

**GENOMIC MICROARRAY WITH SNP ANALYSIS**

Genomic microarray analysis is the latest technology in chromosome testing that can find small pieces of missing or extra chromosome (genetic) material. These missing or extra pieces are known as *copy number variants (CNV)*. Microarray can detect small CNVs that were not detectable by previous technologies, such as a karyotype. CNVs may help us to understand why an individual has congenital abnormalities (e.g. heart defect) or developmental delay (e.g. learning disabilities). Recent studies have shown that approximately 10-20% of individuals with unexplained developmental delay or multiple congenital anomalies (MCA) will have a CNV considered to be clinically relevant.

This SNP microarray platform will also detect absence of heterozygosity (AOH). AOH affecting multiple chromosomes suggests these regions are identical by descent. This information is included in the report for clinical interpretation by the referring clinician. AOH restricted to one chromosome may be suggestive of uniparental disomy (UPD). However, this assay is not designed to offer comprehensive UPD analysis. Standard molecular tests should be ordered if a disorder associated with UPD is suspected.

**CHROMOSOMES & MICROARRAY**

The human body is made up of millions of tiny cells. Inside each cell is a set of chromosomes which contain our genes. A person's genes will determine how they will grow and develop, both physically and intellectually. Microarray can detect missing or extra genetic information that can cause developmental delay or MCA. The clinical features will depend on the function of the missing or extra genes. This test can also find missing or extra genetic information that may not cause developmental delay or MCA, because no important genetic information is affected.

**POTENTIAL OUTCOMES & INTERPRETATION OF TEST RESULTS**

<b>RESULT</b>	<b>INTERPRETATION</b>
NORMAL	No abnormality identified. The cause of the individual's developmental delay or MCA remains unexplained.
PATHOGENIC VARIANT FOUND	A CNV that is associated with a specific pattern of clinical features is identified. An additional blood sample from the child and parents may be recommended to investigate the origin of the CNV. Genetic assessment/counseling will be recommended.
VARIANT OF UNKNOWN SIGNIFICANCE FOUND	A CNV of unclear significance is identified. This variant may or may not be related to the child's developmental delay or MCA. Testing of the child's mother and father may be recommended to assist with the interpretation. Genetic assessment/counseling may be recommended.
UNEXPECTED FINDING	Although this is unlikely, CNVs may be identified that are unrelated to the developmental delay or MCA in the child, but could possibly cause other health problems in the future. Genetic assessment/counseling will be recommended.
ABSENCE OF HETEROZYGOSITY	AOH of multiple chromosome regions suspected to be identical by descent will be reported for clinical interpretation by the referring physician. The laboratory does not use this data for clinical interpretations. AOH results suggestive of UPD of a clinically significant region will require follow-up by molecular tests designed specifically to detect UPD.

*For More Information*

Information regarding requisitions and sample requirements can be found at: [www.sickkids.ca/cytogenetics](http://www.sickkids.ca/cytogenetics)

For more detailed information on microarray technology and its uses, see the pamphlet published by Unique:

[www.rarechromo.org/forum/DisordersLeaflets.asp](http://www.rarechromo.org/forum/DisordersLeaflets.asp)

To locate a genetics center near you, please visit the Canadian Association of Genetic Counsellors website at [www.cagc-accg.ca](http://www.cagc-accg.ca) or the National Society of Genetic Counsellors website at [www.nsgc.org](http://www.nsgc.org)



1. Current microarray technologies will not detect single gene disorders or balanced chromosome rearrangements.

2. A normal microarray result does not rule out the possibility of a genetic cause for an individual's health or developmental concerns.

3. Test results should be interpreted in the context of clinical findings, family history and other laboratory data.

4. This test was developed and its performance characteristics validated by the Cytogenetics Laboratory at the Hospital for Sick Children. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.