



# **The Provision of Direct to Consumer Genetic Tests**

***Guiding Principles for Providers***

**May 2014**

## **The Provision of Direct-to-Consumer Genetic Tests; Guiding Principles for Providers**

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The National Pathology Accreditation Advisory Council (NPAAC) was established in 1979 to consider and make recommendations to the Australian, state and territory governments on matters related to the accreditation of pathology laboratories and the introduction and maintenance of uniform standards of practice in pathology laboratories throughout Australia. A function of NPAAC is to formulate standards and initiate and promote guidelines and education programs about pathology tests.

Publications produced by NPAAC are issued as accreditation material to provide guidance to laboratories and accrediting agencies about minimum standards considered acceptable for good laboratory practice.

Failure to meet these minimum standards may pose a risk to public health and patient safety.

# The National Pathology Accreditation Advisory Council

The National Pathology Accreditation Advisory Council (NPAAC), as outlined in the *Constitution of the National Pathology Accreditation Advisory Council Order No. 1 of 1997* (the Order in Council) is charged with the role of providing advice to the Commonwealth and States and Territories on a range of accreditation issues relating to the provision of pathology services in Australia with the aim of ensuring the quality of the services provided and the safety of the clients receiving those services. The matters that Council addresses include:

- developing policy for the accreditation of pathology laboratories;
- introducing and maintaining uniform standards of practice in pathology throughout Australia;
- adopting coordinated legislation and administrative action in providing pathology services.

Medical testing occurring outside the normal doctor-patient relationship and/or outside the regulated pathology accreditation system has the potential to place patients at risk. NPAAC continues to support the need for strategies to address any risks associated with direct to consumer testing (DTC), and, in particular, direct to consumer genetic testing (DTC-GT). While many providers may argue that the genetic testing that they are offering is for information only and is not health advice, in many cases it is still health and disease related and may result in either the imparting of needless concern or a false sense of security to the client. The provider's processes should match the consequences that would reasonably arise from the consumer's perception of the test result.

With this in mind, this document outlines what is considered by NPAAC to be the guiding principles that should be followed by DTC-GT providers in order to ensure the safety of their clients who seek this type of testing.

Further information about NPAAC's function and publications may be found on the NPAAC website.<sup>1</sup>

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1     *The Provision of Direct to Consumer Genetic Tests: Guiding Principles for Providers*

## Abbreviations

AS	Australian Standard
DTC-GT	Direct-to-Consumer Genetic Testing
DNA	Deoxyribonucleic acid
EASAC	European Academies Science Advisory Council
FEAM	Federation of European Academies of Medicine
HGC	Human Genetics Commission (UK)
HGSA	Human Genetics Society of Australasia
HREC	Human Research Ethics Committee
ISO	International Organization for Standardization
IVD	In Vitro Diagnostic Medical Device
MPS	Massively Parallel Sequencing
NATA	National Association of Testing Authorities
NGS	Next Generation Sequencing
NHMRC	National Health and Medical Research Council
NPAAC	National Pathology Accreditation Advisory Council
OAIC	Office of the Australian Information Commissioner
RCPA	Royal College of Pathologists of Australasia
SNP	Single Nucleotide Polymorphism
TGA	Therapeutic Goods Administration
WGS	Whole Genome Sequencing

# Summary of Recommendations

## **Recommendation 1**

*The provider's laboratory should be accredited to either ISO 15189 or ISO 17025 and should be enrolled in relevant external proficiency programs.*

*The status of accreditation should be disclosed in all documentation.*

## **Recommendation 2**

*The provider should seek clarification from the TGA to ascertain if the DTC service being provided is subject to the IVD regulatory framework.*

## **Recommendation 3**

*The provider should never offer DTC services that test for*

- *High penetrance genotypes associated with serious disorders*
- *Prenatal testing*
- *Preconception carrier screening*
- *Carrier testing in children.*

## **Recommendation 4**

*The provider should not offer DTC nutrigenetic testing services unless extensive validation of both the test and its associated nutritional intervention can be documented.*

## **Recommendation 5**

*The provider should not offer DTC pharmacogenetic testing without strongly advising the consumer not to initiate or alter the dosage of any existing medication, on the basis of the test results, without first consulting a relevant medical practitioner.*

