ABCDE clinical challenge

Questions for this month's clinical challenge are based on articles in this issue. The clinical challenge is endorsed by the RACGP Quality Improvement and Continuing Professional Development (QI&CPD) program and has been allocated 4 Category 2 points (Activity ID: 7651). Answers to this clinical challenge are available immediately following successful completion online at http://gplearning.racgp.org.au. Clinical challenge quizzes may be completed at any time throughout the 2014–16 triennium; therefore, the previous months answers are not published.

Single completion items

CPD

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

Case 1
Aaron Blieck

Aaron Blieck, 44 years of age, is a new patient at your clinic presenting for a general check-up. He also wants to know if he should have genetic testing, as several of his relatives have developed medical conditions over the past few years.

Question 1

Which of the following features of Aaron's family history suggest a potentially heritable condition?

A. His mother developed heart disease at age 64 years.
B. His sister developed breast cancer at age 54 years.
C. His paternal uncle and grandmother had bowel cancer.
D. His father had prostate cancer at age 62 years.
E. His cousin had melanoma at age 45 years.

Question 2

Given his family history, Aaron is at risk of a heritable condition. Aaron is interested in genetic testing. Which of the following should you discuss with him before referring for testing?

A. If he is found to carry the abnormal gene, this means he will inevitably develop the condition.
B. If he is found to carry the abnormal gene, he must disclose this when applying for private health insurance.
C. If he is found to carry the abnormal gene, he has a legal duty to inform his relatives.
D. If he is found to carry the abnormal gene, he must disclose this to his life insurer when updating an existing policy.
E. If he is found to carry the abnormal gene, his children must undergo genetic testing.

Question 3

Aaron wants to know how much it will cost him to have the genetic testing done. Which of the following tests is NOT available on the Medicare Benefits Schedule when ordered by a GP for a patient who meets appropriate clinical indications?

A. BRCA1
B. Haemochromatosis (HFE)
C. Fragile X syndrome
D. Factor V Leiden
E. Karyotyping

Question 4

Aaron asks about 'getting all of his genes tested'. Which of the following statements regarding genome-wide (massively parallel) sequencing is TRUE?

A. It is routinely offered in diagnostics for patients with suspected genetic conditions.
B. The process is very slow.
C. It is much more expensive than sequencing each gene separately.
D. The results give a clear indication of a patient's future health risks.
E. This technology is currently being used in noninvasive prenatal tests.

Case 2
Kelly Armstrong

Kelly Armstrong, 32 years of age, has just presented to you as she has discovered that she is 8 weeks pregnant. You discuss the usual antenatal screening and care, including aneuploidy screening. You inform Kelly of the consequences of a high-risk result, and the risks and benefits of invasive diagnostic testing. Kelly has heard of a new blood test to identify the risk of Down's syndrome and would like to know more about it. You suspect she is referring to non-invasive prenatal testing (NIPT).

Question 5

Which of the following is TRUE regarding NIPT?

A. Kelly could have it at her current gestation.
B. The test costs the patient $200–400.
C. The test cannot identify sex chromosome abnormalities.
D. It reports a risk assessment for trisomy 21, 18 and 13.
E. Results take about 4 weeks to be reported.

Question 6

Kelly asks you how accurate the test is. Which of the following is TRUE?

A. The false-positive rate for trisomy 21 is 2%.
B. The test is less likely to yield a result in obese women.
C. Accuracy for Patau syndrome is 79–92%.
D. About 10% of tests fail to yield a result.
E. Accuracy for trisomy 13 is 99%.

Question 7

Kelly wants to know if she should have NIPT instead of the currently offered first-trimester screening program as NIPT is more accurate. Which of the following statements is true regarding the advantages of NIPT over the current screening program?

A. NIPT has a better detection rate of Down's syndrome, compared with the current screening program.
B. The current screening program is four times more likely to give false-positive result, compared with NIPT.

C. Invasive testing is not needed to confirm a positive result for NIPT.
D. Typically, NIPT can be used for pregnancies that have been conceived with a donor egg, which the current screening program cannot.
E. All of the above.

Question 8

Which of the following is a benefit of the currently offered first-trimester screening program?

A. 44% of non-chromosomal abnormalities are detected.
B. Increased nuchal translucency measurement is also a marker for cardiac abnormalities.
C. Low PaPP-A levels have been associated with intrauterine growth restriction.
D. All cases of gastroschisis are likely to be detected.
E. All of the above.

Case 3

Meaghan Ling

Meaghan Ling, aged 36 years, has come to see you to discuss a website she found offering complete DNA genetic testing. Meaghan is very health conscious and feels she should do everything she can to prevent illness, and wants to know if she should get the genetic testing done.

Question 9

Which of the following is an ADVANTAGE of direct-to-consumer (DTC) DNA genetic testing?

A. It can provide testing for conditions that are not currently publicly funded.
B. Customers are usually well prepared, as they have to go through genetics counseling.
C. Companies offering testing arrange any required medical follow up.
D. There is evidence to show that DTC DNA genetic testing reduces overall mortality and morbidity for patients due to earlier intervention.
E. All of the above.

Question 10

Which of the following should you consider when advising Meaghan about DTC DNA genetic testing?

A. The clinical utility of the test
B. The laboratory validity
C. The clinical validity of the test
D. Your ability to interpret the results
E. All of the above

Question 11

Meaghan wants to know what kind of information DTC DNA testing could give her. Which of the following is TRUE regarding testing for specific conditions?

A. Sickle cell defect is caused by a single gene mutation.
B. Increased risk of breast cancer is caused by a single mutation of BRCA1 or BRCA2 gene.
C. Single nucleotide polymorphisms can be used to identify patients who will go on to develop type 2 diabetes.
D. Pharmacogenetic DNA testing can be used to determine the suitability of warfarin for patients.
E. Pharmacogenetic DNA testing has been shown to be useful in determining appropriate drug doses.

Question 12

Meaghan wants to know how reliable DTC DNA genetic testing companies are. Which of the following is TRUE regarding regulation of DTC DNA genetic testing?

A. The Therapeutic Goods Administration has recently produced a clear set of guidelines regarding DTC DNA genetic testing in Australia.
B. Overseas laboratories offering testing in Australia must comply with the National Pathology Accreditation Advisory Committee guidelines.
C. DTC DNA testing industry has avoided regulatory barriers by claiming that it is a product for information only, not medical decision-making.
D. In the USA, a government investigation found that most DTC DNA genetic testing companies engaged in acceptable marketing practices.
E. All of the above.