SickKids®
THE HOSPITAL FOR SICK CHILDREN
Paediatric Laboratory Medicine

CYTOGENETICS LABORATORY
555 University Avenue
Room 3416, Black Wing
Toronto, ON, M5G 1X8, Canada
Tel: 416-813-7200 x 1
Fax: 416-813-7732
(CLIA # 99D1014032)

Patient Name:
Date of Birth (DD/MM/YYYY):
Sex Assigned at Birth: ○*Male* ○*Female* ○*Unassigned*
Legal Sex (if different): ○*Male* ○*Female* ○*Non-binary/U/X*
Gender Identity: ○*Male* ○*Female* ○*Non-binary/U/X*
Parent's Name:
Address:

GENOMIC SNP MICROARRAY
Referred-In Requisition

For Canada Only
Health Card #:
Issuing Province:
Version:

Testing is provided for medical purposes only and results are not intended for forensic use. The laboratory is not a forensically accredited laboratory.

Complete in full to avoid delay in reporting result.

POSTNATAL Genomic SNP Microarray

Specimen Drawn:
Date (DD/MM/YYYY): _____ Time (HH:MM): _____

Specimen Type:
- ☐ Peripheral Blood in EDTA: 3 mL minimum (1 mL minimum for newborns)
- ☐ Fibroblast cell culture: 2xT25 confluent flasks at room temperature
- ☐ DNA; 1µg at ≥50ng/µL minimum

Karyotype *(if known)*:

Indications for Testing:
- ☐ Developmental delay or intellectual disability
- ☐ Developmental delay or intellectual disability and additional clinical features. Complete Clinical Description Form (page 2).
- ☐ Two or more congenital anomalies. Complete Clinical Description Form (page 2).
- ☐ Microarray/qPCR Family Follow up

Relationship to Proband: _____
Proband Report/Order #: _____

Family History

Pedigree (at least 3-generation, when available and if applicable):

Referring Physician
Name: _____
Address: _____
_____
Phone: _____ Fax: _____
Email: _____
Signature *(required)* _____

Copy Report To
Name: _____
Address: _____
_____
Phone: _____ Fax: _____
Email: _____

Laboratory Use Only

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Phenotypic Description (Clinical symptoms)

Behavior, Cognition and Development

- ☐ Global development delay
- ☐ Fine motor delay ☐ Gross motor delay
- ☐ Language delay
- ☐ Learning disability
- ☐ Intellectual Disability
  - ☐ Mild
  - ☐ Moderate
  - ☐ Severe
- ☐ Attention deficit hyperactivity disorder
- ☐ Autism Spectrum Disorder
- ☐ Psychiatric disorders (Specify below)
- ☐ Other: _____

Neurological

- ☐ Hypotonia
- ☐ Seizures
- ☐ Ataxia
- ☐ Dystonia
- ☐ Chorea
- ☐ Spasticity
- ☐ Cerebral palsy
- ☐ Neural tube defect
- ☐ Abnormality of the CNS (Specify below)
- ☐ Other: _____

Growth Parameters

Weight for age: ☐ <3rd % ☐ >97th %
Stature for age: ☐ <3rd % ☐ >97th %
Head circumference: ☐ <3rd % ☐ >97th %

- ☐ Hemihypertrophy
- ☐ Other: _____

Cardiac

- ☐ ASD
- ☐ VSD
- ☐ AV canal defect
- ☐ Coarctation of aorta
- ☐ Tetralogy of fallot
- ☐ Other: _____

Craniofacial

- ☐ Craniosynostosis
- ☐ Cleft lip ☐ Cleft palate
- ☐ Micrognathia ☐ Retrognathia
- ☐ Facial dysmorphism (Specify below)
- ☐ Other: _____

Eye Defects

- ☐ Blindness
- ☐ Coloboma
- ☐ Epicanthus ☐ Hypertelorism
- ☐ Eyelid abnormality (Specify below)
- ☐ Other: _____

Ear Defects

- ☐ Deafness
- ☐ Preauricular ☐ Pit ☐ Skin Tag
- ☐ Low-set ears
- ☐ Outer ear abnormality (Specify below)
- ☐ Inner ear abnormality (Specify below)
- ☐ Other: _____

