UR MEDICINE | Labs

# Specimens:	Blue:	Lav:	Red:	SST:	Grn:	Gray:	Urine	Micro:	*STAT*
Collect Date:	Time:	By:	Depot:	ABN Signed: ☐					
MR #:	A #:								

REQUIRED (PRINT OR PATIENT LABEL)

Name(Last, First, MI)

Date of Birth | Sex:(circle) M F

Street Address

Street Address 2

City, State, Zip

Phone Number | Chart Number

Indicate primary (1) and secondary (2) insurance

__Blue Cross/Shield __Child Health Plus __MVP

__Blue Choice __Medicaid __MVPG

_Medicare Blue Choice __Medicare __Aetna

__Other _____

1. Primary Contract #: _____

Subscriber's Name: _____

Relationship to Subscriber: _____

2. Secondary Contract _____

Subscriber's Name: _____

Relationship to Subscriber:

Doctor: _____

Address: _____

Phone Results to: Fax Results to:

Ordering Provider's Signature

Date of Signature

Diagnosis Mandatory: Signs/Symptoms or ICD10 Codes

If ordered for screening, list test name here and write "SCREENING" after it

Send Additional Reports To: (Full Name/Address)

Compliance is Mandatory and Regulated. For the laboratory to bill properly and receive payment for tests ordered on Medicare Beneficiaries, specific ICD-10 code(s) or a descriptive diagnosis must be included on each patient for each test ordered.It is critical that the diagnosis provided to the lab is consistent with those recorded in the patient medical record on the date of service.

Microarray CGH

Prenatal/Pediatric Specimens

SPECIMEN TYPE

☐ Amniotic Fluid - Call for Requirements (585) 275-1784

☐ CVS - Call for Requirements (585) 275-1784

Date of Ultrasound ___/___/___ GA on Date of Ultrasound ___ weeks ___ days

☐ Peripheral Blood (URMC) Lavender/Green Top/RmTemp (2-5 mL)

☐ POC Sterile Container/Media/Refrig.

INDICATION

☐ Abnormal Maternal Serum/First Trimester Screen - specify _____
☐ Abnormal Ultrasound
☐ Autism
☐ Congenital Anomalies - specify: _____
☐ Developmental Delay
☐ Dysmorphic Features
☐ Family History of Chromosome Abnormality - specify: _____
☐ Failure to Thrive
☐ History of SAB
☐ Intellectual Disability
☐ Neurological disorders specify: _____
☐ Seizures
☐ Other - specify: _____

GENETIC TESTING

CYTOGENETICS

TEST(S) * Patient Consent required

☐ (25789) Chromosome Analysis (Karotype

☐ (16807) FISH N* (Specify_____)

RESTRICTED TEST

(REQUIRES AUTHORIZED PHYSICIAN'S SIGNATUR

MICROARRAY CGH

TEST(S) * Patient Consent required

☐ 4 X 44K

☐ 4 X 180K + SNP

☐ Direct-prenatal, uncultured

☐ Parental FISH Confirmation*

Family h/o of affected, MRN#_____

PATIENT CONSENT

I have read the information on the consent form and discussed it with my health care provider. I have been given the opportunity to ask questions and have them answered about the tests ordered. I authorize collection and analysis of the necessary sample(s).

Patient/Legal Guardian: Date: Health Care Provider:

SEND ORIGINAL REQUISITION WITH SAMPLE

Microarray CGH Testing
Patient Clinical Information Form

The accurate interpretation and reporting of genetic results is contingent upon the reason for referral, clinical information provided, and family history. To help provide the best possible service, please supply the information requested below and send paperwork with the specimen or return by fax to the Microarray Laboratory, Fax (585) 272-9166

Type of Sample: _____

Patient's Name:	**Gender:** ☐ **Male** ☐ **Female**	**Date of Birth:**

Clinical information - please check all that apply

Perinatal History:
- ☐ Prematurity
- ☐ IUGR
- ☐ Oligohydramnios
- ☐ Polyhydramnios
- ☐ Other: _____

