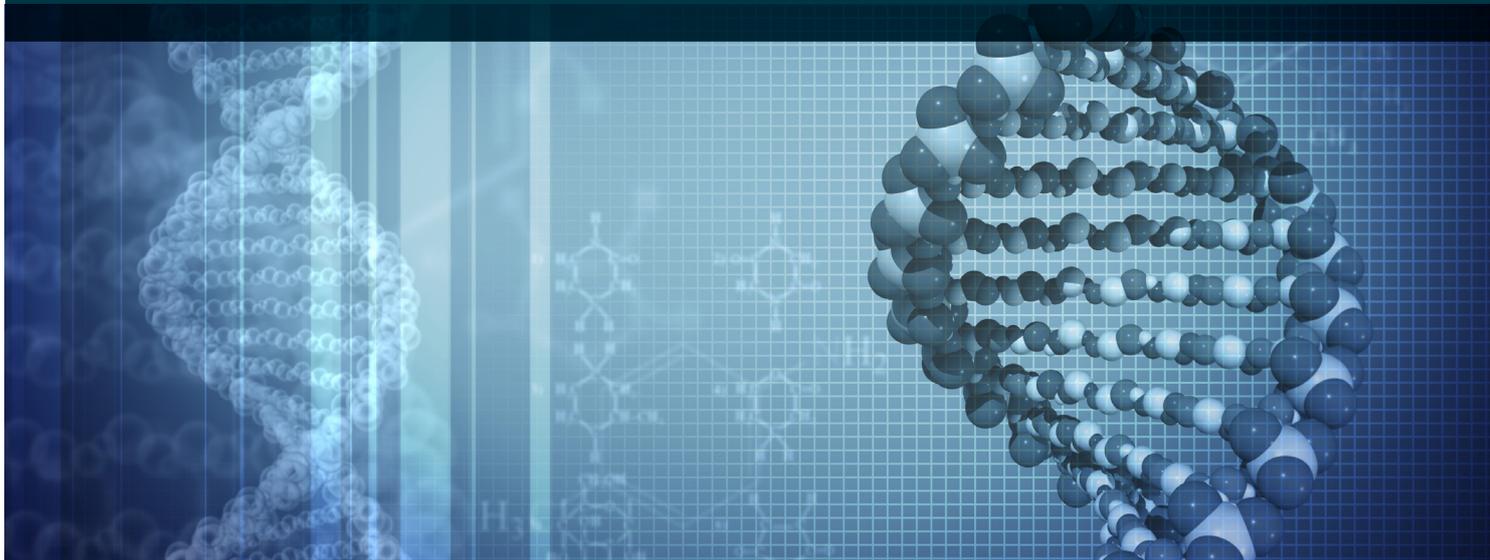




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Discussing Direct-to-Consumer Genetic
DNA Testing with Patients
A Short Guide for Health Professionals

WORKING TO BUILD A HEALTHY AUSTRALIA

DECEMBER 2013 | NHMRC REF#G7

DISCUSSING DIRECT-TO-CONSUMER GENETIC DNA TESTING WITH PATIENTS – A SHORT GUIDE FOR HEALTH PROFESSIONALS

This short guide provides health professionals with information to assist when patients present with genetic test results that have been obtained from organisations marketing directly to the consumer (known as direct to consumer, or DTC, genetic tests), or when patients wish to discuss the option of purchasing such tests. This guide does not apply to subjects undertaking genetic testing in the context of a research study.

This guide will assist health professionals in explaining that DTC genetic tests may have a range of consequences that need to be carefully considered by the patient. These may include results that the patient is not prepared for or predispositions to conditions for which there are no treatments. The test may also impact on an individual's ability to obtain risk rated insurance, such as life insurance. These issues may have implications for both the patient and their family members. It should also be highlighted that genes are not the sole determinant of a patient's future health. Lifestyle, environmental factors, family history and preventative measures including health care options are also important contributing influences.

