In recent years the number of DNA genetic tests that you can buy without needing a referral from your doctor has increased. This booklet informs you about these Direct-to-Consumer DNA genetic tests and the important role of your doctor in providing you with health advice.

What does Direct-to-Consumer DNA genetic testing involve?

Direct-to-Consumer DNA genetic tests are tests available for purchase, usually online, through laboratories that are located overseas. These tests allow you to deal directly with the laboratory for a fee. You do not need to be sick to ask for these types of tests.

Testing involves ordering a test online and then sending a sample from which your DNA can be taken, such as saliva or a swab from the inner cheek, to a laboratory for testing. Your DNA, or deoxyribonucleic acid, which is inherited from your parents, contains genetic information that contributes to your development and functioning. The sample is analysed and the results are then provided directly to you – without the involvement of your doctor.

Things to consider when deciding whether to have a Direct-to-Consumer DNA genetic test

You might like the idea of Direct-to-Consumer DNA genetic tests because they don't involve a blood test, are simple to do and can allow you to keep the results private. Such a test might also appeal to your curiosity to discover more about yourself. Whatever the case may be, it is important to know that while Direct-to-Consumer DNA tests can be taken for fun or personal interest, there are factors such as test accuracy and privacy, which you should consider if you, or someone you know, is thinking about having a Direct-to-Consumer DNA genetic test. These factors are outlined in the rest of this booklet.
How accurate are the tests?

Laboratory analysis of genetic information is usually performed with a certain level of accuracy. This may vary depending on who is doing the test. Companies offering Direct-to-Consumer DNA tests are mostly located offshore and may or may not follow the strict Australian laboratory regulatory requirements.

It can be difficult to determine a laboratory’s quality control standards. This includes knowing how well they will perform the test and whether they analyse and interpret your DNA sample accurately and appropriately. If this is not done correctly, the results and claims made may be misleading, unclear or inaccurate, yet the disclaimers attached to the testing may clear the company of having any responsibility should this be the case.

How useful are the tests?

Direct-to-Consumer tests typically look for specific variations (changes in your DNA), which have been linked to certain diseases, conditions or traits. There are many more variations and details that are yet to be understood. Therefore, the health care value or usefulness of a Direct-to-Consumer test will vary depending on the test. In the case of predictive genetic tests, the variations tested often only have a small influence on your overall risk of developing a disease, and sometimes the link with a disease may be weak. Such tests have been made available directly to consumers because companies view genetic testing as a profitable business opportunity.

Test results and levels of risk can mean different things to different people. Different companies can also return different interpretations of the same sample. Current tests provide only part of the picture. It is important to remember that for most diseases, your genes interact with environmental and lifestyle factors in ways that are not yet well understood. Because of this, the benefits of some Direct-to-Consumer tests may be overstated. It is also possible that you may not necessarily receive all the information for which you paid.

Before undertaking any genetic test—Direct-to-Consumer or not—it might be worthwhile to consider whether the information the test provides will make a difference to you. Is there something that you hope to be able to do after getting the test result that you can’t do now? In the case of health-related predictive tests, for example, unless you are willing to make changes to your lifestyle based on the test results, such tests may not be of much benefit to you.

More information about what Direct-to-Consumer DNA genetic tests can tell you is included below. If you are considering any Direct-to-Consumer test, talk to your doctor (GP). He or she can help you to better understand what a test can and can’t tell you at this point in time.

What can Direct-to-Consumer DNA genetic tests tell me?

Direct-to-Consumer DNA genetic testing companies claim to be able to provide you with different types of information, depending on the type of test. These can be related to:

Health – Your DNA plays an important role in susceptibility to, and in the development and progression of, many conditions. At present, no Australian laboratory offers diagnostic or predictive DNA health tests direct to the consumer. These tests are available in the medical setting on the referral of a doctor. Some Direct-to-Consumer tests available online claim to predict a person’s susceptibility to complex conditions such as heart disease, obesity or depression, which involve many other factors. At this time, studies have yet to prove that such susceptibility tests give accurate results to consumers.
**Kinship** – As your DNA shares genetic ‘markers’ with biological family members, it can be used to determine your relationship to other individuals or groups. This allows you to test for paternity for legal purposes, provide evidence of familial relationships for immigration purposes or to find out about your ancestry. While such tests are available as Direct-to-Consumer tests in Australia, it is important to note that, currently, the results of these tests may not be acceptable in Australian court proceedings.