## **Recommendation 6**

*The provider should always ensure that all promotional and technical claims for any DTC-GT are clear, accurate, unbiased, not overstated, and supported by relevant peer reviewed published evidence.*

## **Recommendation 7**

*The provider should supply to the consumer accurate, appropriate, easy to understand information about the following before obtaining consent:*

- *genetics in general*
- *specific tests being offered*
- *information about the scope, accuracy and limitations, including analytical and clinical validity and utility, of the test*
- *information about likely outcomes of the genetic test and the decisions that the consumer may face after taking the test*
- *counselling both pre and post test where appropriate*
- *how the results will be provided and explained*

- *information about the long term storage of test samples and personal records*
- *that the results might reveal information about genetic relationships*
- *that the results might have implications in relation to purchasing some types of insurance e.g. life insurance*
- *that taking DNA from another person without their consent is unethical and may even be a criminal offence in some jurisdictions*
- *what will happen to the consumer's biological samples and personal and genetic data if the company ceases trading.*

#### **Recommendation 8**

*If the provider intends to use the consumer's biological sample or personal data for research purposes then consent must be obtained from the consumer, and the consumer should be informed about:*

- *the approval provided by an appropriate research ethics committee*
- *whether the biological samples and associated data will be transferred to a biobank or database, and the subsequent security measures to protect the sample and data.*

#### **Recommendation 9**

*Where appropriate, the test provider should supply consumers at the time of testing or subsequently, with information about health professionals who are able to offer further advice or support and genetic counselling.*

#### **Recommendation 10**

*The provider should require consumers to sign a statement confirming that they give their informed consent to the specific genetic tests to be undertaken. Documentary evidence of this informed consent should be retained for the duration of storage of the consumer's biological samples and records.*

#### **Recommendation 11**

*The provider should not release biological samples or records containing personal data and genetic information that can be linked to the consumer to any third party, including insurance companies, health professionals or solicitors, except with the specific prior consent of the consumer on whom the test was performed.*

#### **Recommendation 12**

*If the provider ceases trading, they should dispose of personal and genetic data securely or provide for transfer of responsibilities in accordance with the terms of consent given by the consumer.*

#### **Recommendation 13**

*The provider should ensure that the interpretation of genetic test results is accurate and comprehensible to the consumer, and where test results are provided in the form of a risk assessment, it should be based upon robustly evaluated algorithms, and should make clear the distinction between relative risk and absolute risk. There should also be regular review of the available evidence upon which the interpretation is based.*



**Recommendation 14**

*The test provider should not overstate the value or significance of the results of the genetic test when providing the test results.*

**Recommendation 15**

*Where there are two or more providers, one or more performing the testing, and one or more performing the interpretation, then each should comply with these guidelines where they are relevant to the aspect of the service each has provided.*

**Recommendation 16**

*The provider should ensure that the complaints process is known to the consumer and that complaints are dealt with in a reasonable time-period. Consumers should be informed promptly of the outcome of any complaint.*

**Recommendation 17**

*The provider should not offer direct-to-consumer genomic testing, such as Whole Genome Sequencing (WGS), unless the provider has the appropriate medical infrastructure within their organisation to be able to deal appropriately with the medical ethical issues that arise as a consequence of such testing.*

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# 1. Scope

**This document is specifically designed as a guidance document for providers of DTC-GT who wish to operate within Australia.**

Whilst it is not primarily aimed at consumers, it may also help inform and guide the expectations of potential consumers of DTC-GT. In the first instance consumers are directed to the information resource on Direct-to-consumer DNA Genetic Testing published by the National Health and Medical research Council (NHMRC).<sup>2</sup> This current document focuses particularly upon the provider responsibilities in DTC-GT including all of the key issues listed below that are associated with the provision of such testing.

