Genomic Testing Information Sheet

About this information sheet
This information sheet explains genomic testing. It has been designed to accompany the clinical consent form for genomic testing that your health professional will discuss with you. The clinical consent form needs to be signed before genomic testing is started. Having genomic testing is optional. *It is your choice whether to have this test*.

Questions to ask your doctor/genetic counsellor

Once you have read this form, you may wish to ask your doctor/genetic counsellor these questions:

- What is the chance that the genomic test will identify the cause of my/my child's condition?
- How long will it take to get a result?
- Who will give me the result and how?
- Where will my genomic test be performed?
- What is the cost to me (if any) of my genomic test?
- What (if any) are the implications for other members of my family if I have this test?
- What is the chance of this test finding something that is unrelated to my/my child's current health condition?

Potential outcomes of genomic testing
Your doctor or genetic counsellor will discuss the outcomes of genomic testing including:

- Finding a variant that is the cause of the condition.
- Finding a variant of unknown significance (VUS). The effect of VUS is unknown. Sometimes testing in other family members for the VUS may help to understand if it could be the cause of a condition. The understanding of VUS may change over time.
- No gene variants found that could explain a genetic condition. Reasons for this include:
    - the variant causing the condition cannot be found by the test;
    - the gene causing the condition was not tested ;
    - the gene causing the condition is not yet known.
- Future testing may help clarify this, but the timing for this is unknown.

Potential benefits of genomic testing
Some people wish to have genomic testing to find a genetic diagnosis to help them understand their or their child's condition. A genetic diagnosis can also sometimes help families to access support and services that they need, and to plan for the future. A genetic diagnosis may also help health professionals manage a condition.

A genetic diagnosis may provide families with information about the chance of having another child with the same condition. Sometimes, the genomic test result in one person may also be important for the care of their relatives.

Genomic testing can lead to a diagnosis in 30-50% of people with rare genetic conditions. If a diagnosis doesn't happen today, the genomic test result could be looked at in the future as our understanding improves.

It is important to remember that genomic testing is not a general health test and will not identify *all* gene changes that could contribute to health problems that may develop in the future.

Australian Genomics Health Alliance

Potential risks of genomic testing

Incidental findings
In genomic testing, we are looking at many genes all at once and so there is a small chance they might see a variant in a gene that is not related to your health condition. This is called an incidental finding. It is a variant in a gene not related to the reason for doing the genomic test, but could be important to know about for your health. If your doctor determines that these incidental findings have important consequences for you or other family members, they will raise it with you. If medical follow up is required as a result of an incidental finding, your doctor or genetic counsellor will assist you by making appropriate referrals, if necessary. Your doctor or genetic counsellor will be able to give you some examples of incidental findings.

Insurance
In Australia, genomic testing will not alter your ability to get health insurance or your health insurance premiums. Genomic testing in you or your child could affect how easy it is for you or other family members to get income protection, travel or life insurance; or the price of your premium. An existing diagnosis may already affect your ability to obtain these kinds of insurance. Industry regulation prevents insurers from asking relatives for your genetic test results, and you cannot be requested to have a test by an insurer. Your healthcare provider will not provide your results to an insurance provider without your permission.

Further details can be found at: http://www.genetics.edu.au/Publications-and-Resources/Genetics-Fact-Sheets/FactSheet23A.

Withdrawal from testing
You can change your mind about having genomic testing or being told the results. You can cancel the test at any time before the laboratory finishes the test. You can also choose not to be told the result after the test is finished, but the test result will be placed in your medical record.