**Behaviour and Ability** – Research is uncovering a range of genetic ‘markers’ that may be associated with certain traits, including behavioural traits and performance abilities. It is claimed that individual differences in these markers can be used to estimate a person’s abilities and aptitudes, such as intellectual and sporting ability. Currently, the science remains unclear as to their usefulness.

**Lifestyle** – These tests claim to identify health-related ‘markers’ in your DNA that are associated with differences in lifestyle, in order to provide you with advice on lifestyle choices that is tailored to your genome. Examples include tests to provide information about your responsiveness to a given nutrient or diet (nutrigenetics) and beauty products (dermatogenetics). The usefulness of such tests can be questionable as they often end up providing common-sense dietary and lifestyle advice that would be available to you through other sources and probably at less expense. The accuracy of these tests is also questionable.

**Genomic profiling** – Some overseas laboratories also offer Direct-to-Consumer DNA testing in the form of whole ‘genome scans’ and sequencing. This provides information describing an individual’s entire unique DNA profile, although the amount of analysis and interpretation of one’s complete genome sequence is still limited by our current levels of understanding.

**What happens to my personal information?**

The results of Direct-to-Consumer DNA tests are provided directly to you. Whether these are mailed out or made available online may vary according to the provider, along with the method and period of time that your DNA samples and results are stored. These details are important to consider and should be outlined in their policies.

Prior to testing, it is important to clarify whether your sample and results can be used for other purposes without your consent. Given the unique nature of your DNA, careful consideration should be given before sending your sample overseas to countries that may have different privacy protections to those that exist in Australia.

**What protections are there in Australia?**

In Australia, Direct-to-Consumer genetic testing services are regulated by a combination of laws and voluntary codes of conduct. The Privacy Act 1988 (Commonwealth) covers protection of health information, which now includes reference to genetic information. The Trade Practices Act 1974 (Commonwealth) contains protections for Australian consumers against misleading or deceptive conduct or misleading representations. Fair trading laws operating in each State and Territory aim to protect consumers from unfair trading practices.

Combined with industry codes of practice, these laws help to ensure that businesses operate fairly and competitively, and that all consumers are adequately informed and protected. The Australian Competition and Consumer Commission (www.accc.gov.au) has more information on what your consumer rights are and what you can do if you have any complaints.
What protections are there for offshore tests?

Regulation of testing done in laboratories in other countries is limited. Direct-to-Consumer DNA genetic tests that are offered and conducted by overseas-based laboratories are not regulated in Australia. The protections applicable to Australian residents are not enforceable against overseas based laboratories. This could leave you at risk should problems arise.

Do I need to provide details of my test results to an insurance company if I want to apply for life insurance products?

As genetic testing becomes more accessible, life insurance companies, which offer products such as disability insurance and critical illness cover, view this information as being no different to any other health information. Therefore, it is your responsibility to declare any known health information in life insurance product applications. This includes whether you (or your immediate genetic relatives) have had any genetic testing for which the result is known.

Australian insurance companies are bound by the Disability Discrimination Act 1992 in ensuring the fair use of sensitive genetic information. This means that any decision that a life insurance company makes using such information must be justifiable. Some of the DNA tests offered by Direct-to-Consumer companies have potential health implications but it can be difficult to know how the results from these tests will be interpreted in the context of insurance.

How might the test results affect me emotionally?

Everyone’s reaction to receiving personal results is different. In the case of genetic testing, receiving unexpected Direct-to-Consumer results could cause you significant anxiety or stress. These feelings are more likely to occur in cases where your result claims an increased risk, or diagnosis of a condition—particularly if it is one that has no effective prevention or cure available currently.