This guidance covers:

- The need for demonstrated competence through accreditation
- Regulatory information about in-house in vitro diagnostic medical devices (IVDs), including requirements from regulatory bodies such as TGA, NPAAC, and NATA/RCPA, and their use in health related genetic testing
- Defining the scope of what might be included in DTC-GT, and more importantly, what should be excluded
- General principles that should apply in the performance of DTC-GT and the relationship, both professional and ethical, between the DTC-GT provider and their clients, including:
  - Establishing the analytical and clinical validity of any health-related DTC-GT that may be offered
  - Marketing, advertising and information for prospective consumers
  - Pre-test consent and counselling
  - Post-test counselling and support
  - Sample handling and laboratory processes
  - Interpretation and provision of test results
  - Data protection
  - Ongoing customer support and handling of complaints
- Specific principles that may apply to Massively Parallel Sequencing (MPS)

## 2. Introduction

These guidelines have been produced by the National Pathology Accreditation Advisory Council (NPAAC) in order to address the regulatory gap that currently exists in Australia in the area of direct-to-consumer genetic testing (DTC-GT). These guidelines rely heavily upon “The Common Framework of Principles” for DTC-GT developed by the UK Human Genetics Commission in 2010.<sup>3</sup>

In considering DTC-GT, there are a number of attributes of a test that need to be addressed. The first is the **analytical validity** of the test i.e. the ability of the test result to reflect the actual DNA sequence of the patient. The second consideration is the **clinical validity** of the test i.e. the strength of association between the test result and the clinical state or disease under consideration. The third consideration is the **utility** of the test result i.e. whether the test result provides useable information that is not already available to the consumer being tested. The final consideration is any **ethical, legal, or social consideration** that might flow from the test result.

DTC-GT providers often claim to be able to advise clients about their risks of developing common diseases such as rheumatoid arthritis, cancers and heart attacks. The Nuffield Council on Bioethics acknowledges that such information may provide reassurance or enable people to take preventative action, but they also point out that there are a number of potential disadvantages with such testing<sup>4</sup>:

- The test results can be unreliable and difficult to interpret.
- The clinical validity of the test may be uncertain and not replicated by tests purporting to address the same clinical issue.
- The utility of the test result may be minimal or even dangerous. For example, a result which indicates a reduced risk of disease may lead to unwarranted complacency in lifestyle.
- Conversely, results which indicate an increased risk of disease may be upsetting, particularly if effective interventions are not available. Such results may also lead to patients undertaking unwarranted interventions.
- There is potential for misuse of personal genetic information, such as the unauthorized release of test results or testing a person without consent.
- People may seek unnecessary further tests or advice from their doctor.

The National Health and Medical Research Council (NHMRC), in its consumer information on DTC-GT, makes very similar points to these.<sup>2</sup>

### 3. Regulatory Issues

#### 3.1 Accreditation

The analysis of biological samples, for genetic testing services, should only be provided by competent laboratories. Competence can be established by accreditation to the Australian Standard AS ISO 15189, or to the International Organization for Standardization (ISO) Standards ISO 15189 or ISO/IEC 17025.<sup>5-7</sup>

The National Pathology Accreditation Advisory Council (NPAAC)<sup>1</sup> provides a suite of standards relevant to all aspects of pathology testing, including human molecular genetic testing, with the following standards:

- *Requirements for Medical Testing of Human Nucleic Acids (2013)*
- *Requirements for the development and Use of In-house In Vitro Diagnostic Devices (2007)*
- *Requirements for Participation in External Quality Assessment (2013)*

The requirements promulgated by NPAAC provide appropriate guidance for laboratories in Australia seeking to provide DTC-GT. However, it should be noted that such a laboratory cannot be accredited specifically to NPAAC requirements, as they currently explicitly exclude provision of DTC-GT. Such laboratories should, however, seek accreditation to ISO 15189 or ISO 17025.

Achievement of laboratory accreditation requires monitoring the quality of laboratory performance through external proficiency testing.

Laboratories should have policies in place to apply corrective measures if their performance falls outside of parameters determined by the laboratory's quality assurance programs.

The laboratory personnel should have appropriate professional qualifications that meet recognised standards, underpinned by education and training, to assure competence in laboratory procedures in the provision of genetic tests.

#### **Recommendation 1**

***The provider's laboratory should be accredited to either AS ISO 15189 or to ISO 15189 or ISO 17025 and should be enrolled in relevant external proficiency programs. The status of accreditation should be disclosed in all documentation.***

### 3.2 Is the test an in-vitro diagnostic medical device?

The first question that must be addressed is whether the tests being offered should be classed as in-vitro diagnostic medical devices (IVDs). If this is the case the tests becomes subject to the requirements of the *Therapeutic Goods (Medical Devices) Regulations 2002* administered by the Therapeutic Goods Administration (TGA).

