



Pathology

The Facts

What should I know about Direct-to-Consumer **genetic testing**?

Direct-to-Consumer (DTC) genetic testing refers to tests directly requested from a laboratory by a consumer without requiring a referral from a treating practitioner. The consumer deals directly with the laboratory including collecting the sample and sending it to the laboratory, receiving the results and paying for the test. DTC genetic tests are usually marketed directly to consumers through public media such as the Internet and magazines.

How do DTC genetic tests differ from other types of genetic tests?

Recreational genetics: Some genetic testing carries no risk of impact on health or wellbeing. For example, “recreational” genetics may involve testing the type of earwax one has, while “genealogy” or “ancestry” genetics may involve determining which country a person’s original ancestors came from. DTC can be very convenient for this type of genetic testing.

Paternity testing and forensic genetics: Genetic testing can be used to establish the identity of people, and to test if two people are related to each other. This type of testing is highly regulated by the courts and involves testing in accredited pathology laboratories. (Refer to fact sheet - How safe is the pathology testing process?.)

Community genetics: Community genetics offers testing directly to the consumer, but only as part of a full healthcare program involving accredited pathology laboratories and registered health practitioners. Community genetics is often relevant to consumers from particular geographic origins and community groups, and can be used to screen for

risk of particular inherited disorders such as “Tay-Sachs disease” which is a serious disorder affecting young children.

Medical genetics: Genetic testing is increasingly used to diagnose a range of diseases including inherited disorders and some cancers. This type of testing is only carried out as part of a medical consultation, and the treating practitioner will use an accredited laboratory to test the sample and provide the pathology report.

Did you know?

Some DTC laboratories offer tests for medical genetics, but label the results with a disclaimer that it is “for informational purposes only”. Using these DTC services greatly increases the risks to consumers as they are left without the support of a health practitioner to advise them about which genetic test results are relevant to their particular individual health circumstances, and which genetic test results may require medical follow-up or treatment.





Do DTC genetic tests put me in charge of my own health decisions?

Consumers are encouraged to take greater care and responsibility for their own health, actively participate in their health care decisions and adopt a healthy lifestyle. The rapid advances in genetic testing have meant consumers can now purchase low-cost DTC investigations which might appear to fulfil this 'healthy living' independent philosophy, but is not always the case.

Are DTC genetic test results reliable?

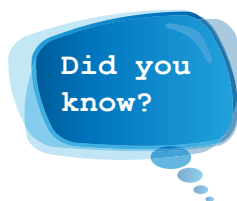
The National Association of Testing Authorities (NATA) and the Royal College of Pathologists of Australasia (RCPA) jointly perform an accreditation assessment of pathology laboratories in Australia. Accreditation is not compulsory for DTC testing. In fact, anybody can offer DTC genetic testing, regardless of qualification or experience, because they are usually offered outside of the regulatory and accreditation framework that applies to most other pathology tests.

The quality of the genetic test result can be assured if an accredited laboratory tests the sample. The consumer should therefore ensure that testing offered by a DTC service is only performed in an accredited pathology laboratory. They can do this by looking for the NATA/RCPA

logos or endorsement certificates that should be clearly displayed in accredited pathology laboratories, or by asking the pathology laboratory staff if they have NATA/RCPA accreditation.



Similarly, accurately interpreting the test result requires an appropriately trained expert health practitioner to review the results.



An individual assessed by more than one DTC genetic testing laboratory can receive quite different predictions for their future risk of the same disease. This is because different DTC laboratories use different risk tables linking the test result with the disease. There is currently insufficient scientific knowledge of the link between a particular genetic test result and the risk of developing a particular medical condition for many genetic conditions.

There is also the risk of testing benefits being overstated. For example, some DTC services claim genetic tests can predict "skin health" or "nutritional health", even though there may be limited evidence about the accuracy or usefulness of such tests.

Interpreting genetic test results

Interpreting genetic test results is not an easy task - even the experts can find it challenging! The results must be interpreted within the context of a person's medical and family history because other factors, such as obesity or smoking, may pose a greater threat to a person's health than their genetic risk profile.

There are also psychological risks involved if a person receives adverse genetic test results without appropriate genetic counselling, as well as the potential for consumers to be falsely reassured or alarmed if their genetic test results are not properly interpreted by an experienced health practitioner.

While a genetic test result can confirm if a particular genetic variation is present or not, this result does not necessarily provide a precise estimate of the risk or severity of disease associated with that gene. This is because a single genetic variation is only one of many factors, such as lifestyle, age and changes in other genes, which determine future health. The context of the test usually determines the medical significance of





the test, which is why an experienced health practitioner, who is aware of the consumer's personal health status and family situation, should interpret genetic test results.

What about consent and privacy issues?

Genetic tests should only be carried out once the consumer has been given sufficient and relevant information about the risks, benefits, limitations and implications of the test, and then freely given their consent.

Consumers should ensure they do not unintentionally consent to the release of their genetic information for secondary purposes, such as research, or for their information to be sold to a third party such as a pharmaceutical company. They can ensure this does not happen by reading the consent form thoroughly, or by asking these specific questions if they have the opportunity.

Pathology laboratories should also take reasonable steps to ensure the specimen received for testing is actually from the person identified as the specimen provider. This is difficult to monitor in some DTC testing because the consumer is usually never sighted by the DTC laboratory staff and may inadvertently accept specimens that are submitted under false names or identities, leading to serious threats to people's privacy.

Did you know?

Health practitioners assist consumers with decisions that are appropriate for their personal situation, and can arrange access to the relevant expertise required to follow through with that decision, such as genetic counselling. They can also discuss the ethical and social issues raised by the specific genetic test/s being considered. These benefits are lost with DTC genetic tests if they are performed without this expert interpretation and counselling.

This fact sheet has been developed by the Royal College of Pathologists of Australasia (RCPA) and the Human Genetics Society of Australasia (HGSA).

For further information on DTC Genetic testing the following document may be useful:

<http://www.hgc.gov.uk/UploadDocs/DocPub/Document/HGC%20Principles%20for%20DTC%20genetic%20tests%20-%20final.pdf>

Reliable information on pathology can be found at:

The Royal College of Pathologists of Australasia (RCPA)
- www.rcpa.edu.au

The Human Genetics Society of Australasia (HGSA)
- www.hgsa.org.au

Centre for Genetics Education
- www.genetics.edu.au

ePathWay (the RCPA's online magazine for consumers)
- <http://epathway.rcpa.edu.au>

The RCPA Manual
- <http://rcpamanual.edu.au>

The Pathology Associations Council (PAC)
- www.pathology.med.pro

Lab Tests Online
- www.labtestsonline.org.au

