Assessing the direct-to-consumer (DTC) genetic testing results of your patient—a quick guide for general practitioners

The National Health and Medical Research Council (NHMRC) recommends that health-related genetic testing should be conducted in an accredited Australian laboratory with professional involvement and support.1 This quick guide provides GPs with support and information when their patients present with genetic test results completed through an external mechanism.

Points for GPs

• GPs cannot be expected to know about every DTC genetic test available.
• Accurate and detailed information about the test your patient has taken, and the credibility of the company providing the test, is critical in informing the interpretation of results. While some companies imply that they are providing a medical test, look carefully at the disclaimers which will highlight the purpose of the test and its limitations.
• DTC testing results are often complex and difficult to interpret.
• It may not be possible for you to interpret test results your patients bring in, and it will often be necessary to refer the patient to a specialist/s for further investigation and/or interpretation of results.
• While testing may benefit some patients and their families, it may also cause harm – the patient might need sensitive management, and genetic counselling.

For health professional support resources, see the following NHMRC documents:

• Medical Genetic Testing: Information for Health Professionals (2010)
• Clinical Utility of Personalised Medicine: Information for health professionals (2011)
• 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 (2009)


Assessing the DTC results of your patient—a quick guide for GPs

• Is the test predictive or diagnostic?
• Explore why the patient wanted the test (known family history? Simple curiosity? Specific fears, concerns, or expectations?)
• Evaluate and interpret the results received:
  – What are the features and limitations of the test?
  – Are the results definitive or suggestive? Clinically significant or useless?
• Is there a need for further investigation?
• What are the ethical, legal and social implications? These include those for life insurance, and the possible tension or conflict between the rights of different family members and the patient to:
  – know, or not to know, information relevant to their own health (autonomy)
  – disclose, or not, personal information (privacy)
  – make an informed decision regarding genetic testing.

It is important to explore with the individual the potential harms and benefits and their reasons for or against disclosure to relatives. A GP may encourage and support the patient to share the information with their relatives, but in most cases, a GP has no duty to inform the relatives of a patient about a positive genetic test result. Nevertheless, changes to the Privacy Act 1988 (Cth) in 2006 allow private health practitioners to disclose patient’s genetic information, whether or not they give consent, under certain circumstances.1
• Refer to genetic support groups, genetics services and/or other specialists as required. In addition to GP support, referral to genetics services, professional counselling and support organisations can be beneficial and, in some cases, necessary for the wellbeing of the patient and/or their family.

(See http://www.genetics.edu.au/conditions/geneticsupportgrouplist for a genetic disease support group list).

Getting the most of the consultation

The more detail your patient provides about the DTC test and company, the more likely you will be able to make an informed interpretation.

The following section includes advice you can provide to your patient who has undergone a DTC genetic test on what to expect and on how to get the most out of the consultation.

What does your genetic test result mean?

Advice for patients on how to get the most out of their GP consultation

• Bring in your direct-to-consumer genetic test results and any other relevant information, such as information about the test you purchased and about the company.

• Interpreting the result will require additional information such as your family health history. Gather as much information as you can about your blood relatives including age at diagnosis and age and cause of death.

• In order to review the results and information, consultations longer than the standard 15 minutes will likely be required. It is advisable to discuss this when you make a booking.

• Understand that genetic diseases by their nature run in families, so your results may have implications for your blood relatives.

• Before your appointment: review your results and make a list of concerns you have and questions you wish to ask before the appointment and take it with you.

• Be aware that your GP may not be able to give you conclusive answers. Certainty depends on the type and purpose of the test you ordered and the detail contained within your test results.

• Additional testing (e.g. a blood test) or further investigation may be indicated in order to validate the original direct-to-consumer genetic test results.

• It may be helpful to bring a trusted family member or friend to the consultation to provide moral support and help recall.


2 See NHMRC’s 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 for the requirements that must be met by private health practitioners if they choose to use or disclose genetic information without patient consent under the Privacy Act 1988 (Cth).