



Sara Bird

Genetic testing: medico-legal issues

Background

The availability and frequency of genetic testing is increasing. Genetic testing poses some unique ethical and legal issues for medical practitioners because of the potential to identify genetic variants that carry implications for the risk of disease in the future for the patient and their relatives. The regulatory framework within which genetic testing is provided in Australia is also changing.

Objective

This article examines some medico-legal issues associated with genetic testing that general practitioners (GPs) are likely encounter in their practices.

Discussion

There is inevitable involvement of the GP in the long term care of a patient (and possibly their family) following genetic testing, regardless of whether or not the GP has ordered the testing. Cases are presented to illustrate some of the medico-legal issues that may arise from direct-to-consumer genetic testing, information disclosure to genetic relatives and requests for parentage testing.

Keywords

genetic testing; medico-legal aspects

Direct-to-Consumer Genetic DNA Testing with Patients – A Short Guide for Health Professionals, warns that health professionals should carefully consider the implications of discussing a direct-to-consumer genetic test result with a patient, noting that a patient may be placed at harm should the health professional attempt to provide therapeutic advice on the basis of a test of unknown analytical and clinical validity.¹

The Guide provides the following advice: Giving advice to patients who present with the test results from a direct-to-consumer DNA genetic test

- Explain that the test may not be of a standard that enables it to be used as the basis for 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's publication 'Direct-to-Consumer DNA Genetic Testing: an information resource for consumers' for more information.¹

Direct-to-consumer genetic testing

Case

A patient aged 28 years presents to her general practitioner (GP) with the results of her genetic tests, which she had ordered online to check her risk of breast cancer after reading about Angelina Jolie.

Direct-to-consumer genetic tests are tests that are conducted by laboratories without requiring a referral from a health professional. These tests may cover health and non-health matters, such as paternity and ancestry. The tests are often promoted on websites by laboratories that are overseas, and, therefore, not subject to Australian regulations, such as the Therapeutic Goods Administration.

A publication produced by the National Health and Medical Research Council (NHMRC), *Discussing*

Disclosure of information to genetic relatives without consent

Case

A patient aged 38 years was recently diagnosed with metastatic bowel cancer. Investigations had revealed an underlying diagnosis of familial adenomatous polyposis associated with a mutation of the APC gene. The patient's discharge summary stated that there was a 50% chance that his siblings and children had inherited the condition and recommended that they undergo assessment. The patient did not want any of his relatives to be informed about the genetic basis for his terminal illness. The patient's sister and his two teenage children also saw the GP as patients. The GP wanted to disclose the information to the patient's relatives to enable them to undergo appropriate management.

The NHMRC's publication *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 outlines the requirements that must be met for GPs (who are working in private practice) to disclose genetic information to relatives without patient consent.² These requirements are that:

- 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.

Requests for parentage testing

Case

A father attends a consultation with his 3-year-old son. The father has brought a genetic testing kit and he asks that the GP undertake 'chain of custody' testing of himself and his son for the purpose of establishing paternity of the child.

Parentage testing refers to testing conducted to confirm or deny the biological parentage of a particular child or individual. Most parentage testing is 'paternity' testing – that is, testing to determine the biological connection between a child or individual and the supposed father.

In general terms, where a child does not have sufficient maturity to make a decision as to whether to submit a genetic sample for parentage testing, such 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 about parentage testing on behalf of the child.

GPs are encouraged to contact their medical defence organisation for advice in a specific situation.

Author

Sara Bird, MBBS, MFM (Clin), FRACGP, is Manager, Medico-Legal and Advisory Services, MDA National. sbird@mdanational.com.au

Competing interests: None.

Provenance and peer review: Commissioned, externally peer reviewed.

References

1. National Health and Medical Research Council. Discussing Direct-to-Consumer Genetic DNA Testing with Patients: A Short Guide for Health Professionals. Canberra: Commonwealth of Australia, 2013.
2. 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: Commonwealth of Australia, 2014.

This article is provided by MDA National. They recommend that you contact your indemnity provider if you have specific questions about your indemnity cover. The scenarios are based on actual medico-legal referrals; however certain facts have been omitted or changed by the author to ensure the anonymity of the parties involved.

correspondence afp@racgp.org.au