Cutaneous

- ☐ Hyperpigmentation
- ☐ Hypopigmentation
- ☐ Other: _____

Respiratory

- ☐ Diaphragmatic hernia
- ☐ Lung abnormality (Specify below)
- ☐ Other: _____

Musculoskeletal

- ☐ Upper limb abnormality
- ☐ Lower limb abnormality
- ☐ Camptodactyly (☐ finger / ☐ toe)
- ☐ Syndactyly (☐ fingers / ☐ toes)
- ☐ Polydactyly (☐ finger / ☐ toe)
  - ☐ Preaxial ☐ Postaxial
- ☐ Oligodactyly (☐ finger / ☐ toe)
- ☐ Clinodactyly (☐ finger / ☐ toe)
- ☐ Contractures
- ☐ Scoliosis
- ☐ Vertebral Anomaly
- ☐ Club foot
- ☐ Other: _____

Gastrointestinal

- ☐ Esophageal atresia
- ☐ Tracheoesophageal fistula
- ☐ Gastroschisis
- ☐ Omphalocele
- ☐ Pyloric stenosis
- ☐ Other: _____

Genitourinary

- ☐ Kidney malformation (Specify below)
- ☐ Hydronephrosis
- ☐ Ambiguous genitalia
- ☐ Hypospadias
- ☐ Cryptorchidism
- ☐ Other: _____

Prenatal and Perinatal History

- ☐ Oligohydramnios ☐ Polyhydramnios ☐ IUGR ☐ Premature birth
- ☐ Fetal structural abnormality ☐ Fetal soft markers in obstetric ultrasound (Specify below)
- ☐ Other :_____

Family History

- ☐ Parents with ≥ 3 miscarriages ☐ Consanguinity
- ☐ List health conditions found in family (describe the relationship with proband)

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Completion of Billing Form NOT required for patients with an Ontario Health Card Number.

BILLING FORM

The hospital, referring laboratory, or a patient/guardian will be billed for the services rendered.

- Invoices are sent upon completion of each test/service.
- Contact SickKids' Genome Diagnostics Laboratory at 416-813-7200 x1 with billing inquiries.

How to complete the Billing Form:

- **Referring Physician completes the appropriate section below to specify billing method.**
- **Send requisition and completed "Billing Form" with specimen.**

Option 1: Complete to have the Healthcare Provider billed:

Your Referring Laboratory's Reference #: _____

Billing address of hospital, referring laboratory:

Name: _____

Address: _____

City: _____ Prov/State: _____

Postal/Zip Code: _____ Country: _____

Contact Name: _____

Contact Telephone #: _____

Option 2: Interm Federal Health Program (IFHP)

Submit a copy of the Interim Federal Health Certificate (Refugee Protection Claimant Document) with the photo and UCI# visible for coverage to be confirmed.

UCI# _____

ICD code *(lab use only)*: _____

Option 3: Complete to have Patient/Guardian billed directly:

If you elect to have patient/guardian billed:

- *Patient/Guardian billing information below must be complete; otherwise, the healthcare provider will be billed.*
- *Please advise the patient/guardian to expect a bill from our laboratory.*
- *Provide us with patient's valid credit card information.*
- *Unfortunately, we cannot accept personal checks.*
- ***In this case, the patient/guardian is solely responsible for the charges.***

Relation to patient (check one): ☐ Patient ☐ Guardian/Parent

Method of Payment (check one): ☐ American Express ☐ MasterCard ☐ Visa

Name as it appears on credit card: _____

Credit card # : _____

Expiry date on credit card: _____

CVS#- found on back of card (Required): _____

Mailing Address of Patient/Guardian (if different from requisition):

Name: _____

Address: _____

_____ Apt. #: _____

City: _____ Prov/State: _____

Postal/Zip Code: _____ Country: _____

Additional Contact Information

Patient's phone # with area code:

_____

- or -

Guardian's phone # with area code:

_____

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

RESULT	INTERPRETATION
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/dplm

For more detailed information on microarray technology and its uses, see the pamphlet published by Unique: 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 or the National Society of Genetic Counsellors website at www.nsgc.org

SickKids

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.

This document was developed by The Division of Clinical and Metabolic Genetics and The Division of Genome Diagnostics, Department of Paediatric Laboratory Medicine, The Hospital for Sick Children