



Pathology

The Facts

What should I know about genetic testing?

Genetic testing has benefits and risks that are different from those associated with other pathology tests. This is due to the predictive nature of certain genetic tests and the shared nature and ownership of genetic information. While there are also different reasons to have genetic tests, such as paternity testing and research, this fact sheet focuses on medical genetic tests which impact on health-related decisions.

The benefits of genetic testing

Genetic tests may:

- clarify an uncertain situation such as confirming a diagnosis
- provide peace of mind if fears of a genetic disposition to a disease prove unfounded
- predict a genetic predisposition to a particular disease. This enables health prevention and screening strategies to be put into place to reduce the risk, severity or impact of the disease.

The risks of genetic testing

The purpose of the genetic test in a specific situation determines the level of medical, psychological, social and ethical risk. This is because a genetic test can be used for multiple purposes with different levels of associated risk. For example, one genetic test may be used to screen a population for at-risk individuals of a particular disease, confirm a diagnosis, predict the risk of developing a particular disease or predict the risk of having a child affected with a genetic disorder.

Did you know?

It can be important to share genetic information with relatives, but an Australian study has shown that some relatives do not necessarily want to know this information. (<http://jmg.bmj.com/content/43/8/665.full>)

The general risks associated with genetic testing include:

- genetic test results can be distressing and may cause psychological harm if they identify a risk of developing a disease; especially if there is no known prevention, treatment or cure
- people can learn distressing information about inherited diseases or disorders involving other family members. This raises issues such as: 'Should I tell the family?' Other family members may not want to know this information and it may interfere with family relationships
- genetic tests results can change existing social obligations within families
- genetic information may lead to difficulties obtaining some forms





Did you know?

Genetic tests don't predict what will happen; they only give an idea of what might happen and therefore don't determine the medical outcome. For example, a genetic predisposition to heart disease does not necessarily mean a person will develop that disease, only that they might develop it. There are other non-genetic factors that influence the outcome such as lifestyle, environment, chance or ageing.

of life, travel or disability insurance or affect family planning decisions

- genetic information can also lead to discrimination in personal, social or work-related matters, even though these outcomes should not occur.

Will I need genetic counselling?

The treating practitioner requesting the test can provide genetic counselling and advice about the genetic condition, or they may recommend referral to a specialist genetic counsellor. The extent of genetic counselling required depends on the level of uncertainty regarding the medical implications of the genetic test result, the potential implications for the patient and the further implications for their family.

Who can request a genetic test?

- treating practitioners can request genetic tests for their patients.
- patients can request genetic tests directly from some private pathology laboratories. This is called Direct-to-Consumer (DTC) genetic testing. (Refer to fact sheet - What should I know about Direct-to-Consumer genetic testing?)

Did you know?

Genetic counsellors in Australia are tertiary trained health professionals with specialist training in genetics and counselling.

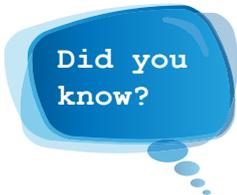
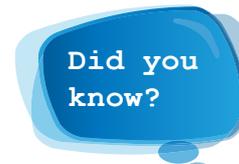
Consent for genetic tests

Before a person consents to a genetic test, their referring practitioner should provide information that helps them clearly understand the risks, benefits, limitations and implications of the test. They should also be given clear information about how long genetic samples and personal data are stored including what happens to these if the pathology laboratory ceases operation. They should also receive information about how results of important genetic tests that may have familial implications can be communicated to others in the family over a time period that may exceed a human lifetime.

Who performs genetic tests?

- specialist genetic testing laboratories in the public hospital system (the usual route for complex testing)
- pathology laboratories in the public and private sectors who perform genetic tests for a few common disorders
- Direct-to-Consumer (DTC) laboratories that advertise directly to patients through public media such as the Internet and magazines.





Pathology laboratories may refer some services to overseas laboratories to access rare or obscure genetic tests.

Pathology laboratory accreditation

The National Association of Testing Laboratories (NATA) and the Royal College of Pathologists of Australasia (RCPA) jointly accredit medical laboratories in Australia. Accreditation is only required if a pathology laboratory seeks a Medicare rebate for certain pathology services. This means that genetic tests could be performed in a pathology laboratory that has not been accredited for that scope of testing.



NATA/RCPA accredits very few medical laboratories outside Australia so assessing the quality standards of some DTC laboratories can be difficult. It is especially risky if the pathology laboratories are located in countries with minimal patient protection laws.

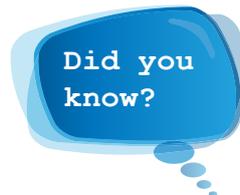
Interpreting genetic test results

Genetic test results should be interpreted by an experienced treating practitioner in the context of other health factors such as family history, environmental characteristics, other health conditions and current medications. The results can be challenging to interpret due to factors such as:

- a positive test result doesn't necessarily translate to a confirmed medical diagnosis, but might only indicate an increased risk of developing a disease or condition

Most genetic tests are offered free of charge to patients if they access them through a genetics service. Lists of these services are freely available through the Centre for Genetics Education – www.genetics.edu.au.

- a negative test result doesn't mean there is no risk of disease. For example, a person may have the same risk of developing the disease as the general population, or carry a genetic alteration that has not yet been identified but increases their risk of that disease
- the same test result can have different implications for different people based on factors such as their age, sex and environment.



A treating practitioner may be authorised under Federal privacy legislation to release information directly to at-risk relatives about a familial disorder. This is only allowed in exceptional circumstances. For further information go to:

http://www.privacy.gov.au/index.php?option=com_content&view=article&id=960&itemid=2147

Confidentiality of genetic test results

Genetic information is not provided to other health professionals, family members or third parties such as employers or insurance companies without the consent of the patient. However, pathology laboratories are required to send a copy of all test results to the requesting practitioner. Additionally, if the test has been referred from one pathology laboratory to another for testing, then the testing laboratory is required to send a copy of the report to the requesting laboratory.





Potential misuse of genetic information

Genetic test results, and their implications, may be misunderstood and misused by those who have access to them if they are not interpreted accurately. There is also the risk of intentional interference with test samples in certain situations such as during family law proceedings.

Will I have to pay for my genetic test?

Patients should ask their treating practitioner, or the pathology laboratory performing the genetic test, about the financial costs involved before they attend to have their test. This is because some genetic tests are provided free-of-charge through public hospitals, and a select few are funded by Medicare, while many genetic tests are offered privately, but only via a fee-for-service arrangement.

Genetic testing and the insurance industry

Under Australian law, applications for life and disability insurance are usually required to disclose any known health or genetic information about themselves or genetic relatives. Insurance companies and employers traditionally obtained this information by asking questions about the health and causes of death of close genetic relatives.

Did you know?

There is a risk that a person might not have a genetic test, or seek genetic counselling, in case the test results jeopardise access to life insurance at the expense of potentially lifesaving medical interventions.

As genetic testing becomes more accessible, insurers are viewing genetic test results in the same context as other health information and expect them to be disclosed.

However, the insurance industry in Australia has agreed not to require an applicant or insured person to have a genetic test as part of a policy application. If they already have information about a genetic relative, they don't extract that information and insert it into the person's file.

In Australia, private health insurers cannot charge a higher premium based on a person's medical history, including their genetic test results.

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

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