Growth of genetic and genomic medicine

Advances in genetics and genomics have greatly improved our capacity to understand, diagnose and screen for some genetic disorders, and research is continuing to translate gene discoveries into disease treatments. However, many genetic tests are not yet underpinned by robust evidence of their clinical value. While thousands of genes have been mapped, relatively few have been identified as being involved in a genetic disorder. Further, the vast majority of medical conditions with some genetic link involve either the complex interaction of a number of genes, or the complex interaction between genes, lifestyle and the environment.

Consequently, the interpretation of genetic test results is complex and needs to be carried out by appropriately qualified professionals, who can also offer or refer patients to pre- and post-counselling and management advice.

What is direct-to-consumer DNA genetic testing?

Whereas conventional genetic testing is initiated by a health professional, direct-to-consumer genetic testing is initiated by the consumer. These tests are often promoted on websites by laboratories that are located overseas. The consumer deals directly with the laboratory online, for a fee. A test kit is sent to the consumer, who sends a sample, usually taken from a cheek swab, to the laboratory. The laboratory staff analyse the sample and provide the results directly to the consumer.

Consumers purchase these tests not only for health information, but to establish parentage and for general interest, such as tracing ancestry.

The prevalence and uptake of direct-to-consumer genetic tests is increasing, so it is important for health professionals to be aware of the issues surrounding these tests and their limitations.

Note that DTC genetic testing is different to 'Community Genetics', where health professionals arrange for genetic testing to be offered to the relevant targeted at-risk community. The consumer then initiates testing, which is offered in conjunction with appropriate educational materials and programs, involves access to counselling, and testing is performed in an Australian accredited laboratory. An example of Community Genetics is testing for Tay-Sachs disease in Ashkenazi Jewish communities. Community Genetics has the full support of the Royal College of Pathologists of Australasia, the Human Genetics Society of Australasia and State and Territory Health Departments.

Key issues with direct-to-consumer DNA genetic testing

Overseas laboratories may not be required to meet the rigorous quality standards that apply in Australia

In Australia, genetic tests are categorised as in-vitro diagnostic medical devices (IVDs), and those used for health-related purposes are regulated by the Therapeutic Goods Administration (TGA). The TGA requires that genetic tests performed by Australian laboratories comply with rigorous standards relating to quality, safety and performance. In addition, the National Pathology Accreditation Advisory Council (NPAAC) develops quality standards which Australian pathology laboratories must meet in order to gain accreditation by the National Association of Testing Authorities of Australia (NATA), which enables them to claim Medicare benefits for their services.

Providing genetic test results that have been performed for medical purposes directly to the patient is illegal in Australia, in recognition of the importance of specialist advice and counselling on the results of such tests.

Overseas companies offering direct-to-consumer genetic tests may not be required by their respective authorities to follow these standards to which Australian laboratories must adhere. There is no consistent oversight of the genetic testing marketplace internationally, and some countries may exert little or no regulatory control over these tests.

Direct-to-consumer genetic tests may not have analytical and clinical validity

Direct-to-consumer genetic testing provided by overseas companies and purchased through the internet are not subject to scrutiny by the TGA, and are not evaluated by the TGA for their safety, quality and performance. The TGA and NPAAC requirements which must be met in Australia cover all aspects of testing, including laboratory facilities, manufacture of the medical device, the proper collection of specimens, the accuracy and quality of the test method and test results, and strict record keeping, to ensure that test results are not mixed up and provided to the wrong person.

When these standards are not met, test results may be misleading, unclear or inaccurate. They may not provide any medically useful information, or worse, may be harmful, for example by prompting patients to pursue unnecessary medical interventions; or by falsely reassuring patients, who consequently do not take follow-up action. Some genetic tests are not diagnostic but predictive, and their predictive power is often limited. They may suggest an increased risk for developing a condition which, given the complex interaction of genes, lifestyle and environment, many people will never develop.

Direct-to-consumer genetic tests should not be used as the basis for clinical decision making and health care

Health professionals should carefully consider the implications of discussing a direct-to-consumer genetic test result with a patient. A patient may be placed at harm should the health professional attempt to provide therapeutic advice based on a test of unknown analytical validity (how accurately the test detects the presence or absence of a particular gene or genetic variation) and/or clinical validity (how well the gene being analysed is related to the presence, absence or risk of a specific disease).

