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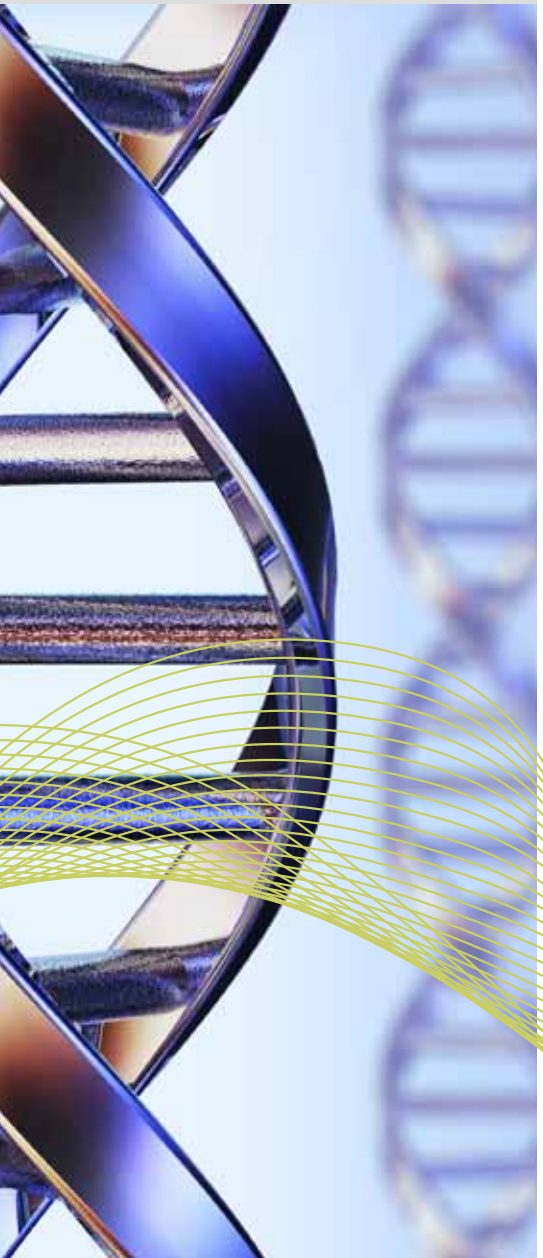
DNA Genetic Testing in the Australian Context:

A Statement from the National Health and Medical Research Council (NHMRC)

Inside



Genetic tests are available both through health professionals and now increasingly directly to consumers via the internet for a range of applications. Test applications include the determination of parentage and ancestry, prediction of future susceptibility and the diagnosis of genetic disorders. The developments and challenges for health professionals and consumers presented by developments in genetic testing continue to be a focus of the National Health and Medical Research Council (NHMRC).

This NHMRC statement is aimed at health professionals and policy makers. It concerns DNA genetic tests that are used in the diagnosis of medical conditions or in an attempt to provide information about an individual's risk of developing health-related conditions. It distinguishes between the fields of medical genetic testing and direct-to-consumer testing, serves to highlight the factors considered integral to the appropriate delivery and use of health-related genetic testing, and assists health professionals in managing the health and wellbeing of Australians.

Genetic testing, along with the methods by which a genetic test is offered, has both benefits and pitfalls. Individuals may undergo Direct-to-Consumer (DTC) genetic testing out of curiosity for seemingly trivial things, such as to discover more about their ear wax or whether they possess genetic variations for curly hair, allowing greater engagement and appreciation for science and for what makes them unique. However, of potential concern is the packaging of such tests 'for fun' when they also offer findings that affect, or are considered as affecting, clinical outcomes.[1] 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:

- professional involvement and education
- a robust evidence base
- consumer information and support.

The Importance of Professional Involvement and Education

Professional involvement is important in providing individuals and their families with balanced information as most common diseases involve multiple and complex genetic and environmental factors.

Medical genetic testing refers to genetic tests offered in a health care setting, at the request of a qualified health professional. In Australia, most DNA diagnostic and susceptibility genetic testing is conducted in specialised clinics where the results are explained and counselling is offered. Medical genetic tests are conducted in an Australian Standard 4633 (ISO15189) accredited environment, thereby adhering to the appropriate quality management system and requirements that are pertinent for the delivery of medical genetic testing.

Direct-to-consumer (DTC) DNA genetic testing bypasses the medical profession. DTC genetic tests are increasingly available for purchase over the internet through overseas based companies that market their products online. Currently, there are no specific standards and quality systems in place for DTC tests.

