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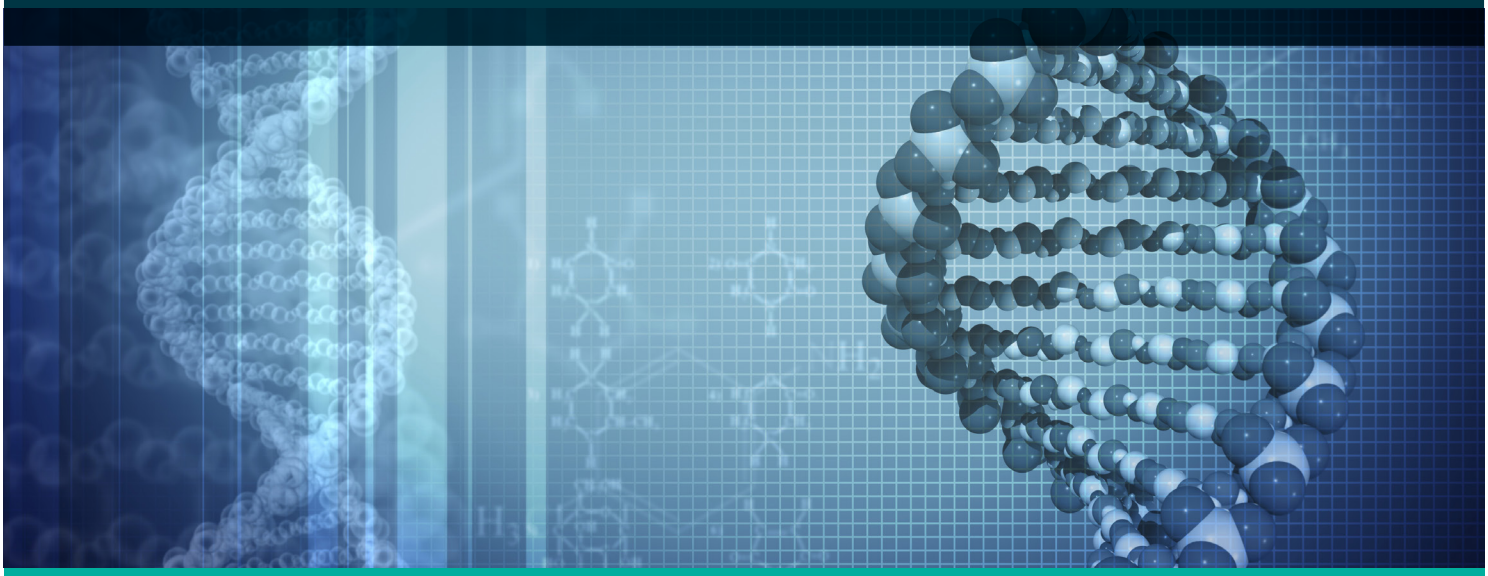

Direct-to-Consumer Genetic Testing
A Statement from the National Health and Medical Research Council (NHMRC)

WORKING TO BUILD A HEALTHY AUSTRALIA

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DIRECT-TO-CONSUMER GENETIC TESTING-

A STATEMENT FROM THE NATIONAL HEALTH AND MEDICAL RESEARCH COUNCIL (NHMRC)

Direct to consumer (DTC) genetic tests are purchased directly by consumers, often over the internet and usually without the involvement of a medical professional.

The challenges for health professionals and consumers presented by the recent proliferation of DTC genetic tests are a focus of the National Health and Medical Research Council (NHMRC). This statement concerns DNA genetic tests that are being, or potentially could be, used in the diagnosis of medical conditions or to indicate pre- disposition to particular diseases. The statement highlights important issues for the appropriate delivery and use of health- related genetic testing.