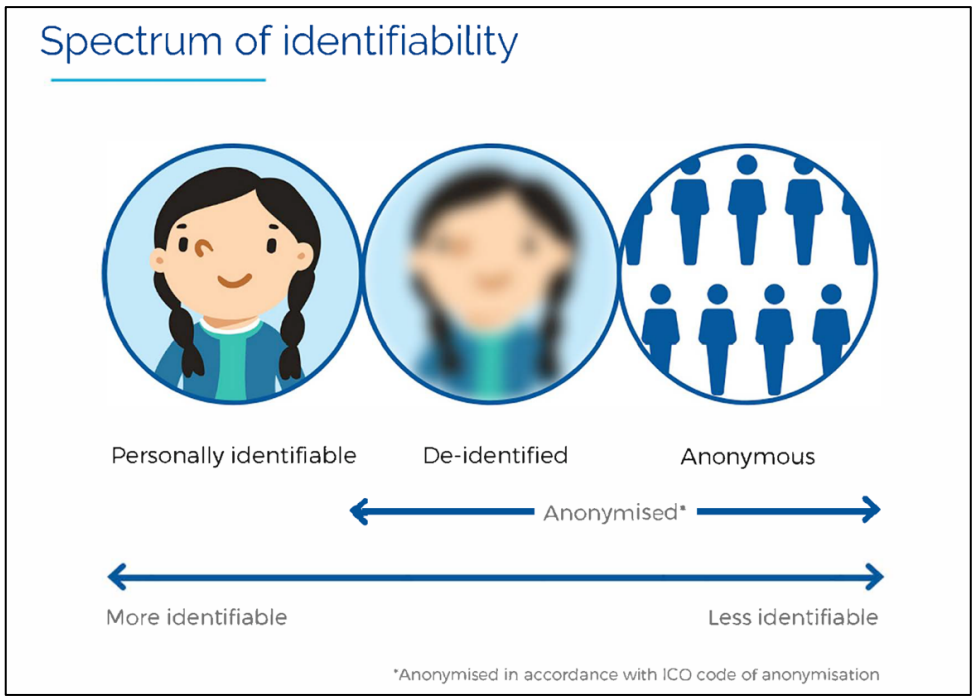
Sharing results to help family members
Genetic services will not usually contact your relatives. However, your relatives may be referred to genetic services to arrange testing for themselves or their children when you tell them, or, when they find out there is a genetic condition in the family. With your permission, your test results may be released to another genetics service to help with the care of other family members. As genetic changes are unique in families, it is helpful for genetic services to be able to share information (for example – exactly what genetic change is present in a family), so that the correct testing can be offered to others in the family who may be at risk. All efforts will be made to ensure that your identity is not revealed to those family members unless you wish to provide that information.

Data and Sample Sharing
Your identified results and genomic data from the test will be stored in secure, access controlled databases that meet Australian/international security standards and laboratory guidelines.

Your identified results and genomic data, your identified sample, or the fact that you have had genomic testing, will not be used or disclosed outside of your care without your consent, unless required or allowed by law. The health professionals involved in your care may order further testing of your sample or share your genomic data with each other to help work out what your test results mean.

Providing consent for genomic testing also allows for the sharing of your sample, genomic data and related health information to advance scientific knowledge. Your information will be shared in a way that keeps you anonymous. This may include sharing with large databases to help improve understanding of gene variants and related conditions by comparing your results to those from other people.

Your information and sample will only be shared when safeguards are in place to help protect your privacy. Personal identifiers will be removed (including your name, date of birth, and address) and stringent security measures will help prevent unauthorised access or misuse. Sharing will also involve only the least information necessary. These safeguards make it difficult to know whether the information is about you or other people; however, there is always a very small chance that you might be re-identified. Given that the potential to identify you is significantly reduced, you are unlikely to directly benefit from this sharing.

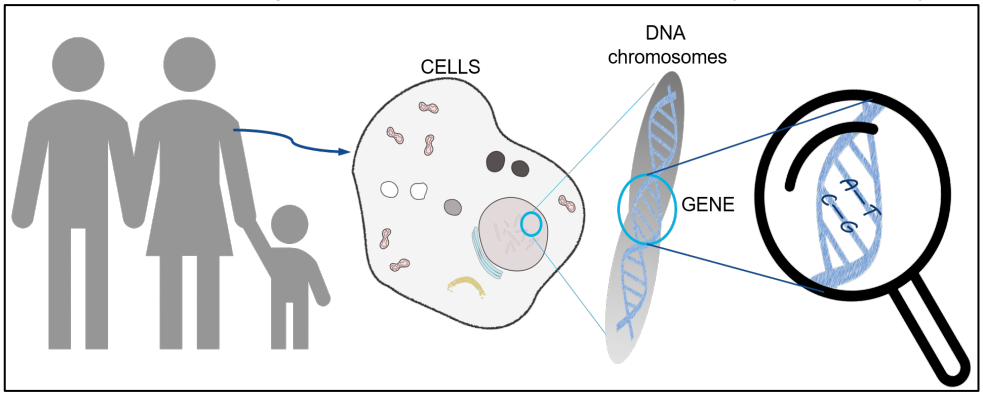


You can also provide consent to sharing for research into the **same or related condition** in a way that may be linked back to you. In this context, the identifiers described above can be removed from your information or sample, and replaced by a code. If there are findings from this research that have implications for your future clinical care, it may be possible to re-identify you, so that your results can be returned. However, participating in research does not guarantee direct benefits to you.

All researchers must respect the relevant laws (including privacy and security requirements) and ethical guidelines for biomedical and health research. Sharing in this context will only happen for projects approved by a human research ethics committee. You can choose not to provide consent to this sharing.

