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RCPA
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Position Statement

Subject:	**Genetic Tests that are Marketed Directly to Consumers**
Approval Date:	July 2013 (RCPA approved) September 2014 (HGSA approved), March 2 017, August 2018
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Review By:	Genetic Advisory Committee
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Genetic factors are recognised to play a role in almost every aspect of health and disease. Our understanding of these genetic influences is increasing, as is our ability to test for them.
Many people are interested in using genetic tests. Genetic tests can be utilised for medical and non-medical purposes, to determine ancestry, predict medication sensitivity, predict the likelihood of developing particular diseases and of passing this predisposition to their children, and testing for acquired genetic changes in the oncology setting which may help determine prognosis or treatment.

Genetic testing for Medical purposes
Genetic test results may have significant health implications, not only for the individual being tested, but potentially also for their relatives when testing for heritable genetic changes.

The Royal College of Pathologists of Australasia (RCPA) strongly advocates that complex medical tests always be requested by, and after discussion with, an experienced medical practitioner or other appropriately qualified health practitioner. This approach applies to all medical tests. It is particularly relevant for complex genetic tests that predict the medical future of a child. It is not appropriate for genetic tests that deal with significant clinical issues to be marketed directly to patients.
(RCPA Media Release on DTC - July 2018)

Furthermore, some genetic tests have the potential for complex outcomes, with profound implications for some individuals. NPAAC provides guidance on the classification of genetic tests into two levels: Level 1 (“standard”) tests, and Level 2 (“potential to lead to complex clinical issues”) tests. Level 2 genetic tests require specific written consent, and are associated with counseling issues requiring the involvement of appropriately experienced medical specialists or individuals specifically trained in the provision of genetic counselling (e.g. genetic counsellors) ..
(NPAAC Requirements for Medical Testing of Human Nucleic Acids 2013)

Genetic testing for non-Medical purposes
Some genetic tests are also available for non-health purposes, such as tests for distant ancestry or for lifestyle or behavioural traits (see category table above). Where these categories of tests have no medical implications, there is no requirement for a medical or healthcare practitioner to be necessarily involved, and such tests could potentially be accessed through a variety of routes, some of which potentially might be Direct-to-Consumer (DTC).
(NHMRC Guide on medical genetic testing
http://www.nhmrc.gov.au/_files_nhmrc/publications/attachments/e99.pdf

NHMRC Statement on DNA genetic testing in the Australian context
https://www.nhmrc.gov.au/_files_nhmrc/publications/attachments/ps0002_dna_genetic_testing_consumers.pdf

NHMRC Guide on discussing DTC genetic testing with patients
https://www.nhmrc.gov.au/_files_nhmrc/publications/attachments/g9_direct_to_consumer_genetic_testing_nhmrc_statement_141208.pdf

NPAAC Genetic DTC Guiding Principles (May 2014)
https://www.health.gov.au/internet/main/publishing.nsf/content/F57C68E5E41DC946CA257EEC000396F2/$File/Genetic%20DTC%20Guiding%20Principles%20-%20May%202014.pdf

Categories of Genetic Testing
Genetic tests may be categorised to help in discussion of which tests are, or are not, appropriate for access through a DTC route. The following table (adapted from the UK Human Genetics Commission) is an example of broad categories of genetic tests:

	Broad category of genetic test	**Description**
1	Diagnostic tests	Tests intended to diagnose a medical condition in a person with symptoms and/or signs.
2	Pre-symptomatic tests	Tests intended to predict that an asymptomatic person has a high probability of developing a condition, for example, mutation testing in some autosomal dominant single – gene disorders. This is sometimes referred to as predictive testing.
3	Carrier testing (including reproductive and pre-conception testing)	Tests intended to show that a person is a carrier of a condition, so that although they are not themselves affected, there is a risk they may have affected children with another carrier.
4	Pharmacogenetic/Pharmacogenomic tests	Tests intended to predict the response profile of an individual to a drug or course of therapy.
5	Susceptibility/Pre-dispositional health tests	Tests intended to provide an indication of the absolute lifetime risk and/or relative risk of an individual developing a condition compared with the general population.
6	Lifestyle/behavioural tests	Tests intended to provide information about an individual's: • behavioural propensities • performance capacities (physical or cognitive) or • response to certain environmental conditions and that are designed to assist the individual to modify the outcomes of any of these by elective changes in behaviour (not including the administration of prescribed medicines).
7	Nutrogenetic tests	Tests intended to provide information about how an individual metabolises nutrients.
8	Phenotype tests	Tests intended to provide information about how an individual's phenotype is conditioned by their genotype for example, tests that indicate the genetic basis of a person's eye colour.