Genetic testing, along with the methods by which a genetic test is offered, has both benefits and pitfalls. Individuals may undergo DTC genetic testing for a range of reasons, which often reflect personal priorities. Such motivation allows greater engagement and appreciation for science and for what makes us unique. However, of potential concern is the packaging of such tests 'for fun' or 'for informational purposes' when they also offer findings that have the potential to affect health outcomes. Irrespective of whether a genetic test is offered through a health professional (medical genetic tests), or directly to consumers (DTC genetic tests), NHMRC considers the following three elements to be integral to the appropriate delivery and use of health- related genetic testing:

- Consumer information and support;
- A robust evidence base; and
- Professional involvement and education.

The status of DTC genetic testing is currently being reviewed by Australian regulatory authorities to ensure that it is included within the national quality and regulatory framework. When performed in accredited laboratories that meet quality standards, and when used for selected conditions where clinical validation and utility has been demonstrated, they may indicate the presence of genetic disorders and offer the potential for prediction of future susceptibility to medical conditions.

Given the wide range of issues that this type of genetic testing presents, NHMRC considers that there is a need for caution for individuals opting to undertake DTC genetic testing. The need for caution is supported by the recent action of the US Food and Drug Administration (FDA) ordering a major DTC genetic testing company to cease providing health-related testing until such time that the company can clinically validate its test results.

NHMRC has developed a range of information and guidance products on genetics and genetic testing. These are listed at the back of this Statement. These resources are intended to assist health care professionals in their consultations with patients, and to assist consumers who are considering DTC genetic testing.

The need for consumer information and support

NHMRC encourages individuals interested in undertaking a genetic test to exercise caution and, before initiating DTC testing, to consider the implications for themselves and their family.

Genetic testing results can have implications for family members due to the shared nature of genetic information. Individuals should consult their general practitioner about these potential implications as well as the various available options, including whether there is an actual need for medical genetic testing and/or standard pathology. Following this consultation, individuals may then wish to discuss with their family, both their intention to access DTC genetic testing before they commence such testing, as well as their DTC results after testing.

Consumers also need to be aware that results of genetic tests are generally required to be declared when seeking to obtain risk-rated insurance products, such as life insurance or income protection insurance. Where test results (whether genetic or non-genetic) identify increased health risks, this could have an impact on an individual's ability to obtain this type of insurance.

DTC genetic tests can seem appealing because sample collection can be done comfortably at home and access to results by others may seem limited. However, what may not be immediately clear to the consumer is that their DNA sample and the results of analysis are held by a third party, usually based overseas. Australian law protects an individual's privacy rights for services provided in this country, but these protections do not extend to overseas-based services. For example, some DTC testing policies allow for genetic test results to be sold to third parties. Consumers also need to be aware that the disclaimers attached to some DTC genetic testing may absolve the company of responsibility for inaccurate test results. Consumers should carefully examine the privacy policies and terms and conditions of their chosen DTC genetic testing company to ensure that these are what the individual finds acceptable.

The need for robust evidence

NHMRC considers that genetic tests should be underpinned by robust scientific evidence about their utility and potential role in clinical care. This means that the use of a genetic test should lead to clinical decisions that improve patient outcomes.

Every individual's DNA will have a large number (approximately one million) of variations from the reference human genomic sequence. However, only a relatively small number (approximately 20 thousand) of human genes have been identified, as well as an additional as-yet-unknown number of (usually non-coding) regulatory elements. This numerical disparity means that only a small fraction of these many variants have been demonstrated to be associated with medical conditions. At our current stage of understanding, the vast majority of variants will be benign and unlikely to be medically significant. It is a complex and scientifically demanding process to validate a disease association with a genetic variant, and to demonstrate clinical utility. Only a tiny fraction of these many genetic variants have so far been demonstrated to inform clinical care and/or assist in improving patient outcomes.

