

Genetic testing

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Patients and questions of genetics.

Personalised medicine, involving the capacity to predict disease development and influence decisions about lifestyle choices or to tailor medical practice to an individual, is now a reality made possible by advances in genomic technology.¹

The increasing availability of genetic testing brings the promise of improved patient outcomes and disease prevention. However, as with any change in medical practice, this situation presents challenges and potential risks.

What are the medico-legal issues that GPs may face when dealing with genetic testing?

Direct-to-consumer DNA genetic testing

How will you respond to an anxious and distressed 18-year-old patient who wants your advice about the '23andMe' DNA analysis test results she has just received?

The National Health and Medical Research Council (NHMRC) has useful guidelines for healthcare professionals about discussing direct-to-consumer tests with patients:

- Explain that the test may not be of a standard that enables it to be used as the basis of therapeutic advice.
- Explain that as well as concerns about the quality of overseas tests, for most diseases, a person's genes interact with environmental and lifestyle factors, and this is another reason why a course of action based solely on the test is not appropriate.
- Explore why the patient wanted the test. Informed by your knowledge of the patient's medical history, determine whether the clinical problem warrants the need for diagnostic investigation through an accredited Australian laboratory.

- Consider also whether there is an alternative test that may be more likely to yield actionable information.
- Consider also referring the patient to a genetics specialist or counsellor with the necessary expertise in the area of concern to the patient, who can also explain the implications that genetic tests may have for their blood relatives.
- Document the advice provided, and any agreed action, in the patient's medical record.
- Direct the patient to the NHMRC publication 'Direct-to-consumer DNA genetic testing: An information resource for consumers' for more information.

Reproduced with permission from the National Health and Medical Research Council. Discussing direct-to-consumer genetic DNA testing with patients: A short guide for health professionals. Canberra: NHMRC, 2013.

Disclosure of genetic information to relatives without the patient's consent

Your 48-year-old patient has just been diagnosed with Huntington disease (HD), and does not want his family to know. His daughter is also your patient. She is planning a pregnancy and, if she were aware of her father's HD, you believe she may choose to have pre-implantation embryonic diagnosis.

Under the *Privacy Act 1988*, disclosure of genetic information to relatives without a patient's consent is strictly regulated:

- Use or disclosure of genetic information without consent may proceed only when the authorising medical practitioner has a reasonable belief that this is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.
- Specific ethical considerations must be taken into account when making a decision about whether or not to use or disclose genetic information without consent. Any departures from maintaining confidentiality must be taken very seriously and should be the exception rather than the rule. The decision should involve consideration of the likely effect on the patient of breaching confidentiality and the possible ambivalence of genetic relatives to receiving genetic information.
- Reasonable steps must be taken to obtain the consent of the patient (or his/her authorised representative) to use or disclose genetic information.
- The authorising medical practitioner should have a significant role in the care of the patient and sufficient knowledge of the patient's condition and its genetic basis

to take responsibility for decision-making about use or disclosure.

- Prior to any decision concerning use or disclosure, the authorising medical practitioner must discuss the case with other health practitioners with appropriate expertise to assess fully the specific situation.
- Where practicable, the identity of the patient should not be apparent or readily ascertainable in the course of inter-professional communication.
- Disclosure to genetic relatives should be limited to genetic information that is necessary for communicating the increased risk and should avoid identifying the patient or conveying that there was no consent for the disclosure.
- Disclosure of genetic information without consent should generally be limited to relatives no further removed than third-degree relatives.
- All stages of the process must be fully documented, including how the decision to use or disclose without consent was made.

Reproduced with permission from the 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. Canberra: NHMRC, 2009.

Requests for parentage testing

A father presents with his two-year-old son and a genetic testing kit. He wants you to complete 'chain of custody' testing and documentation for Family Court proceedings.

This type of testing should only be performed with the written informed consent of all persons with parental responsibility for the child or pursuant to other lawful authority, such as a court order. Specialist counselling may be required as part of the consent process.

Where one person with parental responsibility cannot be contacted or withholds consent, the GP should advise the parent that a court should be contacted and authorised to make a decision related to parentage testing on behalf of the child. 🗣️

Reference

1. National Health and Medical Research Council. Information for health professionals: Clinical utility of personalised medicine. Canberra: NHMRC, 2011. Available at www.nhmrc.gov.au/_files_nhmrc/publications/attachments/ps0001_clinical_utility_personalised_medicine_feb_2011.pdf [Accessed 27 April 2017].

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