	Broad category of genetic test	Description
9	Genetic relatedness tests	Tests intended to determine or provide information about a genetic relationship, including paternity and maternity.
10	Ancestry tests	Tests intended to provide information about an individual's relatedness to a certain ancestor or ancestral group and /or how much of an individual's genome is likely to have been inherited from ancestors from particular geographical areas or ethnic groups.
11	Genetic matching	Tests intended to determine the likelihood that an individual is the source of a sample of DNA recovered from a secondary object or material.

Table adapted from: (UK Human Genetics Commission: "A Common Framework of Principles for direct-to-consumer genetic testing services: *http://webarchive.nationalarchives.gov.uk/20121102202005/http://hgc.gov.uk/client/document.asp?DocId=214&CAtegoryId=3*

Using the above table as an example, categories 1-5 should be regarded as categories of "Genetic Testing for Medical Purposes", categories 6-10 could be regarded as Genetic Testing for non-Medical Purposes" and category 11 as an example of a test for non-Medical purposes that could never be regarded as a potential DTC test.

It must be specifically noted that labelling a medical test as "For informational purposes only" (or similar wording) does not change its category of testing from that of a complex medical test, to a lesser non-medical category. *It is the inherent nature of the genetic test that determines its categorisation, not the labelling that the provider places on it.*

Important considerations for all Genetic Tests

For genetic tests that are used for any medical purpose (see categorisation table above), it is a legal requirement in Australia that most tests *must* be performed in a NATA/RCPA accredited laboratory. This ensures that the results are analytically correct and meet appropriate quality standards, and that the test meets criteria for scientific validity. It also ensures that there is appropriate clinical supervision and oversight of the testing process and its interpretation.

For other genetic tests that may not necessarily be used for a medical purpose, it is strongly recommended that such tests *should also* be performed in an accredited laboratory, for the same reasons of ensuring appropriate standards of analytical accuracy and quality.

Testing performed in an accredited pathology laboratory in Australia also ensures that the laboratory observes standards in relation to protecting patient privacy and confidentiality.

Note that population screening programs are not considered DTC, even where professional consultation takes place only after testing, provided the following conditions are met:

- The program is undertaken within an appropriate medical supervision and governance framework,
- Testing is performed in accredited laboratories, and
- Positive screening test results are reported by a qualified healthcare professional, with an option to be relayed to the participant's usual medical practitioner for further management.

Important additional considerations for all Direct-to-Consumer Tests

Many laboratories offering DTC testing are not necessarily accredited to medical standards. Some laboratories are also based overseas and are not bound by Australian consumer protection laws. While this may not be relevant to the purpose of the tests being offered, it does mean that the laboratory might not be obliged to observe some of the customary safeguards medical laboratories in Australia.

It is strongly recommended that the full “Privacy Policy” (however named) and “Terms and Conditions” of a Direct-to-Consumer laboratory service be carefully read and understood *before providing any sample for testing*. In particular, some of these laboratories reserve the right to release, forward or even sell samples or genetic information to external organisations. Once genetic information has been released to external parties, it is not usually possible to reverse or recover this information and it may have privacy consequences both for the individual and their relatives.

It is also recommended that for non-medical testing, scientific claims are reviewed for plausibility. While some testing laboratories offering non-medical genetic services (such as Ancestry testing) use techniques based on sound scientific principles, others offering testing to predict (for example) physical beauty, athleticism, intelligence or romantic compatibility should have their scientific validity carefully considered before accepting their claims at face value. As already noted, consumer protection and truth-in-advertising laws may not necessarily apply for DTC services offered from overseas through sources such as the Internet.