Genomics in pregnancy and medico-legal risk

Quick guide:

- Carrier screening and genomic testing is an evolving area, so keep up to date with College guidelines and seek advice if you are unsure.
- Multiple consultations may be required for patients to understand the implications of screening for themselves and their genetic relatives.
- Patients may still be left with uncertain information and difficult decisions – consider also mental health support.

Introduction

As carrier screening and genetic tests become more widely available, genomic medicine is moving from a highly specialised field to a routine part of medical practice.

Advances in genomics-based research are changing the prevention, diagnosis, and treatment of many conditions, across all areas of practice. More doctors are finding their practice involves some aspects of genomic medicine.

As with any emerging area of practice, it is important to understand the potential limitations and medico-legal issues that genomic testing may present.

Carrier screening

Current guidelines recommend all women planning a pregnancy or in the first trimester of pregnancy should be offered information about carrier screening for genetic conditions (see RANZCOG Guidelines: Genetic carrier screening, and RACGP resource Genetic carrier screening). These recommendations apply in addition to consideration of referral for genetic counselling and specific testing based on family history, ethnicity or population risk.

Explaining carrier screening can be complex, even where patients have a family history of risk factors. For women and couples with no such history, it is particularly important to help them understand:

- what screening may reveal about chances of their child being affected by a genetic condition,
- issues that carrier screening may not identify and additional tests that may still be necessary (including non-invasive prenatal testing and combined first trimester screening), and
- what options they may have based on their carrier status.

Doctors who are not clinical geneticists are not expected to provide the extent of information or counselling that a clinical geneticist would provide. However, courts have found that it is not enough simply to tell a patient about genetic counselling or screening options available. You need to ensure they are given enough information about implications of a screening test for themselves and their child(ren), so that they can make an informed decision.

- Follow relevant college guidelines on genetic carrier screening.
- Keep a careful note of your discussion with the patient, copies of resources you provided and the outcome of the discussion.
- Consider that patients may need multiple consultations to understand their options and make a decision.
- While carrier screening can be performed at any time, screening before a couple conceives gives them the most time to understand and consider their options, such as preimplantation genetic diagnosis.
- If during pregnancy a couple is found to have an increased chance of having an affected pregnancy, alert them to any clinical time restraints on their possible subsequent decisions such as diagnostic testing or termination.

Informed consent for testing

As with any procedure or treatment you recommend to your patient, you need to give them enough information so they can make an informed decision. The focus of the consent process is the discussion and shared decision making with the patient. For more information on the consent process, see Avant's factsheet Consent: the essentials.

Genomic testing can present additional issues you need to keep in mind – particularly because:

- the science and ability to detect genetic variations is changing rapidly.
- testing may have implications for individuals other than the patient – including other family members, parents or donors.
- risk can be especially difficult to understand in the context of genetic variation.

Patients may find it difficult to understand that tests showing genetic variation may still not give them certainty about whether they or their child will ever go on to develop a condition. As broader panel tests become available, there is increased concern about 'variations of unknown significance' being identified.

In general, you should discuss:

- Details of the condition(s) being tested for.
- If tests are being performed outside of Australia.
- The potential outcomes and implications for the patient.
- Options and timing constraints (particularly for testing in the antenatal context).
- The possibility of unexpected findings.
- Possible implications for other genetic relatives.
- Whether the testing is part of a research program and any research their data may be used for. Generally, if the test is part of a research program the patient should receive a specific consent form and specific information.
- Costs of the tests. Reproductive carrier screening generally incurs out of pocket expenses where there is no family history of the condition.

As with any consent process, it can be useful to refer to written or illustrated material and other information to assist a patient's understanding. It may be necessary to refer the patient for genetic counselling or to another specialist for further information.

Privacy and confidentiality issues

Sending samples overseas

Since many genetic tests are conducted overseas, it is important to understand your obligations under the Privacy Act (Cth) and Australian Privacy Principles and follow your practice policies if you are disclosing a patient's personal information to an overseas recipient (such as sending a sample overseas for testing).

Disclosure to relatives without consent

You should explain to your patients that in some circumstances, genetic information may be disclosed to genetic relatives without their consent.