Due to the potential to cause unnecessary physical or psychological harm, NHMRC considers that genetic tests should be ordered where tests are reliable and where there is a professionally identified, clinical need. Ideally, such tests would be an approved medical genetic test, conducted in a professional health care setting so that results can be contextualised appropriate to the tested individual's circumstances.

Beyond appropriate levels of technical understanding, medical genetic testing also requires the health professionals involved to give due consideration to the wide range of ethical, legal and social issues that can result from genetic testing. These include the potential implications for the individual being tested, and for their genetic relatives.

In Australia, in vitro diagnostic devices (IVDs) for human genetic testing, including devices intended specifically for the collection of samples for human genetic testing, are classified by the Therapeutic Goods Administration (TGA) as having a high individual risk[2] (Class 3 – see box, right) and are regulated accordingly.[3] However, DTC genetic tests are increasingly available for purchase by Australian consumers from overseas companies through the internet. The remit of the TGA does not extend to regulating devices and their associated testing services that are provided outside of Australia.

Class 3 devices present a moderate public health risk, or high individual risk, where an erroneous result could lead to a patient management decision resulting in a significant detrimental impact on patient outcome. The high individual risk includes consideration of the potential for stress and anxiety resulting from the information and the nature of the possible follow-up measures.

Consequently, the cross-jurisdictional availability of DTC genetic tests, driven by market forces and growing consumer demand, presents a new set of challenges.

The growing demand for DTC genetic tests is motivated by self-directed individuals who are now taking greater responsibility for their own health. While individuals can opt to undertake a health-related genetic test directly through the purchase of a kit from an overseas based company on the internet, receipt of DTC results has mixed outcomes. DTC results might serve to satisfy curiosity or increase interest in the field of genetic science, however they can also raise considerable anxieties and cause confusion.

Access to professional advice throughout the DTC process is often limited or provided indirectly. NHMRC encourages the involvement of health professionals, including general practitioners, clinical geneticists and genetic counsellors in the genetic testing process. Explaining the implications of an identified genetic variation is not always straightforward and requires considerable expertise, particularly as different companies can return different interpretations of the same sample.[4]

To support patients who have had a genetic test, be it in a clinical setting or otherwise, health professionals need to become more aware of the commercialisation of genetic tests and understand what the results mean.[5] Ideally, professional development should be facilitated by the relevant associations, colleges and societies, rather than provided on an *ad hoc* basis.

The Need for Robust Evidence

NHMRC considers that genetic tests should be underpinned by robust evidence. Approximately 20,000 human genes have been identified but only a few are currently tested in clinical practice as medical genetic tests to inform clinical care and/or assist in improving patient outcomes.

Most DTC genetic tests are still research activities that need to be confirmed, and at this time, are of questionable clinical value.[6,7,8,9,10] Not all genetic tests are underpinned by the same level of evidence. International reports on DTC genetic testing have found that consumers can be provided with misleading information that is also of little or no practical use.[11] DTC genetic testing companies have been found to provide information that is contradictory based on the same DNA sample and some companies have also been found to provide disease predictions that conflict with established medical conditions.[8,11]

The Need for Consumer Information and Support

Genetic testing results can have implications for family members due to the familial nature of genetic information. Health professionals have an important role in providing advice and support. NHMRC encourages individuals interested in undertaking a genetic test to exercise caution and consider whether there may be implications for their family members. Interested individuals should consult their general practitioner about the potential implications of genetic testing and to discuss the various options available, including whether there is an actual need for testing. For example, a particular genetic test may not provide clinically significant information and traditional tests (such as body mass index for diabetes) could be more appropriate for determining susceptibility in type 2 diabetes.

The internet has facilitated the exponential growth of the genetic testing market. DTC genetic testing, in particular, has flourished through targeted marketing using this medium. Such tests can be appealing because sample collection can be done at home and access to results by others is limited. However, a consumer's DNA sample is then held by a third party, most likely in an overseas jurisdiction. This is of even greater concern in cases where the DNA of another person is surreptitiously obtained and tested without their consent.

While DTC genetic testing companies have their own privacy policies available online, these can vary and it is not known whether these are read and understood by consumers. Consumers must be aware that protections applicable to Australian residents, which are enforceable for Australian based medical genetic testing, are not enforceable for DTC genetic testing companies with overseas based laboratories. These jurisdictional differences could potentially leave consumers vulnerable should problems arise.

