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

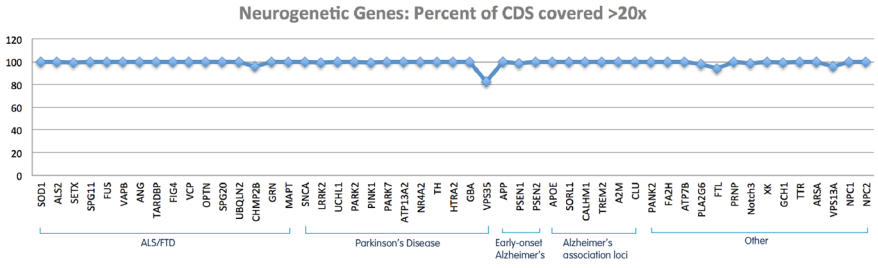
Clinical exome sequencing by VCGS

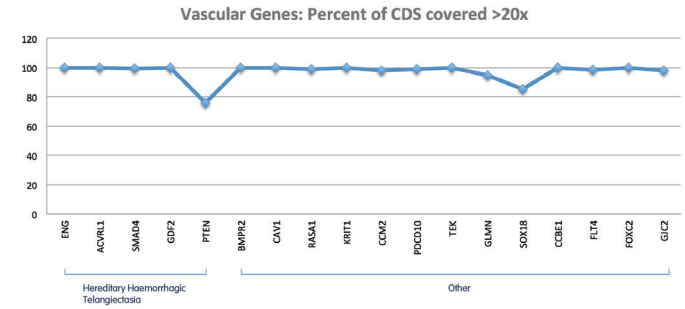
Clinical Exome Sequencing is a powerful tool used to help identify the cause of health and developmental problems.

One of the key advantages of performing clinical exomes is that sequencing data from the whole exome is captured. This allows you to dynamically adjust your clinical gene list to suit your patient's needs.

In addition, Clinical Exome sequencing enables you to re-analyse your patient's data at a later date as new evidence associated with your patient's disease becomes available.

Our 'augmented' exome sequencing approach is designed to specifically improve coverage over genes known to cause disease in humans*. Therefore, the vast majority of genes associated with neurogenetic and vascular diseases have 100% of their coding region covered at 20x or more, making our Clinical Exome sequencing a highly valuable tool for the identification of causative variants.





*Ashley EU. Towards Precision Medicine. Nature Rev Genet. Aug 2016, 17(9):507-22.

At VCGS, we use a phenotype driven analysis approach, using gene lists to prioritise variants for interpretation. This means there is little risk of incidental findings as analysis only progresses until a likely pathogenic variant has been identified, or until all high impact variants found have been determined not to be associated with your patient's clinical presentation. Using this approach allows us to analyse the most likely candidate genes for your patient, giving us the best chance to identify causative variants.

To complement our Clinical Exome sequencing service, we also offer exon deletion screening for APP, PSEN1 and PSEN2.

The Clinical Exome service at VCGS is a well-established and fully accredited test providing a flexible & cost-effective option for causative variant detection in neurogenetic and vascular disorders.

Of course, extensive testing options for other genes, as well as predictive and segregation testing are also available.

Contact us:

We highly value a close relationship with our requesting clinicians. We have a dedicated genetic counsellor available to facilitate referrals and discuss any Clinical Exome sequencing related queries or concerns.

For further information please contact Dr. Sebastian Lunke, Head of Translational Genomics Unit

P (03) 8341 6210
E exomes@vcgs.org.au
W vcgs.org.au/clinicalexomes