Up to 20% of individuals with developmental delay and/or multiple congenital anomalies will have a gain or loss of chromosomal material.

Effective March 1, 2011 postnatal microarray testing will be available in Ontario at Credit Valley Hospital and The Hospital for Sick Children laboratories.

Who Is Eligible for Testing?

Ontario residents insured by OHIP with:

- Developmental delay including a delay in gross motor, fine motor, learning , speech& language, and/or social development

and/or

- Multiple congenital anomalies

Not recommended as a first tier test for:

Strong clinical suspicion of a single gene disorder (ex. Rett syndrome)

Couples experiencing infertility/recurrent miscarriage

Suspected aneuploidy (ex. Down syndrome, trisomy 13, trisomy 18, Turner syndrome, Klinefelter syndrome)

How to Arrange Testing for Your Patient

The requisition for microarray testing is available on our website.

www.cvh.on.ca/genetics

For first time access: You are required to contact the Genetics clinic (mwood@cvh.on.ca) for the username and password

Sample Requirements:

Minimum 3-5mL [1 mL for newborns] peripheral blood collected in an EDTA (lavender top) tube.

Shipping Address:

GENETICS LABORATORY
The Credit Valley Hospital
2200 Eglinton Ave. W., Rm 2H144
Mississauga, ON, L5M 2N1 Canada

P: (905) 813 – 1100 x6288
F: (905) 813 - 3854

Revised: March 1, 2011

CREDIT·VALLEY
THE CREDIT VALLEY HOSPITAL

Regional Genetics Program

Microarray Testing in Ontario

What is Microarray?

Microarray testing is a relatively new technology that looks for small gains or losses of chromosome material that may provide an explanation for why an individual has developmental delay and/or multiple congenital anomalies.

For more information about microarray testing, please visit our website at www.cvh.on.ca/genetics.

First-Tier Testing

Canadian College of Medical Genetics position statement (Sept 2009):

"Genomic microarray should be the first line test for developmental delay, mental retardation, autism, multiple congenital abnormalities or dysmorphic features"

Benefits

1000x higher resolution than routine chromosome analysis.

Limitations

Balanced structural chromosomal rearrangements such as translocations and inversions cannot be detected by microarray

Microarray is not designed to screen for single gene disorders (ex. Duchenne Muscular dystrophy)

Detection Rate

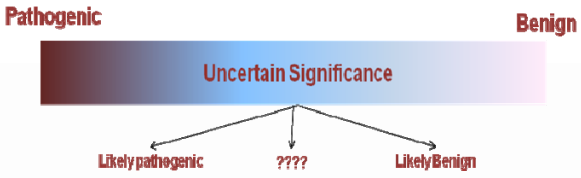
>99% pathogenic copy number changes (CNCs) can be detected by microarray

Turn around Time

Expected to be 4-6 weeks. Results could take longer if further analysis and/or family studies are to be done.

Interpretation of Result

Since microarray testing is relatively new, and the information generated is extensive, the interpretation of a variant identified in an affected individual can be difficult.



Factors considered when assessing the clinical significance of a CNCt:

Known syndromic region

Size of deletion/duplication

Inherited or *de novo*

Clinically relevant genes in deleted/duplicated area

Frequency of variant in patient and control populations

Genetics Follow Up

Normal Microarray Result:

Could consider a referral to genetics if you are still suspicious of an underlying genetic cause, such as a single gene disorder, to account for your patient's features.
Note: G-banding (karyotyping) is not recommended for patients with a normal microarray result.

Abnormal Microarray Result:

A referral to your local Genetics center is recommended. The clinic will arrange family studies (if recommended), offer an interpretation of the specific abnormality, as well as discuss recurrence risks and (possible) prenatal testing for future pregnancy