The TGA operates under the Therapeutic Goods Act (the Act) and IVDs are a class of 'Therapeutic Good'. The *Therapeutic Goods Act 1989* defines a therapeutic good as a good which is represented in any way to be, or is likely to be taken to be, for therapeutic use.<sup>8</sup>

Therapeutic use is defined as use in or in connection with:

- preventing, diagnosing, curing or alleviating a disease, ailment, defect or injury;
- influencing, inhibiting or modifying a physiological process;
- testing the susceptibility of persons to a disease or ailment;
- influencing, controlling or preventing conception;
- testing for pregnancy; or
- replacement or modification of parts of the anatomy.

The TGA website defines an IVD as follows<sup>9</sup>:

“A medical device is an *in vitro* diagnostic medical device (IVD) if it is a reagent, calibrator, control material, kit, specimen receptacle, software, instrument, apparatus, equipment or system, whether used alone or in combination with other diagnostic goods for *in vitro* use. It must be intended by the manufacturer to be used *in vitro* for the examination of specimens derived from the human body, **solely or principally for the purpose of giving information about a physiological or pathological state, a congenital abnormality** or to determine safety and compatibility with a potential recipient, or **to monitor therapeutic measures**.”

It is important to note that definition of a therapeutic good is not determined solely by the provider's intentions. It is also important to note that the above TGA definition of an IVD is quite clear in that an IVD is “used **solely or principally for the purpose of giving information about a physiological or pathological state...**”

It would seem to be an inescapable conclusion that testing used in DTC-GT is absolutely designed to give information about a physiological state, and that the provider of such a test **must** seek clarification from the TGA as to whether any such test being provided is subject to the IVD regulatory framework in Australia before offering such testing in Australia.

The legislation, set out in the *Therapeutic Goods (Excluded Purposes) Specification 2010*, prohibits supply in Australia of self-testing IVDs for genetic testing to determine the presence of, or predict susceptibility to, disease in humans, except where the device is also to be used for any of the following purposes:

- a) for testing for a disease or condition as part of a public health screening program sponsored by the government of the Commonwealth or a State or Territory;
- b) for self-testing to monitor a diagnosed disease or condition;
- c) for export only.

An IVD for self-testing is defined in the Act as an IVD medical device intended to be used:

- a) in the home or similar environment by a lay person;
- b) in the collection of a sample by a lay person and, if that sample is tested by another person, the results are returned directly to the person from whom the sample was taken without direct supervision of a health professional who has formal training in a medical field or discipline to which the self-testing relates.

The TGA can be contacted by telephone on 1800 020 653 and select option 3, by email to [info@tga.gov.au](mailto:info@tga.gov.au), or by writing to the following address: TGA, PO Box 100, Woden ACT 2606, Australia.

### **Recommendation 2**

***The provider should seek clarification from the TGA to ascertain if the DTC service being provided is subject to the IVD regulatory framework.***

## **3.3 Testing for mutations which carry a high risk of disease**

The ethical and regulatory implications of DTC-GT will depend on the medical implications of the test result. A test for a mutation that is associated with high risk of disease is associated with greater regulatory and ethical concerns than a test which carries few if any medical consequences.

In 2010, the Nuffield Council on Bioethics noted that DTC-GT for disease susceptibility, while possibly giving reassurance or enabling people to take preventative action, had some significant potential problems.<sup>4</sup> Two years later the European Academies Science Advisory Council- Federation of European Academies of Medicine (EASAC-FEAM) working group determined that all kinds of human genetic testing require an appropriate and relevant level of professional advice. Consequently the EASAC-FEAM working group<sup>10,11</sup> considered that the following should be currently **excluded** from the services offered by DTC-GT companies:

- High-penetrance genotypes, including monogenic disorders, that are associated with serious disease
- Pre-natal testing
- Preconception carrier screening
- Carrier testing in children.

They further considered that DTC-GT companies should be **discouraged** from offering the following:

- Nutrigenomics
- Pharmacogenetics.

