed by four or five suggested answers or completions. Select the most appropriate statement as your answer.

Clinical challenge

Case 1

Lucas, aged 36 years, presents to you as a new patient. He was diagnosed with Fabry disease at the age of 13 years and is very well informed regarding his condition. He has recently moved to the area and is seeking a new general practitioner (GP).

Question 1

With regards to the epidemiology of rare diseases, which one of the following statements is CORRECT?

A. A rare disease is defined as a condition with a prevalence of less than one per 3000.
B. Approximately 75% of rare conditions have a prevalence of 0.1–10 per 100,000.
C. 20% of rare diseases have a genetic basis.
D. A delayed diagnosis is reported in 50–60% of cases.

Question 2

Lucas's condition went undiagnosed for several years. All of the following statements regarding the importance of diagnosis are correct EXCEPT:

A. A diagnosis informs prognosis and management.
B. A diagnosis can restore reproductive confidence.

C. A diagnosis validates an individual's symptomatology.
D. A diagnosis should be made during the initial consultation.

Question 3

Lucas's condition was eventually diagnosed after his GP recognised the link between his recurrent presentations of joint pain and abdominal pain.

Which one of the following conditions is most commonly associated with the combination of joint subluxation and joint pain?

A. Birt-Hogg-Dubé syndrome
B. Neurofibromatosis
C. Ehlers-Danlos syndrome
D. Lipedema

Question 4

Your consultation with Lucas causes you to reflect on the challenges of managing rare diseases at a population level.

In Australia, current strategies to address the challenge of managing rare diseases include:

A. the *Undiagnosed Diseases* telemedicine program
B. the Agrenska centre for care of multiple rare disease groups
C. multidisciplinary clinics for specific rare disorders

D. a coordinated national plan for rare diseases.

Question 5

Key components of a national plan for addressing rare diseases include which of the following?

A. Raising awareness of the burden of rare diseases
B. Improving healthcare for people with rare diseases
C. Promoting scientific and social research on rare diseases
D. Providing educational resources for health professionals
E. All of the above

Case 2

Doris, aged 79 years, presents with a 3-month history of weight loss and poor appetite. You are her regular GP in a small country town. She has previously been well and is the full-time carer of her husband. You suspect she may have an underlying malignancy.

Question 6

Which of the following is the strongest risk factor for cancer of unknown primary?

A. History of heavy smoking
B. Being from a remote area

C. Being male

D. Increasing age

E. History of high alcohol consumption

Question 7

You arrange investigations, which confirm a metastatic cancer on histopathology. However, appropriate specialised investigations fail to identify the site of origin.

Which one of the following is the correct diagnosis for Doris?

A. Malignancy of unknown origin

B. Confirmed cancer of unknown primary

C. Provisional cancer of unknown origin

D. Metastatic colorectal carcinoma

Question 8

Doris wants to know why it is important to try to discover the origin of a cancer. You explain to her that cancers have varying prognoses and treatments.

Which one of the following subtypes of cancer of unknown primary has the poorest prognosis?

A. Merkel cell adenopathy of unknown origin

B. Poorly differentiated neuroendocrine carcinoma

C. Squamous cell carcinoma involving cervical lymph nodes

D. Females with papillary adenocarcinoma of peritoneal cavity

E. Squamous cell carcinoma of the adominopelvic cavity

Question 9

Which one of the following subtypes of cancer of unknown primary has a favourable prognosis?

A. Multiple metastatic lytic bone disease (non-prostate-specific antigen)

B. Poorly differentiated carcinoma with midline distribution (extragonadal germ cell syndrome)

C. Multiple cerebral metastases

D. Adenocarcinoma metastatic to the liver or other organs

E. Squamous abdominopelvic cancer of unknown primary

Case 3

Jessica, 28 years of age, presents to you for the first time, requesting blood tests to diagnose Lyme disease. She describes a 6-month history of fatigue and intermittent joint aches. She has done some research online and has decided her symptoms must be due to Lyme disease. You feel that this is very unlikely and you attempt to explore other possible diagnoses. However, she becomes frustrated, stating that she is sure that it is Lyme disease.

Question 10

What is the most accurate description of Jessica's presentation?

A. Malingering

B. Chaotic illness

C. Contested illness

D. Hypochondriasis

E. Enigmatic illness

Question 11

The following week, you see Jessica's 24-year-old sister, Mallory, as a patient. Mallory tells you she has been tired and emotional for many years now. She also has ongoing issues with abdominal pain, which has been extensively investigated previously with no cause found. These symptoms are creating issues in her workplace. She discloses that she was sexually abused by an uncle as a child. You decide that Mallory's presentation is consistent with a chaotic illness and that validation is an important part of therapy.

Which of the following approaches can provide validation as part of managing a chaotic illness?

A. Acknowledge that the patient has survived a difficult life.

B. Avoid discussing difficult issues such as childhood trauma.

C. Advise that childhood trauma should not affect the central nervous system.

D. Explain that you can help her find a simple solution.

E. Advise that life should not be overwhelming.

Question 12

You continue to see Mallory regularly over the next year. Given that managing chaotic illness can be challenging for the doctor and patient, you aim to include harm minimisation in your management.

Which of the following is consistent with harm minimisation in the context of a chaotic illness?

A. Actively manage preventive health.

B. Attend to physical and psychological issues separately.

C. Be prepared to acknowledge when a consultation does not go well.

D. Protect Mallory as much as you can from health professionals who are judgemental.

E. All of the above are correct.