To date, only a small minority of DTC genetic tests have established their clinical value. While this will undoubtedly change with time, at present the majority of DTC genetic tests are still research activities. Because of this lack of agreed evidence base, different DTC genetic testing companies have sometimes been found to return different interpretations of the same DNA sample, due to different analytical or post-analytical approaches.[1] Some companies have also been found to provide disease predictions that conflict with established risk for medical conditions.[2]

The importance of professional involvement and education

NHMRC encourages the involvement of health professionals, including medical practitioners, clinical geneticists and genetic counsellors in the genetic testing process.

The growing uptake of DTC genetic tests is in part driven by individuals who are taking more responsibility for their own health. However, they can also experience worry and confusion if there are unexpected findings or unclear results. While there are different models of DTC result reporting in use, some DTC genetic testing services provide results directly to the consumer without the involvement of a health professional, which means that access to professional advice may be limited, or provided indirectly or only after the fact.

Professional involvement is important in providing individuals and their families with balanced information and an appropriate clinical context. Most common diseases involve multiple and complex interactions between genetic and environmental factors.

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1 P C Ng, S S Murray, S Levy & J C Venter, 'An agenda for personalized medicine', Nature, Vol 461, 2009, pp. 724-26

2 G Kutz, 'Direct-to-Consumer Genetic Tests, Misleading Test Results Are Further Complicated by Deceptive Marketing and Other Questionable Practices' Proceedings of the Subcommittee on Oversight and Investigations, Committee on Energy and Commerce, House of Representatives, 2010.

To avoid causing unnecessary physical or psychological harm and potentially unnecessary follow-up testing and examination, medical investigations should only be ordered where tests are reliable, where there is a professionally identified clinical need, and where clinical utility and effectiveness have been established. These same principles apply to non-genetic testing, to established genetic testing, and now also to DTC genetic testing. To support patients who have had a DTC genetic test, health professionals need to take into account the quality and reliability of test services. Laboratory services should be accredited to Australian medical testing standards (or international equivalents). Test reports should conform to the emerging standards and recommendations of peak pathology and genetics professional organisations.

While it is important to be familiar with the details of a particular DTC genetic service in order to interpret reports from that particular provider, the health professional will likely also need supplementary support and development such as that typically facilitated and coordinated by relevant professional organisations.

Explaining the implications of an identified genetic variation requires considerable expertise. In addition to technical understanding, medical genetic testing requires that health professionals give due consideration to the wide range of ethical, legal and social issues that result from genetic testing. The body of scientific and medical knowledge is rapidly expanding, and health professionals should either ensure they remain abreast of current practice, or seek the advice of appropriate others, for example a clinical geneticist or genetic counsellor.

The future of DNA genetic testing

Rapid advances in genetics have led to major impacts upon health practitioners' needs to better understand and provide information and referrals on genetic conditions for patients and their families. Health practitioners are increasingly likely to find themselves providing advice on genetics, and perhaps also advising on the utility of and/or results from DTC genetic tests.

Ease of access and falling costs are likely to result in continued consumer interest in DTC genetic testing. The medical role of much DTC genetic testing is evolving and is yet to be fully established. The clinical use of unvalidated or unaccredited DTC tests results should be discouraged. By focusing on ensuring the quality of testing and the scientific evidence base of clinical utility of test results, health professionals can assist patients and consumers make informed decisions as to whether DTC genetic testing is appropriate for them.

Through its Human Genetics Advisory Committee, NHMRC will continue its ongoing dialogue with professionals and the Australian community to support the development of knowledge on genetic testing in the interests of the health of Australians.

Additional information on genetics:

- Understanding Direct-to-Consumer Genetic DNA Testing: An Information Resource for Consumers
  http://www.nhmrc.gov.au/guidelines/publications/g8
- Discussing Direct-to-Consumer Genetic DNA Testing with Patients: A Short Guide for Health Practitioners
  http://www.nhmrc.gov.au/guidelines/publications/g7
- The Provision of Direct-to-Consumer Genetic Tests: Guiding Principles for Providers
  http://www.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-path-bestpractice
- 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