**Recommendation 3**

***The provider should never offer DTC services that test for:***

- ***High penetrance genotypes associated with serious disorders***
- ***Prenatal testing***
- ***Preconception carrier screening***
- ***Carrier testing in children.***

**Recommendation 4**

***The provider should not offer DTC nutrigenetic testing services unless extensive validation of both the test and its associated nutritional intervention can be documented.***

**Recommendation 5**

***The provider should not offer DTC pharmacogenetic testing without strongly advising the consumer not to initiate or alter the dosage of any existing medication on the basis of the test results without first consulting a relevant medical practitioner.***



## 4. Marketing and Advertising

The UK Human Genetics Commission in 2010<sup>3</sup> produced principles for the provision of direct-to-consumer genetic testing services. They have developed the following code to cover marketing and advertising:

- Where relevant, the test provider should comply with any legislation or voluntary codes for advertising of medical tests, including genetic tests or other clinical services and they should also comply with more general guidance (including legal guidance) covering consumer advertising.
- Promotional and technical claims for genetic tests should accurately describe both the characteristics and the limitations of the tests offered, and the test provider should not overstate the utility of a genetic test.
- Where a claim is made about the clinical validity of a genetic test, the claim should be supported by the citation of relevant evidence published in peer reviewed scientific literature and the test provider should give standard references to this literature.
- The test provider should be aware of the risk of bias when quoting evidence and ensure that evidence is presented transparently with reference to the criteria used to include and/or exclude published literature when this is cited as evidence of the applicability or effectiveness of the test.
- Information about tests which are available only in the context of a consultation with a health professional or are only provided to consumers with both individualised pre- and post-test counselling should make it clear that tests are available only in that context.

The Therapeutic Goods Advertising Code, administered by the TGA, ensures that the marketing and advertising of therapeutic goods is conducted in a manner that promotes the quality use of the product, is socially responsible and does not mislead or deceive the consumer.

### **Recommendation 6**

***The provider should always ensure that all promotional and technical claims for any DTC-GT are clear, accurate, unbiased, not overstated, and supported by relevant peer-reviewed published evidence.***

## 5. Provision of Information to Prospective Consumers

The HGC, in their Principles document, have recommended that the following information should be provided to prospective consumers:

- It is important for providers of DTC-GT to supply easily understood, accurate, appropriate and adequate information before obtaining consent for genetic tests:
  - general information about genetics to enable a consumer to understand the scientific basis of genetic testing, the role of genes in health and disease, conditioning phenotypes, and the technologies applied to generate the knowledge
  - a clear explanation of the relative roles of genetics, environmental factors, lifestyle choices and other factors in determining health, disease and phenotype
  - specific information about genetic tests offered
  - information about counselling offered in connection with the test including whether counselling is included in the cost of the test and for what costs the consumer will be liable if they withdraw following pre-test counselling
  - information about the presentation of results in statistical form, such as relative and absolute risk assessments or likelihood of inclusion or exclusion as a genetic relative, so that the consumer can understand test results that are provided
  - information about measures taken by the test provider and laboratories to ensure the confidentiality of personal records and security of biological samples
  - information about the maximum period of storage of the biological sample and personal records, and procedures for storage, transfer and disposal of biological samples and personal records
  - information about whether biological samples may be used for any secondary purposes, such as additional research purposes, and whether personal genetic information may be passed to third parties and, if so, under what conditions and to whom
  - information about procedures for handling and resolving consumer complaints
  - information about the manner in which the test results will be provided and, if applicable to the genetic test, information about the requirement for pre- and post-test counselling
  - a statement that the results of the test might be able to reveal information about genetic relationships
  - a statement that the results of the genetic test might have implications when purchasing some forms of insurance (such as life insurance, disability insurance, and travel insurance)
  - a statement that third parties, such as law enforcement agencies, can have access to consumers' biological samples without their consent if laws exist that would permit this
  - information about specific procedures that might need to be followed if the test is to be used for official purposes, such as certain chains of evidence

