VCGS
Leaders in Genetic Health

Victorian Clinical Genetics Services
Murdoch Childrens Research Institute
The Royal Children's Hospital
Flemington Road, Parkville VIC 3052
P (03) 8341 6201 W vcgs.org.au

exomes

clinical exome sequencing

Clinical Exomes Brochure 2

What happens to your genetic information?

The information generated by a clinical exome is stored in data files. Data may be re-analysed, either for the same set of genes in the initial analysis, or for a different set of genes, without obtaining another sample from you. Data may also be used for research, with your consent (see examples of use below).

Research to advance knowledge may lead to new or more certain information about genetic variants.

Advances in knowledge mean that it is possible that the interpretation of a result may change with time. This is most likely to occur for variants of unknown significance or if no gene mutations were identified in your test.

You may wish to check with your doctor periodically (e.g. every 2-3 years) to see if there has been a change in the interpretation of a result. On occasion, the lab may contact your doctor if new information is obtained that affects the interpretation of your result.

How will my genetic information be used?

A patient consent form must be complete prior to a clinical exome test being performed. This form describes how your DNA, exome data and associated health information (collectively known as 'information') may be used for different purposes. Some of the uses will occur without the laboratory or healthcare team re-contacting the patient (part A).

For other uses, patients will be re-contacted (part B).

Part A: Information that does not identify you

If you sign the consent form and have your exome sequenced, you agree to your information being shared with approved researchers in Australia and internationally (point 7 of the consent form). Your information will be used in a way that means you aren't easily identifiable (anonymised).

- Safeguards to protect your privacy include:
- Personal identifiers are removed (i.e. name/date of birth);
- Your personal details are kept separate;
- Your data is coded;
- Only information relevant to the question being asked will be shared;
- Stringent security measures prevent unauthorised access or misuse.

This use of your information has the potential to improve understanding of normal and abnormal genetic variants. Your information, along with information from thousands of other people, is compared to help improve our understanding of how genes vary across the population.

Your information may be included in online secure databases (i.e. a server/cloud providers) that meet international security and safety standards.

You may not benefit directly from this work, but your information may be used for advancement of knowledge generally. You will not be contacted about any results or analysis of your information because your personal details will not be provided to researchers.

Part B: Information that may identify you

Some examples of how your information could be used in a way that may identify you are listed below. These uses may reveal information that may be important to your health, or the health of your blood relatives. For all the examples below, your information can be linked to you so any information with important health implications can be returned to you. Examples of use:

Activities to clarify the cause or management of your condition.
For example, your doctor may share your information with international groups trying to identify variants common to people who all have the same condition. The greater the number of patients with the same condition and the same variants, the more likely the variants are contributing to the condition.

Research to identify new genes associated with your condition.
Research opportunities aimed at finding the cause of your condition may be offered. This may involve a re-analysis of your genomic data. With your permission, your doctor and the laboratory could share your information, meaning researchers may not need to re-sequence your genome and you would not need to provide another sample.

Trying to clarify what a specific variant does in the cells of the body.
To help clarify whether the variant affects cell functioning, the laboratory may ask for your information so they can do further work that may provide clues to the role of the variant.

Will information from a clinical exome affect my insurance?

Provided you are having the exome sequencing test to determine the cause of your condition, the test is unlikely to affect your ability to obtain insurance products, such as life and/or income protection insurance. You may wish to seek independent advice about this.

Further information

More information about genomic sequencing can be obtained from your requesting doctor, your genetic counsellor or on the VCGS website at: vcgs.org.au/exomes

Contact details

VCGS is a not-for-profit provider of a comprehensive range of clinical and laboratory genetics services. We provide genetic counselling support before and after testing.

Molecular Genetics Laboratory
Victorian Clinical Genetics Services,
4th Floor Murdoch Childrens Research Institute
50 Flemington Road, Parkville, Victoria 3052

P (03) 8341 6201 E exome@vcgs.org.au
F (03) 8341 6366 W vcgs.org.au

VCGS
Leaders in Genetic Health