

# exomes

clinical exome sequencing

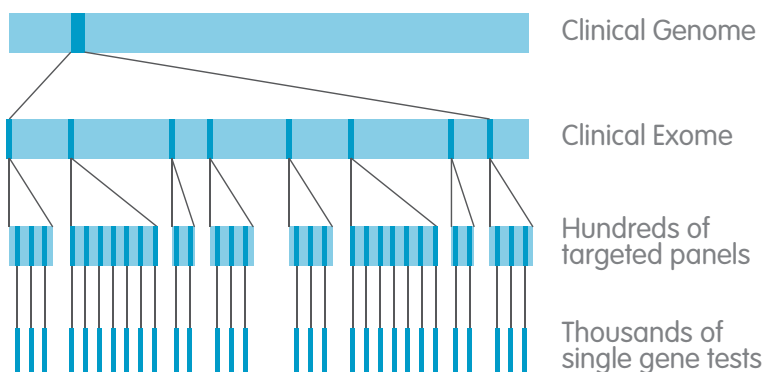
## VCGS and Clinical Exomes

VCGS provides Australia's most cost-effective, NATA accredited clinical genomics sequencing service. We take a multidisciplinary approach which includes clinical geneticists, genetic counsellors, scientists and bioinformaticians. Our fully integrated service offers segregation, cascade and prenatal testing where required.

Clinical exome testing can be applied in a wide range of genetic health investigations and may also be used by specialists (e.g. neurologists, cardiologists, nephrologists, medical oncologists, etc) to investigate specific causes of known but genetically heterogeneous conditions.

**Comprehensive analysis:** Our two-step approach using detailed phenotype-based information has been shown to be the most effective and reliable way to focus our attention on the most likely disease-causing variants.

**Clinical Exomes:** Consolidation of many single tests into one test



By focusing our analysis on the most disease relevant parts of the genome, we are able to provide very cost-effective genomic diagnostics.

VCGS provides Australia's most cost-effective, NATA accredited clinical genomics sequencing service, with highly optimised validated bioinformatics.

## Our Clinical Genomics Service

### Clinically driven:

We use the patient's clinical presentation as a means to drive our exome testing.

### Expert variant prioritisation and interpretation:

Genes related to the individual's phenotype are prioritised for analysis. If no diagnosis is reached in these genes, our highly experienced analysts apply an expanded gene list to maximise the diagnostic rate.

### Trio sequencing:

VCGS offers trio sequencing. Trio sequencing with joint variant analysis offers increased likelihood of a definitive result, especially for complex phenotypes.

### Incidental findings:

We minimise the chance of detecting unrelated health problems when testing children by filtering out variants in genes known to cause adult-onset conditions.

## How do I arrange this test for my patient?

VCGS accepts referrals for clinical exome sequencing from clinical geneticists and specialists. The following information is required in order to request a test:

- Pathology Request Form (4ml EDTA blood) for 'Clinical exome sequencing'.
  - Detailed phenotype information.
  - Suggested **Gene List\*** relevant to the patient's clinical phenotype.
  - Signed **Consent Form\*\***.
  - Completed Billing Authorisation Form.
- \* **Gene lists** from previously tested patients with a shared phenotype are available. The genetic counsellor can provide this information if required.

\*\* **Consent Form** signed by the patient or parent/guardian is a requirement of VCGS clinical exome testing. The Consent Form is available on request ([exome@vcgs.org.au](mailto:exome@vcgs.org.au)) and provides information to families regarding the risks and benefits of the requested test, including the chance of incidental findings and authorisation for re-analysis in the case of negative findings.

Please contact the Clinical Exome Genetic Counsellor for further information regarding these requirements.

## Clinical Exome Report: What to expect

A clinical exome test looks for any variation within a person's coding DNA sequence when compared to a reference exome. We use detailed phenotype information to prioritise relevant genes for analysis.

Our multidisciplinary team of medical scientists, genetic clinicians and specialists will assess the latest information from the scientific literature and clinical databases to interpret candidate variants using the American College of Medical Genetics based guidelines variant classification scheme.

To minimise the risk of incidental findings, we liaise with clinicians to only analyse genes that are likely to be relevant for the patient's clinical presentation. Analysis will progress until a likely cause for the patient's condition has been determined or until all variants in the genes deemed relevant to the patient's condition have been found to be likely benign. Accordingly, your patient's report will contain a list of variants that were found to be the likely cause for the patient's condition, as well as variants

# VCGS: Sharing Knowledge and Supporting Choice.

that could not be unambiguously interpreted (Variants of Uncertain Significance). In addition, we will provide a detailed interpretation of the reported variants, a list of genes we considered during the interpretation, as well as a list of potentially relevant regions of the exome that we were unable to assess.

## About VCGS

VCGS is a not-for-profit wholly owned subsidiary of the Murdoch Childrens Research Institute (MCRI) based at The Royal Children's Hospital (RCH) in Melbourne, Australia. MCRI and RCH are Australia's largest child health research organisation and largest children's hospital respectively.

### Integrated Service:

VCGS offers a unique, fully integrated service that provides a range of childhood and adult pathology testing and clinical genetics services. We are scientists, bioinformaticians, genetic counsellors and clinical geneticists that work together to provide the most comprehensive service for patients and health professionals.

### Expertise:

VCGS has been offering testing and genetic counselling services for 30 years. Combined with the Murdoch Childrens Research Institute, we have the largest genetic expertise in the Southern Hemisphere, with well-established pipelines for translating clinical research into diagnostics and genetic support services.



A request form with detailed phenotype information is required from a specialist doctor.



This test also requires completed Patient Consent and Billing Authorisation Forms.



Blood sample collection from pathology collection service (4ml EDTA blood).



Test results are reported to the referring doctor and can take up to 3 months.

VCGS provides access to pre-curated evidence based gene lists, which can be tailored to the needs of your patient.

## Contact Details

**Clinical Genomics Service  
Victorian Clinical  
Genetics Services**

4th Floor Murdoch Childrens  
Research Institute  
50 Flemington Road,  
Parkville, Victoria 3052

**Clinical Exome  
Genetic Counsellor**  
P (03) 8341 6201

E [exome@vcgs.org.au](mailto:exome@vcgs.org.au)

For more information and  
details about our clinical  
genomics service please visit:  
[www.vcgs.org.au](http://www.vcgs.org.au)