Growth:
- ☐ Failure to Thrive
- ☐ Overgrowth
- ☐ Short stature
- ☐ Other: _____

Development:
- ☐ Fine motor delay
- ☐ Gross motor delay
- ☐ Speech delay
- ☐ Other: _____

Cognitive:
- ☐ Learning disability
- ☐ Intellectual Disability
  List IQ/DQ, if known: _____
- ☐ Other: _____

Behavioral:
- ☐ Asperger syndrome features
- ☐ Autism
- ☐ Oppositional-defiant disorder
- ☐ Obsessive-compulsive disorder
- ☐ Pervasive developmental delay
- ☐ Other: _____

Other
- ☐ Other: _____

Neurological:
- ☐ Ataxia/dystonia/chorea
- ☐ Hypotonia
- ☐ Neural tube defect
- ☐ Seizures
- ☐ Spasticity
- ☐ Structural brain anomaly
- ☐ Other: _____

Cardiac:
- ☐ ASD
- ☐ AV canal defect
- ☐ Coarctation of aorta
- ☐ Hypoplastic left heart
- ☐ Tetralogy of Fallot
- ☐ VSD
- ☐ Other: _____

Craniofacial:
- ☐ Cleft lip +/- cleft palate
- ☐ Cleft palate alone
- ☐ Coloboma
- ☐ Craniosynostosis
- ☐ Dysmorphic facial features
- ☐ Ear malformation
- ☐ Macrocephaly
- ☐ Microcephaly

  List HC, if known: _____
- ☐ Other: _____

Cutaneous:
- ☐ Hyperpigmentation
- ☐ Hypopigmentation
- ☐ Other _____

Musculoskeletal:
- ☐ Contractures
- ☐ Club foot
- ☐ Diaphragmatic hernia
- ☐ Limb anomaly
- ☐ Polydactyly
- ☐ Scoliosis
- ☐ Syndactyly
- ☐ Vertebral anomaly
- ☐ Other: _____

Gastrointestinal:
- ☐ Gastroschisis
- ☐ Hirschsprung disease
- ☐ Omphalocele
- ☐ Pyloric stenosis
- ☐ Tracheoesophageal fistula
- ☐ Other: _____

Genitourinary
- ☐ Ambiguous genitalia
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Kidney malformation
- ☐ Undescended testis
- ☐ Urethra/ureter obstruction
- ☐ Other: _____

Family History:
- ☐ Parents with ≥ 2 miscarriages
- ☐ Other relatives with similar clinical history (please explain below)
- ☐ Parental Sample
  Proband _____

Clinical descriptions - please include any additional relevant clinical information not provided above:

Patient Name_____ Date of Birth _____

INFORMED CONSENT FOR GENETIC TESTING

1. Genetic testing will look for changes in the DNA, genes, or chromosomes which may be associated with a specific genetic condition. A positive test result is an indication that the individual may be predisposed to or have the specific disease or condition tested for and may wish to consider further independent testing. If a positive result is obtained, medical and/or genetic counseling follow-up may be advised.

2. As with any test, in some cases, results may not be obtained and a repeat sample may be requested or parental samples may be requested to further understand the findings. When parental testing is performed, it may show changes related to those found in the individual originally tested (usually a child or fetus). In those cases, this information regarding parental test results would become part of the child's (fetus's) test report. Thus it would appear the the child's(mother's) medical record.

3. Some types of genetic testing such as fluorescence in situ hybridization (FISH) are approved by the New York State Department of Health:

   * FISH, which uses DNA probes which bind to a specific region of a chromosome, is helpful in identifying "marker" chromosomes, variations in chromosome structure, or small deletions or dulications on a chromosome.

   * Microarray CGH analysis looks for extra or missing pieces of DNA that are too small to been seen by standard chromosome testing. It uses tiny "DNA probes" to look for thousands of possible changes at the same time. Many results will be negative, but some will find changes that are: (1) associated with known genetic syndromes, (2) not well understood, or (3) "normal variations" in the general population. Because certain types of chromosome changes (translocations, inversions, low level mosaicism, etc.) cannot be detected by array CGH, testing is often performed with standard chromosome analysis.

