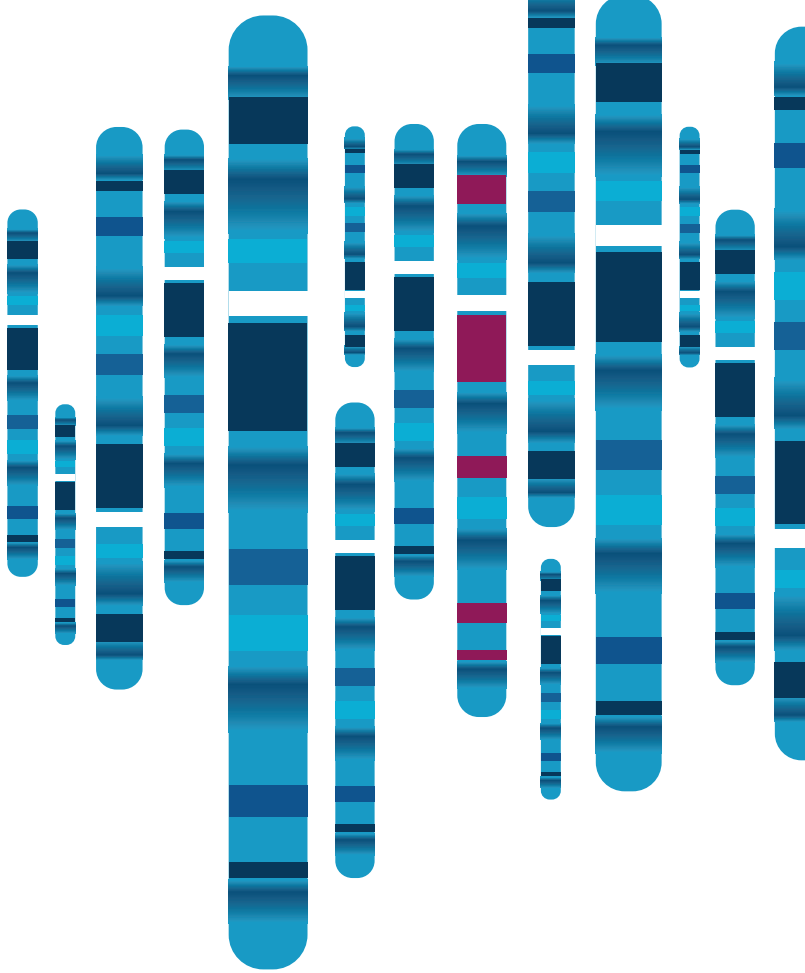
Reporting microarray results

Everyone has CNVs; they are what make us unique. Most do not affect our health, however some CNVs are known or suspected to affect health and development. Expert interpretation is needed to discriminate pathogenic CNVs from the benign.

Pathogenic variants	• Known, well documented deletion/duplication syndromes.
Uncertain variants	• Recurrent microdeletions/microduplications. • Susceptibility loci for neurodevelopmental/behavioural phenotypes. • Often inherited with incomplete penetrance and variable expressivity.
Unknown variants	• Novel; no comparable CNVs in any existing database. • Depending on genomic size, gene content and inheritance, may be clinically significant or a rare/benign familial variant. • Parental investigations required to determine significance.
Benign variants	• Commonly documented CNVs observed in individuals not suspected of having an underlying genetic condition. • Benign CNVs are not reported by VCGS.
LCSH	• Long continuous stretches of homozygosity may indicate a genetic imprinting disorder, or a recessive genetic condition. • VCGS can perform detailed genotype mapping where a recessive condition is suspected.



Ordering microarray from VCGS

Microarray analysis can be performed using blood* or saliva.

Our easy to use saliva collection kits (including return postage) can be ordered for your practice – see website below.

**Check with your local phlebotomy service to ensure sample goes to VCGS*

Chromosome microarray analysis and related genetic tests (e.g. like Fragile X which has a Medicare item number) are bulk billed.

VCGS is a not-for-profit specialist genetics service.
For information about chromosome microarray please see our website:
W) vcgs.org.au/tests/paediatric-microarray **P)** 1300 11 8247

CT-E--025 26/6/19

Advanced analysis to detect clinically significant chromosome changes

VCGS Leaders in Genetic Health | Paediatric microarray

VCGS are experienced providers of chromosome microarray (CMA) using saliva

✓ Using saliva is stress free for you and your patients

- quick and painless (20 secs)
- any age group
- sample is stable for nearly 2 months

✓ Saliva can be used for any of our genomic based tests (e.g. CMA, Fragile X, Prader-Willi)

✓ Saliva is accurate and reliable; equivalent quality to blood when tested at VCGS.

✓ Saliva tests both lymphocytes and fibroblasts - which is useful for detecting tissue limited mosaicism.

Our integrated laboratory + clinical service provides more clinically relevant reporting.

Our microarray service

Chromosome microarray (CMA) or molecular karyotype, is an advanced cytogenetic tool that detects small genetic changes that are known, or suspected to cause genetic and neurodevelopmental/behavioural conditions.

We use only high density single nucleotide polymorphism (SNP) arrays – a more advanced platform than comparative genomic hybridisation (CGH) or CGH/SNP blended arrays.

This platform detects:

- Copy number variants (CNVs), also known as deletions & duplications
- Low levels of genetic mosaicism*, down to 7-10%
- Copy number neutral changes, such as uniparental isodisomy# (UPD)
- Clinically relevant 'long continuous stretches of homozygosity' (LCSH) which may be indicative of imprinting disorders and recessive single gene conditions

**Where two different cell lines exist in one individual. Typically one normal and one chromosomally altered cell line.*

 # UPD is where both pairs of one chromosome are inherited from the one parent. UPD is a cause of imprinting conditions like Prader-Willi, Angelman, Beckwith Weiderman and Russell-Silver syndromes.

We manage a large internal database of variants (>500,000) from the local population. This, combined with our integrated clinical and laboratory service, gives you the most accurate, evidence based reporting for clinically relevant genetic variants.

1st lab in Australia to offer diagnostic CMA

7 fellowship accredited HGSA clinical cytogenetics scientists

>20,000 CMAs performed on saliva

>80,000 CMAs performed in paediatric setting

>500,000 genetic variants in our in-house database

SNP

We only use a high density SNP based platform

HGSA - *Human Genetics Society of Australasia*