- that might need to be maintained in some jurisdictions, if the test is to be used in the courts of law
  - a statement that taking DNA from someone else without their consent is generally ethically inappropriate and is a criminal offence in some jurisdictions
  - information about what will happen to consumers' biological samples, and personal and genetic data, if the company ceases trading.
- The test provider should provide information to consumers about the degree of association between a genetic variant and a disease, condition or trait for each genetic test that they offer in a format that is easy to understand.
- The test provider should make available to consumers information about the scope, accuracy, and limitations of the test. Information about the analytical and clinical validity of each of the genetic markers used in the test should be made available. Other factors, such as behaviour or environmental conditions that will play a role in determining the development of the condition or trait under investigation, should also be listed.
- The test provider should provide information about the likely outcomes of the genetic test and the decisions that a consumer may face after taking the test. They should also identify prospectively any likely further investigations that a consumer or member of their family may wish to pursue after receiving the test results.
- If a test provider intends to use a consumer's biological samples and/or associated personal or genetic data for secondary purposes, such as research, the consumer should be informed whether the research has been approved by a Human Research Ethics Committee (HREC) or other competent authority, whether the biological sample and data will be transferred to or kept in a biobank or database, and about measures to ensure the security of the sample and data. The consumer must be informed of any risks or potential benefits associated with participating in the research and whether they will receive feedback on research findings that relate to them. Consent must be received from the consumer for the research to be performed unless such consent has been waived by a formally constituted HREC duly registered with the NHMRC except where the research uses collections of non-identifiable data and involves negligible risk.<sup>12</sup>
- If a test provider intends to use the results of a genetic test to make a recommendation to a consumer to purchase a therapeutic product, such as a nutritional agent or supplement, the test provider should make available all or any information about the link between the genetic test result and the efficacy of the indicated product. The test provider should also provide information about other lifestyle choices and behavioural modifications that are known to have a preventative or therapeutic value in relation to the trait linked to the genetic markers tested and whether the consumer can purchase the recommended therapeutic product elsewhere.
- The provider should declare any conflict of interest arising as a result of any relationship between the DTC-GT provider and any drug or medical product supplier, or any nutrigenomic product supplier.
- Where the test result indicates that the consumer may benefit from an alteration in the dosage of a medicine, or from an alternative medicine to one currently being

taken, the test provider should make available comprehensive information about the link between the genetic test result and the metabolism of the indicated medicines in terms suitable for the consumer.

- The test provider should make it clear how and whether a consumer can receive updated test results as part of the service they supply, and how this would be provided.
- Where appropriate, outside the context of a consultation with a suitably qualified health professional, the test provider should inform consumers about recommendations or known actions that may help the consumer to take informed decisions about their health or welfare in the light of the test results, including informed interaction with the health care system.
- Where appropriate, the test provider should supply consumers with information about appropriately qualified health professionals who are able to offer further advice or support.

#### **Recommendation 7**

***The provider should supply to the consumer accurate, appropriate, easy to understand information about the following before obtaining consent:***

- ***genetics in general***
- ***specific tests being offered***
- ***information about the scope, accuracy and limitations, including analytical and clinical validity and utility, of the test***
- ***information about likely outcomes of the genetic test and the decisions that the consumer may face after taking the test.***
- ***counselling both pre and post test where appropriate***
- ***how the results will be provided and explained***
- ***information about the long term storage of test samples and personal records***
- ***that the results might reveal information about genetic relationships***
- ***that the results might have implications in relation to purchasing some types of insurance e.g. life insurance***
- ***that taking DNA from another person without their consent is unethical and may be a criminal offence in some jurisdictions.***
- ***what will happen to the consumer's biological samples and personal and genetic data if the company ceases trading.***

#### **Recommendation 8**

***If the provider intends to use the consumer's biological sample or personal data for any secondary purpose, including research, then consent must be obtained from the consumer, and the consumer should be informed about:***

- ***the approval provided by an appropriate research ethics committee***
- ***whether the biological samples and associated data will be transferred to a biobank or database, and the subsequent security measures to protect the sample and data.***

## 6. Counselling and Support

Where the test is a genetic test in the context of inherited or heritable disorders, that test should only be provided to consumers who are given a suitable opportunity to receive pre- and post-test genetic counselling. Note that genetic counselling is an accredited healthcare profession in Australia.

The counsellor should have the appropriate skills and competencies and be certified by the relevant professional body in Australia, the Human Genetics Society of Australasia (HGSA).<sup>13</sup> The genetic counsellor should be independent of the DTC-GT provider.

After receiving the information provided in part 4 and receiving any offer of pre-test counselling, consumers should have the opportunity to cancel purchase of the test without incurring further costs relating to the test.