   * Microarray CGH-SNP analysis looks for extra or missing pieces of DNA described above as well as regions of homozygosity (ROH) throughout the genome. Presences of constitutional ROH are consistent with uniparental idodisomy (UPD), ancestral relatedness, or consanguinity (two individuals who share a common ancestor). Also, CGH-SNP testing may uncover non-paternity, adoption, or consanguinity.

4. FOR URMC PROVIDERS ONLY: Some genetic tests are performed at only a few laboratories; therefore, the sample may need to be sent to a laboratory that is not certified by the New York State Health Department. In these cases, approval for testing will be obtained from New York State.

5. No tests other than those authorized by the patient (or guardian) will be performed on the sample and the sample will be destroyed when testing is complete or not more than sixty days after the sample was taken, unless permission is granted in Section 8 to retain the sample for research.

6. The patient may wish to obtain professional genetic counseling priot to signing this consent.

7. Test results will not be released to anyone other than the referring doctor(s) and the University of Rochester Medical Center Medical Records.

8. The Microarray CGH Laboratory retains patient samples indefinitely for validation, educational purposes, and/or research. The submitted clinical information and test results are also included in a HIPAA-compliant, de-identified public database as part of the National Institute of Health's effort to improve diagnostic testing and the understanding of the relationship between genetic changes and clinical symptoms (for information about the database, visit www.iccg.org). Confidentiality of each sample is maintained. patients may request to withdraw consent for storage of their sample and/or use of the data by: (1) initialing the statement below or (2) calling the laboratory at 585-758-0494 and asking to speak to a Microarray CGH technologist.

_____YES, this sample may be kept as long as the names and other identifying information are removed

_____NO, this sample may not be kept and must be discarded at the end of the testing process.

The patient (or legal counsel) is required to sign the consent prior to genetic testing being performed.
Therefore, please sign this form at the bottom of page 1.

Microarray (page 4 of 4)

Patient Name_____ Date of birth_____

Chromosomal Microarray Testing and the ISCA Consortium Database

Your doctor has ordered chromosomal microarray as part of your or your child's medical evaluation. This test is used to look for a genetic cause of problems in the physical, intellectual and behavioral development of children and adults. The chromosomal microarray has significantly improved our ability to find the underlying cause of many developmental and medical concerns, allowing families to learn about specific result and make informed plans for medical and/or educational interventions.

The chromosomal microarray is away for the laboratory to look at the entire genetic make-up of a person in order to find missing (loss) or extra (gain) pieces of the chromosomes. Many of the losses and gains found by microarray are common and have a well understood pattern of medical problems, However, some microarray results are rare and have not been seen before. In some cases, it is not a problem or not, or what medical problems may be expected. Parental testing can sometimes help to clarify this type of uncertain result.

More information is needed to understand rare results and you can help!

Your or your child's sample is being sent to URMC's Microarray CGH lab for microarray testing. URMC_Labs is a member of the International Standard Cytogenomic Array (ISCA) Consortium, an organization of more than 85 laboratories working together to gather the information needed to understand the meaning of rare chromosomal microarray results. For each order received for microarray testing, URMC_Labs contributes the microarray result along with the reason for testing (such as autism or heart defect) to the central ISCA Consortium database.

Privacy is of the utmost importance to us, therefore all patients identity information is removed (de-identified) before results are submitted. Your confidentiality is maintained.

The ISCA Consortium database is only possible through the contribution of individual patient results. With your help, as the ISCA database grows over time, laboratories will be able to use the information to improve the reporting of rare results. Patients with an uncertain result may then learn the true meaning of their microarray result and optimize their medical care.

To Opt-Out of the ISCA Database

There are three ways to opt-out:

1. Check the opt-out box on page 3 section 8 of test requisition form.
2. Calling client services at 1-800-747-4769 option 3
3. Calling the Microarray CGH Laboratory at 585-758-0494

Please call either client services or the Microarray CGH Laboratory at the numbers above if you have questions about the use of your information or sample.