Results of Direct-to-Consumer tests are not always ‘black and white’. For health-related predictive tests, a result that says you have an ‘increased risk’ of developing a certain condition compared to the average person may cause needless alarm, while a test result indicating a ‘decreased risk’ may give you a false sense of security. This might also mean that possible symptoms or the benefits of leading a healthier lifestyle and/or taking early detection tests are ignored, which can then lead to long term and potentially serious health problems.

It is also important to note that non-health-related genetic tests that do not claim to provide specific information about disease risk, such as ancestry tests, can also reveal information that may have implications for your health. This is because some health conditions are more common in people with certain ancestry than others.

Before deciding to take a test, it might be useful to think about the range of possible impacts and ask yourself questions such as:

- Why am I considering taking this test?
- What do I hope to get out of this test?
- Will the test be able to answer my question(s)?
- What if I do not get the result I was hoping for or expecting?

Genetic counselling is usually available to help with these questions if you have a genetic test ordered by a doctor. However, with much Direct-to-Consumer testing, genetic counselling is often not routinely available, limited or not sought. If you wish to talk through the potential uses and implications before taking a genetic test, speak to your doctor (GP). The final page of this leaflet includes further suggestions on who to speak to.
**Will the results have any implications for my family?**

It is important to realise that your DNA test result can be relevant to your genetic (‘blood’) relatives. Given the nature of genetic information, you may want to consider sharing relevant information with them, particularly in the case of genetic conditions that may be inherited. Knowing this type of information may benefit your family members, especially if it helps them make plans for their future, such as undertaking regular screening for prevention and early detection.

However, the sensitivity and impact of genetic information may also affect existing social and legal obligations within families. Genetic testing results can affect how family members relate to each other, particularly in the case of identity related testing, such as paternity tests. While the sharing of information may benefit some, other family members may find out more than they wish to know.

**What support and information can I get if I decide to take a Direct-to-Consumer test?**

Some Direct-to-Consumer tests lack scientific validity and others provide health results that are meaningful only when given in the context of other information. This is why access to suitable professional advice and counselling is still important. Health professionals including clinical geneticists, genetic counsellors and your doctor (GP) can help you understand your test results and any implications. They can also advise you if there are other methods of finding out the information that you are seeking.

While health professionals are often bypassed in Direct-to-Consumer testing, if you have any questions or concerns about your current and future health, your doctor (GP) should be your first point of contact. He or she can offer support and help you make an informed choice about whether to proceed with genetic testing, and refer you to genetic specialists as required.

State and Territory genetics services can also be useful points of contact and can provide you with information on services available in your region. The Centre for Genetics Education (www.genetics.edu.au) lists laboratory and genetics services for genetic counselling, prenatal testing, family cancer and other specialities in Australia. The Centre for Genetics Education also provides a list of available genetic condition support groups.

Your family may also be able to offer you support. You may want to consider involving them as they are likely to be affected by your decision to get tested. Finally, the company selling the test should also be able to provide more information prior to testing and may offer counselling services via the internet or by telephone. If you do not feel comfortable about the advice or level of support you receive through their services, it is worth talking to your doctor and/or seeking a second opinion.

**How else can I find out the information I’m seeking?**

If you are considering taking a Direct-to-Consumer test for health-related information, genetic tests are not the only way to get this information. Several standard, non-genetic clinical tests can provide you with a good indication of your health risks, such as tests to measure cholesterol levels. Your doctor (GP) is able to advise you about these and offer you advice about lowering your risk of many future medical disorders.

Talking to your family can also be a useful way of finding out more about your family’s medical history. Should you have a family history of a condition you are concerned about, speak to your doctor. Should the need for genetic testing be suggested, you can also have this done within the properly regulated clinical setting, which provides you with greater protection, counselling and support. More information on this kind of testing, known as medical genetic testing, is available from the NHMRC website.

**How was this information developed?**

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

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