



EDUCATION

Genetics in general practice



AFP in Practice questions are designed to get you started in a small group learning (SGL) activity in your practice or with colleagues. Requirements to earn 30 Category 1 CPD points for a SGL activity are: minimum of four and a maximum of 10 people, minimum of 8 hours of discussion in a year, and at least two GPs. Groups may include anyone else who has an interest (ie. practice nurses, community health workers, allied health professionals). A kit with all the instructions and forms you need is available at www.racgp.org.au/afpinpractice. You can also earn Category 2 points based on these questions at AFP practice challenge. Visit www.racgp.org.au/practicechallenge.

Carolyn O'Shea

Learning objectives

After completion of this activity participants will be able to:

- integrate an understanding of Down syndrome screening issues into a consultation that allows patient informed decision making
- gather a family history and apply that information to produce recommendations for the individual patient
- prepare an article for the practice newsletter about haemochromatosis
- discuss critically the ethical issues around genetic screening
- describe methods of identifying patients who should be offered screening for a condition based on their family history.

Category 1 – SGL questions

Domain 1 – communication skills and the patient-doctor relationship

ICPC codes: W78

Melissa and Tom come to see you excited about being pregnant for the first time. Based on dates, Melissa is about 6 weeks pregnant. They report knowing little about pregnancy care and request 'all the usual tests, test for everything'. Focusing on Down syndrome screening: is it a 'usual test'? What information is needed for informed consent pretest? What about when the results are available?

- * Suggested learning activities: group discussion about 'usual tests'; role play the discussion about Down syndrome screening and tests in the current consultation; role play a consultation informing the result at 14 weeks gestation with a Down syndrome screening result of 1 in 150 risk. Discuss the meaning of this and options for future investigation and management.

Domain 2 – applied professional knowledge and skills

ICPC codes: A21, A23

Family history is often easier to interpret when recorded in a genogram. Using a genogram, or another form of recording family history, record a family history and make specific recommendations about indications for screening for conditions or referral to a genetics service.

- * Suggested learning activity: each group member creates a family history genogram, which can be fictional or based on a de-identified patient. Once the history has been taken, group members can compare genograms. Then as a group, decide if there are any indications for screening or referral based on the family history.

Domain 3 – population health and the context of general practice

ICPC code: B99

You have decided that the next practice newsletter should contain an article on haemochromatosis. This is prompted by patients saying it 'runs in their family', but often not knowing more about the disease.

- * Suggested learning activity: in pairs, produce an outline of an article then compare and discuss these outlines in a group. Split the topics up and each write a paragraph to cover your section. You will then have an article for the practice newsletter.

Domain 4 – professional and ethical role

ICPC code: A27

Maria is a patient who you know is looking at becoming pregnant in the next few months. She is estranged from her family, however you also look after Maria's mother, who has told you that Maria's sister recently had a stillbirth related to what sounds like hydrops fetalis. You have asked Maria about her family history and she did not raise this. You suspect she is not aware of the

stillbirth. What are your professional and ethical duties? To whom do you have a duty of care? What are the confidentiality issues? How might you manage the issues in practice?

- * Suggested learning activity: a group discussion about the issues raised in this case.

Domain 5 – organisational and legal dimensions

ICPC codes: A21

The National Bowel Cancer Screening Program has commenced in your area. In discussion, you establish that there have been a few cases of bowel cancer diagnosed in patients of the practice. Some have been via the screening program, however some cases are in younger patients. In reviewing this, you establish that some of these younger patients had indications for screening for bowel cancer based on their family history. How might you identify patients in your practice who have indications for screening? What are the practical steps involved? The National Bowel Cancer Screening website lists indications for screening ([www.health.gov.au/internet/screening/publishing.nsf/Content/bw-familial-guide/\\$File/familial-guide.pdf](http://www.health.gov.au/internet/screening/publishing.nsf/Content/bw-familial-guide/$File/familial-guide.pdf)).

- * Suggested learning activity: develop a method in your practice to identify and manage patients with an indication for screening.