### **Recommendation 9**

***Where appropriate, the test provider should supply consumers at the time of testing or subsequently, with information about health professionals who are able to offer further advice or support and genetic counselling.***

## 7. Consent

A genetic test should be carried out only after the consumer concerned has given free and informed consent. Informed consent can only be provided when a consumer has received sufficient relevant information about the genetic test to enable them to understand the risks, benefits, limitations and implications (medical, social, financial and legal) of the genetic test.

The provider should have processes that can demonstrate that the consumer's consent is informed. Simple signed forms, with no evidence as to whether the consumer has understood the possibly technical wording of this consent, are not sufficient to demonstrate consent is informed.

The test provider should take all reasonable steps to ensure that a biological specimen provided for testing was obtained from the consumer identified as the sample provider.

The test provider should require consumers to sign a statement confirming that they give their informed consent to the specific genetic tests to be undertaken on their biological material.

The test provider should retain documentary evidence of the provision of informed consent by the consumer for the duration of storage of the consumers' biological samples and personal records.

Separate, specific, informed consent should be required by the test provider if the test provider wishes to perform further tests that are not covered by the original consent or if biological samples are to be stored by the test provider after the consumer has been provided with the genetic test results. Likewise, separate informed consent should be required by the test provider before biological samples are used for any secondary purposes, e.g. research, or before any third party is permitted access to biological samples.

### **Recommendation 10**

***The provider should require consumers to sign a statement confirming that they give their informed consent to the specific genetic tests to be undertaken. Documentary evidence of this informed consent should be retained for the duration of storage of the consumer's biological samples and records.***

## 8. Data Protection

Genetic information is sensitive personal data and requires the highest level of security and confidentiality. There are explicit requirements under the Federal *Privacy Act* regarding the management of genetic information by organisations and individuals in the private sector. These can be found in the *National Privacy Principles*. These are available from the Office of the Australian Information Commissioner (OAIC) on [the OAIC website](#). Detailed explanatory guidelines regarding use and disclosure of genetic data were published by the National Health and Medical Research Council (NHMRC) in December 2009.<sup>14</sup>

The test provider and laboratories must not release biological samples or records containing personal data and genetic information that can be linked to an identifiable person to any third party without the prior specific consent of the person to whom they relate, unless required to do so in accordance with legislation for example, pursuant to a Court order.

If a test provider ceases trading, they must dispose of personal and genetic data securely or provide for transfer of responsibilities in accordance with the terms of consent given by the consumer.

### **Recommendation 11**

***The provider should not release biological samples or records containing personal data and genetic information that can be linked to the consumer to any third party, including insurance companies, health professionals or solicitors, except with the specific prior consent of the consumer on whom the test was performed or in accordance with a court order.***

### **Recommendation 12**

***If the provider ceases trading, they should dispose of personal and genetic data securely or provide for transfer of responsibilities in accordance with the terms of consent given by the consumer.***

## 9. Test Result Interpretation

A qualified professional should be available and be responsible for the interpretation of genetic test results, and should ensure that the interpretation of genetic test results is accurate, and take steps to ensure that these results are comprehensible to the consumer.

Where genetic test results are provided in the form of a risk assessment, the risk assessment should be based on robustly evaluated algorithms. Standard statistical methodologies should be used to convert risks reported in scientific literature to the risk of a disease, condition or trait for an individual compared with the general population risk over a defined period e.g. 10 years, as well as lifetime risks or lifetime incidences. Results should make clear the distinction between relative risks and absolute risk.

Test providers should regularly review the available evidence on which their interpretation is based.

### **Recommendation 13**

***The provider should ensure that the interpretation of genetic test results is accurate and comprehensible to the consumer and, where test results are provided in the form of a risk assessment, it should be based upon robustly evaluated algorithms and should make clear the distinction between relative risk and absolute risk. There should also be regular review of the available evidence upon which the interpretation is based.***



## 10. Provision of Test Results

The results of genetic tests and the significance that should be attributed to a particular genetic test result should be described to the consumer in a format that is easy to understand.

When testing for a condition or trait, where such conditions or traits are determined, at least in part, by other, non-genetic factors in addition to genetic markers, the test provider should make consumers aware of these other factors when providing results of genetic tests. In addition, the test provider should supply an indication of the level of significance that an individual should attribute to the genetic test results in comparison with the significance of these other factors. This should be provided to the consumer in a format that is easy to understand.