While genetic tests have traditionally been available only in a clinical setting as medical genetic tests, the availability of DTC tests through the internet allows individuals greater personal autonomy. The growing public enthusiasm for DTC genetic testing is encouraging as it suggests a willingness by individuals to engage more actively with own their health and wellbeing. However, the provision of unnecessary and/or inaccurate DTC genetic tests has the potential to produce an expanding cohort of 'worried well' and place unnecessary strain on Australia's health system.[12] Conversely, risks also remain in the inappropriate self-management of susceptibility and conditions. In such cases, an individual may feel empowered by the information they have sought for themselves through undertaking a DTC test. However, the accompanying perception that they are adequately managing any uncovered conditions and/or predispositions (however accurate these may be) may not be true, particularly in instances where the support of a health practitioner is indicated but not offered or sought.

Because it is not possible to regulate many transactions conducted on the internet, NHMRC considers that a more effective approach is to ensure that consumers have realistic expectations of what information genetic tests, both DTC and medical, can provide. This can be achieved through access to accurate information, appropriate support and counselling when needed. NHMRC, through its Human Genetics Advisory Committee, has been actively working in this area.

Addressing the Rise 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 to patients and their families. Health practitioners are increasingly likely to find themselves providing advice on genetics, and perhaps also advising on the utility and/or results of DTC genetic tests. To assist health professionals to best support their patients and consumers in making informed choices, NHMRC has a range of information and guidance products on genetics and genetic testing.

General practitioners can access *Genetics in Family Medicine: The Australian Handbook for General Practitioners* and its accompanying patient factsheets from the NHMRC website.[13] Released in 2007, this national education resource was an Australian Government initiative supported by NHMRC.

NHMRC's *Clinical Utility of Personalised Medicine: Information for Health Professionals*[16], released in 2011, provides an overview of the role of genomics in personalised medicine and its potential to improve health care.

As each individual is both genetically unique and linked to others by their genome, genetic testing is of interest to individuals and to families. For health professionals, this relates to the issue of disclosure, which is pertinent due to legislative changes to the *Privacy Act 1988* (Cth) (see box – right). NHMRC has released Guidelines[14] to assist health practitioners in the private sector with making decisions about the use or disclosure of genetic information. The Guidelines establish when, by whom and in what manner, use or disclosure of genetic information may take place. Approved by the Privacy Commissioner, the Guidelines outline the factors that should be considered when determining whether use or disclosure is necessary to lessen or prevent a serious threat to an individual's life, health or safety.

Did you know?

As a result of legislative changes to the *Privacy Act 1988* (Cth), Australian health practitioners in the private sector are now permitted to use or disclose patients' genetic information, without their consent, in circumstances where they reasonably believe that doing so is necessary to lessen or prevent a serious threat to the life, health or safety of a genetic relative.

NHMRC's *Medical Genetic Testing: Information for Health Professionals*[15], highlights the ethical, legal and social issues resulting from genetic testing and supports health professionals in:

- assisting patients when considering genetic testing
- ordering the appropriate genetic test
- interpreting the test result in the context of clinical decision making
- providing follow-up care and support to the patient and family.

Accompanying this NHMRC Statement on DNA genetic testing is the online release of *Medical Genetic Testing: Health Information for You and Your Family*, which is a complementary information resource on medical genetic testing for consumers.

Both *Medical Genetic Testing: Information for Health Professionals* and the complementary consumer information resource also feature commentary on the issue of DTC genetic testing. Given the wide range of issues that DTC genetic testing presents, NHMRC considers that a multi-faceted approach, including information, education and regulation should be taken in relation to DTC testing.[16]

In addition to the development of informational resources, NHMRC also collaborated with the UK Human Genetics Commission in the development of a *Common Framework of Principles for DTC Genetic Testing Services*.[17] The Principles, released in August 2010, aim to promote consistency and high standards in the provision of DTC genetic tests at an international level and facilitate the progression of country-specific codes of practice,[18] though it should be noted that these are not legally enforceable.

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.

List of key NHMRC products on genetics

- Direct-to-consumer (DTC) DNA genetic testing: An information resource for consumers
- Direct-to-consumer (DTC) DNA genetic tests
- Medical Genetic Testing: Health information for you and your family
- Medical Genetic Testing: Information for Health Professionals
- 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
- Genetics in Family Medicine: The Australian Handbook for General Practitioners
- Clinical Utility of Personalised Medicine: Information for health professionals
- DNA Genetic Testing in the Australian Context: A Statement from the National Health and Medical Research Council (NHMRC)

These resources are available from www.nhmrc.gov.au

How was this statement developed?

NHMRC developed this Statement in consultation with its Human Genetics Advisory Committee 2010–12, with input from the Therapeutic Goods Administration on matters regarding regulation. The Statement was recommended to the CEO for publication by the Council of NHMRC in 2011.

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