Under the Privacy Act, health practitioners working in private practice in Australia may be permitted to disclose genetic information without the consent of their patient if:

- reasonable steps to obtain consent from the patient are unsuccessful, and
- disclosure is necessary to lessen or prevent a serious threat to the life, health, or safety of a genetic relative of that patient.

Practitioners must follow the National Health and Medical Research Council's (NHMRC) guidelines and process for engaging with patients and their genetic relatives.

Doctors in health organisations covered by state or territory information privacy provisions (such as public hospitals) would need to follow the legislative requirements in their state or territory.

In practice, we understand that with time and counselling, most patients agree at least to inform relatives they should consider testing. If patients do not consent the situation can be complex and challenging, and you should consider getting legal advice from Avant.

- Be aware of your privacy obligations and make sure you follow the legislation and your practice policies, particularly if you are transferring information outside Australia for testing.
- Seek advice if you believe you may need to disclose information to genetic relatives without the patient's consent as the situation can be complex.

Test results and follow-up

Some issues to be aware of in the context of genomic testing are:

- the complexity and variety of test results
- availability of direct-to-consumer testing
- emerging understanding of implications of genetic variants.

Given the variety of tests available in Australia and overseas, and the differing way in which results are reported, reading test results may not be straightforward.

A 'negative' result may mean a particular variant was not tested, or that the significance of a particular variant is still unclear.

There have been cases where patients have been given incorrect information about their risk of developing or passing on a genetic disorder because practitioners were unfamiliar with the test or reporting format.

Also, identifying a particular variant does not necessarily indicate an increased risk of disease in the absence of family history or other known risk factors.

Direct-to-consumer genetic tests

DNA tests available to the public can add to patient and practitioner confusion.

Genetic self-tests to detect disease are currently prohibited by the Therapeutic Goods Administration from supply in Australia. A recent TGA review highlighted concerns about security and integrity of personal information collected by these tests, as well as accuracy and interpretation of the results. However, patients may access such tests from overseas or may get information from DNA tests for diet, wellness or lifestyle purposes.

You have no obligation to follow up direct to consumer tests that a patient may have had, but your patient may ask you about using these tests or about the implications of the results.

The NHMRC has published a guide for health professionals when discussing direct-to-consumer DNA testing with patients. It warns against using these tests as the basis for clinical decision making because the test has unknown analytical or clinical validity. Instead, ask what prompted the patient to consider undertaking a genetic test and what they hope to gain and determine whether and how you can provide care.

Changing classification of variants

As genomic science develops, the implications of particular variants may be better understood. This raises practical and ethical issues about how to follow up with patients as new information comes to light.

- Use the NHMRC guidelines (and your own practice policy of you have one) to guide your discussions with patients about direct consumer tests.
- Check your policy for delivering results and follow-up, ensure it is clear who is responsible for following up with patients. For more information on follow-up, see Avant's factsheet: Patient follow-up and recalls.
- Ensure your patient understands the implications of variants of uncertain significance and whether they should follow up or retest at a time in the future.

Important references

Genetic testing and carrier screening

RANZCOG: Statement on genetic carrier screening C-Obs 63

RACGP: Beware the rare – professional and patient resources on genetic carrier screening

RACGP Genomics in general practice

Avant Genomic testing and medico-legal risk – discussion paper

National Health and Medical Research Council. Discussing Direct-to-Consumer Genetic DNA Testing with Patients

Privacy and confidentiality

Office of the Australian Information Commissioner: Guide to Health Privacy, see particularly Chapter 8: Using and disclosing genetic information in the case of a serious threat

National Health and Medical Research Council. Use and disclosure of genetic information to a patient's genetic relatives under Section 95AA of the Privacy Act 1988 (Cth): Guidelines for health practitioners in the private sector.

Office of the Australian Information Commissioner Chapter 8: APP 8 – Cross-border disclosure of personal information - Home (oaic.gov.au)

Avant resource page: Data breaches: all you need to know

Consent

Australian Genomics: National clinical consent forms

Avant factsheet: Consent: the essentials

Further information

For more information or immediate medico-legal advice, call us on 1800 128 268, 24/7 in emergencies.
avant.org.au/mlas

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