When providing consumers with the test results for pharmacogenetic tests, the test provider should strongly recommend that the consumer not alter the dosage of any existing medication on the basis of the test results and take the results of the pharmacogenetic test to a medical practitioner for personalised interpretation of the test result. The test provider should give the consumer appropriate information to take with them to their medical practitioner to aid the interpretation of the test results.

The test provider should not overstate the value or significance of the results of the genetic test when providing the test results.

The test provider should state clearly when a genetic test result can only give an indication of relative risk in relation to the general population as opposed to an absolute risk, bearing in mind that either might only be calculable in the context of a family history analysis.

The test provider should have in place a process to evaluate how well consumers are able to understand the background information and test results they have received, and take steps to improve their information and results provision in accordance with the findings.

The test provider should ensure that the provision of genetic test results is undertaken in such a way as to retain the confidentiality of personal and genetic data. When genetic test results are provided electronically, the test provider should ensure that appropriate security measures are in place to maintain the confidentiality of data transmitted. If the option of sending test results via email is offered by the test provider, consumers should be made aware that this method is generally not secure.

Test providers who interpret data obtained from genetic tests that have been provided by a third party laboratory should comply with all the aspects of these Principles that are relevant to the services they provide. Likewise, test providers who only undertake the genetic analysis and do not interpret the test results should comply with all the aspects of these Principles that are relevant to the services they provide.

The test provider should be able to provide consumers, at the time of testing or at any subsequent stage, with information about opportunities that are available for any further consultation with health professionals, including any conflict of interest that may arise.

**Recommendation 14**

***The test provider should not overstate the value or significance of the results of the genetic test when providing the test results.***

**Recommendation 15**

***Where there are two or more providers, one or more performing the testing, and one or more performing the interpretation, then each should comply with these guidelines where they are relevant to the aspect of the service each has provided.***

## 11. Handling Complaints

The test provider should have written procedures in place for acknowledging and investigating complaints that are freely available to the consumer. Staff who manage and respond to complaints should have received training relevant to the test and the consumers of the test.

The test provider should ensure that complaints are dealt with in a reasonable time-period and consumers should be informed promptly of the outcome of the complaint.

If a consumer remains dissatisfied with the investigation or outcome of their complaint, they should be made aware of what further recourse might be available to them, including through the TGA, the Health Ombudsman or other relevant bodies.

TGA relies on the public, healthcare professionals and industry to report problems with IVD medical devices (see [the TGA website](#)).

Problems can be reported to the TGA via the following email address - [iris@tga.gov.au](mailto:iris@tga.gov.au)

### **Recommendation 16**

***The provider should ensure that the complaints process is known to the consumer and that complaints are dealt with in a reasonable time-period. Consumers should be informed promptly of the outcome of any complaint.***

## 12. Massively Parallel Sequencing

With the continued development of massively parallel sequencing (MPS) combined with dramatic improvement in sequence assembly software design, whole genome sequencing (WGS) will become cheaper and, as noted in the recent PHG publication *Next steps in the sequence*<sup>15</sup>, the entire industry is becoming increasingly geared towards a genome-wide approach to analysis. Whether to have a test and how to interpret the results will become significantly more problematic because of the increased likelihood of results from WGS containing clinically or personally significant findings that could be classed as incidental. This leads to significant ethical issues that need to be addressed before and after performing such testing on consumers. Amongst these issues are:

- (a) Genomic testing should only be performed if the consumer has received appropriate pre-test counselling by a qualified genetic counsellor or a relevant medical specialist. Such counselling should include discussion around both expected and incidental findings.
- (b) The results of genomic testing should be given to the consumer during post test face-to-face counselling.
- (c) The provider of genomic testing should have developed and should provide the consumer with a written policy regarding reporting of incidental findings.
- (d) The provider of genomic testing should also be aware of, and have considered, their responsibilities to other family members of the consumer.

Consequently, it is strongly recommended that genomic testing is not offered direct-to-consumer unless the provider has the appropriate medical infrastructure within their organisation.

### **Recommendation 17**

***The provider should not offer direct-to-consumer genomic testing, such as Whole Genome Sequencing (WGS), unless the provider has the appropriate medical infrastructure within their organisation to be able to deal appropriately with the medical ethical issues that arise as a consequence of such testing.***

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