MICROARRAY and Q-PCR Follow-up Requisition

Genetics Diagnostic Laboratory
Eastern Ontario Regional Genetics Program
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https://www.cheo.on.ca/en/clinics-services-programs/genetics-diagnostic-laboratory.aspx

Patient Name: _____
Last First Initial
Health Card Number: _____
DOB: (yy/mm/dd) _____
Address: _____
_____
Telephone: _____
Sex (circle one): ◯ Male ◯ Female

ALL SECTIONS MUST BE COMPLETED

Collection Date: _____ Time: _____
Collection Centre:
☐ CHEO Inpatient ☐ CHEO Outpatient
☐ Other location: _____
(specify)
Specimen collected by: _____

Specimen Type

Blood (Collect blood specimens in an **EDTA** tube - 3mL for infants, 7-10mL for patients ≥ 2yrs. Do not freeze or spin.)

For any other sample types, please contact the laboratory directly.

Health Care Provider Requesting Test

Name: _____
Registration #: _____
Telephone: _____ Fax: _____
Address: _____
_____

***COPY to*:**
Name: _____
Registration #: _____
Telephone: _____ Fax: _____
Address: _____
_____

Test Requested

☐ **Microarray analysis - please indicate phenotype below**
☐ **Q-PCR Array Follow-up** ☐ Proband ☐ Family Member - Relationship to Proband _____

Clinical Information - Reason for test *(complete phenotypic description is required for appropriate interpretation of results)*

Neurological:
☐ Developmental delay
☐ Learning/Intellectual disability
☐ Autism spectrum disorder
☐ Macrocephaly ☐ Microcephaly
☐ Cortical malformation
☐ Seizures
☐ Other _____

Growth:
☐ Intrauterine growth retardation
☐ Failure to thrive
☐ Short stature
☐ Overgrowth

Other: _____
_____

Craniofacial:
☐ Dysmorphism, non-specific
☐ Dysmorphism suggestive of del/dup:
_____
☐ Craniosynostosis
☐ Structural eye anomaly/visual disability
☐ Choanal atresia/other nasal anomaly
☐ Cleft lip and/or palate
☐ Mandibular anomaly
☐ Structural ear anomaly/deafness

Cardiovascular:
☐ Structural heart anomaly: _____

Gastrointestinal:
☐ EA/Tracheoesophageal fistula
☐ Diaphragmatic hernia
☐ Intestinal atresia
☐ Malrotation

Genitourinary:
☐ Hydronephrosis
☐ Structural renal anomaly:
_____
☐ Uterine anomaly
☐ Hypospadias
☐ Other genital/reproductive anomaly

Musculoskeletal:
☐ Pectus excavatum or carinatum
☐ Scoliosis
☐ Vertebral anomaly
☐ Rib anomaly
☐ Oligodactyly/Polydactyly/Syndactyly
☐ Other upper extremity abnormality
☐ Other lower extremity abnormality
☐ Other: _____

☐ Homozygosity Suspected, specify chromosome/locus: _____ ☐ Parents Consanguineous

Ethnicity / descent from isolated community: _____

Specify any suspected syndrome(s) and or genes of interest: _____

PAST TESTING RESULTS (Please attach copy of reports if available)

Previous Cytogenetics: _____ Previous Microarray: _____

CHEO Genetics Pedigree Number: _____ Lab # _____

MICROARRAY TESTING: A BRIEF GUIDE FOR PHYSICIANS

What is microarray testing and when to order it?

Microarray analysis is a modern technology that is orders of magnitude more sensitive than a karyotype in detecting partial chromosome deletions and/or partial chromosome duplications.

In patients who have developmental delay, intellectual disability, autism spectrum disorder, dysmorphism or multiple congenital anomalies that are not suggestive of common chromosome abnormalities, microarray analysis is recommended as the **first tier test** by the Canadian College of Medical Geneticists (CCMG). Microarray analysis does not detect balanced structural chromosome rearrangements such as translocations and inversions, low-level mosaicism or point mutations.

What type of microarray testing is done at CHEO and how long does it take?

The CHEO Genetics Diagnostic Laboratory utilizes the Applied Biosystems^{TM} Cytoscan HD assay. Across the genome, deletions and duplications of 50kb or greater are detected. Additionally, results are assessed for long contiguous stretches of homozygosity of 5Mb or greater on autosomes.

The turn-around time for this test is approximately 6 weeks for routine samples.

What type of results can you expect?

Scientific literature and databases used for interpretation of the microarray results are continually growing; however, we currently have limited information on many regions of the genome. Thus, different types of results can be generated:

Abnormal result

Patients with a clearly pathogenic deletion or duplication should be referred to the CHEO Genetics clinic to receive a clinical assessment, appropriate genetic counselling and additional familial studies, when indicated.

Result of unclear significance

Further analyses are required to help in the interpretation of copy number alterations that are likely pathogenic or of unknown significance. The required samples for the proband and their parents will be indicated in the microarray report. It is very helpful and time saving when parental studies are performed prior to, or concurrent with, a referral to the CHEO Genetics clinic.

Long contiguous stretches of homozygosity (LCSH)

LCSH may be identified in a patient whose parents share a common ancestor (identity by descent) or, more rarely, when uniparental disomy is present. The microarray report may include recommendations to investigate the potential clinical significance of LCSH.

Unexpected result (also called incidental or secondary findings)

Rarely, testing may identify a copy number alteration that is not related to the reason for which microarray testing was performed but that could be potentially medically actionable. Patients with an unexpected result should be referred to the CHEO Genetics clinic to receive a clinical assessment, appropriate genetic counseling and additional familial studies as required.

Normal result

A normal result implies that the patient does not have a duplication or deletion larger than the sizes outlined above. Standard chromosomes analysis (karyotype) is **not** indicated for patients with a normal microarray result. A normal microarray does not rule out mosaicism, genetically balanced rearrangements, smaller deletions or duplications, imprinting defects, uniparental disomy, nor point mutations. A microarray does not address the possibility of a single gene condition. There are many circumstances under which a referral to the CHEO Genetics clinic is appropriate, regardless of the results of microarray.