Giving advice to patients considering accessing direct-to-consumer genetic testing

- Ask what prompted the patient to consider undertaking a genetic test and what they hope to gain.
- Explain that genetic tests conducted overseas may lack scientific validity and the results may be misleading, unclear, or inaccurate due to the possibility that overseas laboratories may not be required to observe the same quality standards as Australian laboratories.
- Explain that even genetic tests that do meet high quality standards may not provide any medically useful information.
- Explain that, due to the shared nature of genes, there may be family issues that need to be considered.
As an example, obtaining one's own genetic tests results may reveal information about one's blood relatives. Conversely, if other family members obtain genetic test results, it may reveal information about your patient, which the patient may or may not wish to know.
- Explain that there may be social, cultural and legal issues that need to be considered for certain groups. As an example, Aboriginal and Torres Strait Islander Peoples believe that information about their heritage is often collectively owned and so it is important for them to consider the issues that their decision to be tested may raise for their community .
- If the patient's concern is clinically valid, consider whether there is an alternative test that may be more likely to yield actionable information.
- Consider also referring the patient to a genetic specialist or genetic 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 and whether the test will provide them with clinically useful information.
- Advise that genetic tests become part of the person's medical record and may have implications for third parties such as employers and insurers: in Australia, applicants for life insurance are legally obliged to disclose genetic test information to the life insurer. Under the Disability Discrimination Act, insurers are allowed to consider genetic information in underwriting life insurance decisions.

Example: A male in his 20s was denied full life insurance cover because he revealed that he had discussed genetic testing with a genetic counsellor. After having a genetic test and being diagnosed with a mutation in the MSH6 gene, he disclosed this information to another two insurance companies and was again denied life insurance. After researching the disease and learning that annual colonoscopies would greatly reduce his risk of cancer, he made a complaint to the Human Rights Commission (HRC). After advising the third company of his HRC complaint, the company then offered him full coverage. This would suggest the company did not have evidence or data that would have supported its earlier decision.¹

- Request a genetic test through an accredited Australian laboratory only when the results will contribute to diagnosis and/or management.
- Advise that Medicare rebates and tests funded through state governments are available for some clinically indicated genetic tests when performed by an accredited Australian laboratory, but tax payer subsidised rebates are not available for direct-to-consumer DNA genetic tests.
- Direct the patient to the NHMRC's publication '*Direct-to-Consumer DNA Genetic Testing: an information resource for consumers*' for more information.
- Document the advice provided, and any agreed action, in the patient's medical record.

¹ Keough, LA & Otlowski, MFA, Life insurance and genetic test results: a mutation carrier's fight to achieve full cover, *Ethics and Law, MJA*, vol 199, 2 September 2013. P 363-366.

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.

Additional information on genetics:

- Understanding Direct-to-Consumer Genetic DNA Testing: An Information Resource for Consumers
<http://www.nhmrc.gov.au/guidelines/publications/g8>
- Direct-to-Consumer Genetic Testing: A Statement from the National Health and Medical Research Council (NHMRC)
<http://www.nhmrc.gov.au/guidelines/publications/g9>
- Clinical Utility of Personalised Medicine: The Australian Handbook for General Practitioners
<http://www.nhmrc.gov.au/guidelines/publications/ps1>
- Genetics in Family Medicine: The Australian Handbook for General Practitioners
<http://www.nhmrc.gov.au/your-health/egenetics/health-practitioners/genetics-family-medicine-australian-handbook-general-prac>
- Medical Genetic Testing: Information for Health Professionals
<http://www.nhmrc.gov.au/guidelines/publications/e99>
- Medical Genetic Testing: Health Information for You and Your Family
<http://www.nhmrc.gov.au/guidelines/publications/ps3>
- 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
<http://www.nhmrc.gov.au/guidelines/publications/pr3>