About the test – What is Genomic testing?

Our bodies are made up of billions of cells. In most of our cells we have a complete copy of our genetic information (genome). We all have about 20,000 genes in our genomes. Our genes are made of DNA and contain the instructions for growth and development of the body. Until recently, doctors and scientists were only able to test one gene at a time. Genomic technology allows us to test all of our genes at once (genomic testing).



DNA can be taken from your body cells to identify changes that contribute to or cause disease.

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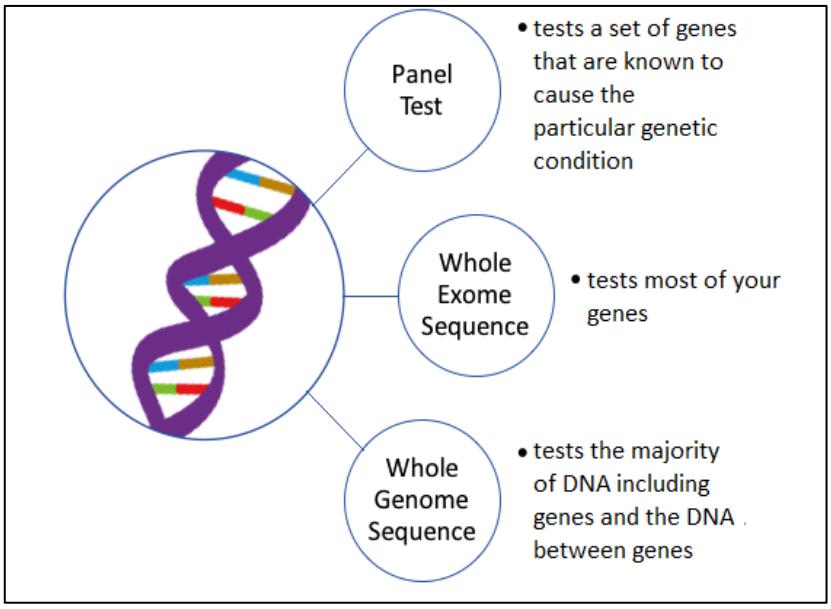
Types of genomic testing
Most genomic testing uses DNA that comes from a blood, tissue, saliva, or mouth swab sample. There are three main types of genomic testing:



Types of genomic tests

What are genetic variants?

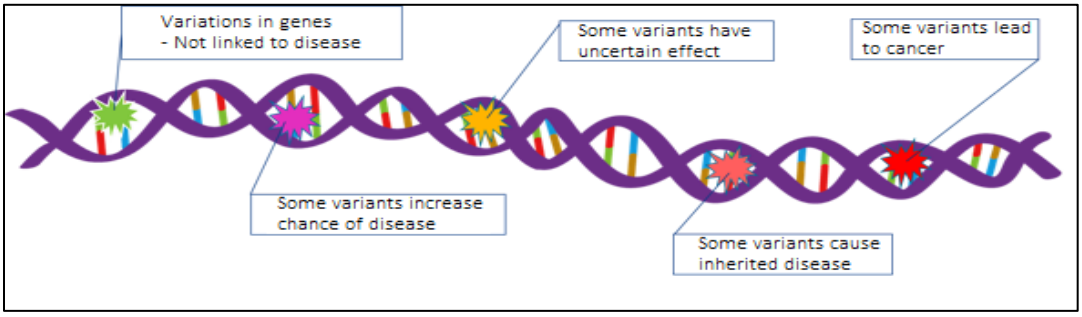
Each person's genome contains many genetic differences (variants). Most of these are harmless and do not change how the gene works in the body. Genomic testing is done to find gene variants that *do* change how a gene works and therefore cause genetic conditions.

Example:

Think of a gene as like a sentence in an instruction manual and consider the following sentence **MUM CUT THE HOT DOG**

MOM CUT THE HOT DOG	Some spelling changes look different but *don't* change the meaning of the sentence. Therefore, the gene still works.
MXM CUT THE HOT DOG	Other spelling changes look different and *do* change the meaning. Therefore, the gene doesn't work.
MEM CUT THE HOT DOG	Other spelling changes look different but we do not know if the meaning changes. Therefore, we are uncertain what the effect on the gene might be.

Different types of DNA variations have different effects on the genetic code and body functions.



Questions?
This information sheet provides general information about genomic testing. It supports the information you should receive from a medical specialist or genetic counsellor. If you have any questions about the test or any of the information in this sheet, please contact your doctor or genetic counsellor.