



# Specimens:	Blue	Lav:	Red:	SST:	Grn:	Gray:	Urine:	Micro:
Collect Date:	Time:		By:	Depot:		ABN Signed: <input type="checkbox"/>		
MR #:	A #:							

**\*STAT\***

**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 \_\_\_\_\_

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)

2 OraCollect saliva swabs for DNA (Room Temp)

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 \*Patient Consent Required**

**CYTOGENETICS**

TEST(S)

(25789) Chromosome Analysis (Karyotype)

(16807) FISH N\* (Specify \_\_\_\_\_)

RESTRICTED TEST  
(REQUIRES AUTHORIZED PHYSICIAN'S SIGNATURE)

**MICROARRAY CGH**

4 X 180K + SNP (CNV Platform)

Direct-prenatal, uncultured

Parental MaCGH confirmation test  
 Report additional findings

Parental FISH Confirmation\*  
Family h/o of affected, MRN# \_\_\_\_\_

**PATIENT CONSENT**

*I have read the information on the back of this form and discussed it with my health care provider and 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: \_\_\_/\_\_\_/\_\_\_

Initial here if de-identified sample may be used for research: \_\_\_\_\_

**HEALTHCARE PROVIDER CONSENT**

*I attest that I have reviewed the requirements for genetic testing order on the requisition with the patient. I have conveyed the required information to the patient and obtained consent*

Health Care Provider: \_\_\_\_\_

Date: \_\_\_/\_\_\_/\_\_\_

**SEND ORIGINAL REQUISITION WITH SAMPLE**

## 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 child's(mother's) medical record.
3. Some types of genetic testing 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 duplications on a chromosome.
  - \* Microarray CGH analysis looks for extra or missing pieces of DNA that are too small to be 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 isodisomy (UPD), ancestral relatedness, or consanguinity (two individuals who share a common ancestor). Also, CGH-SNP testing may uncover non-paternity, adoption, or consanguinity.
  - \* Chromosome genome mapping identify DNA structural variants (SVs), namely, insertions, deletions, and duplications as well as inversion and translocations (balanced and unbalanced), losses and gains of whole chromosomes or segments of chromosomes. These constitutional chromosomal aberrations are of clinical relevance to: (1) Diagnose (2) Indicate a greater likelihood of developing a disease or condition (3) Establish eligibility for a specific treatment, and/or (4) Provide prognostic information that influences patient management/treatment decisions, (5) Provide information on treatment adherence.
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 to retain the sample for research.
6. The patient may wish to obtain professional genetic counseling prior 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. If consent for research is obtained, the Microarray CGH Laboratory retains patient samples for validation, educational purposes, and/or research in the approved DNA/Cell Repository (STUDY00007544). 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](http://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 at the bottom of page one(2) calling the laboratory at 585-758-0494 and asking to speak to a Microarray CGH technologist or calling Client Services at 1-800-747-4769, option3.

Consent is required from the patient (or legal counsel)  
prior to genetic testing being performed

Therefore, please sign this form at the bottom of page 1.

# 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:** \_\_\_\_\_

<b>Patient's Name:</b>	<b>Gender:</b> <input type="checkbox"/> Male <input type="checkbox"/> Female	<b>Date of Birth:</b>
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**Clinical information - please check all that apply**

<p><u>Perinatal History:</u></p> <p><input type="checkbox"/> Prematurity</p> <p><input type="checkbox"/> IUGR</p> <p><input type="checkbox"/> Oligohydramnios</p> <p><input type="checkbox"/> Polyhydramnios</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Growth:</u></p> <p><input type="checkbox"/> Failure to Thrive</p> <p><input type="checkbox"/> Overgrowth</p> <p><input type="checkbox"/> Short stature</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Development:</u></p> <p><input type="checkbox"/> Fine motor delay</p> <p><input type="checkbox"/> Gross motor delay</p> <p><input type="checkbox"/> Speech delay</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Cognitive:</u></p> <p><input type="checkbox"/> Learning disability</p> <p><input type="checkbox"/> Intellectual Disability</p> <p>List IQ/DQ, if known: _____</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Behavioral:</u></p> <p><input type="checkbox"/> Asperger syndrome features</p> <p><input type="checkbox"/> Autism</p> <p><input type="checkbox"/> Oppositional-defiant disorder</p> <p><input type="checkbox"/> Obsessive-compulsive disorder</p> <p><input type="checkbox"/> Pervasive developmental delay</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Other</u></p> <p><input type="checkbox"/> Other: _____</p>	<p><u>Neurological:</u></p> <p><input type="checkbox"/> Ataxia/dystonia/chorea</p> <p><input type="checkbox"/> Hypotonia</p> <p><input type="checkbox"/> Neural tube defect</p> <p><input type="checkbox"/> Seizures</p> <p><input type="checkbox"/> Spasticity</p> <p><input type="checkbox"/> Structural brain anomaly</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Cardiac:</u></p> <p><input type="checkbox"/> ASD</p> <p><input type="checkbox"/> AV canal defect</p> <p><input type="checkbox"/> Coarctation of aorta</p> <p><input type="checkbox"/> Hypoplastic left heart</p> <p><input type="checkbox"/> Tetralogy of Fallot</p> <p><input type="checkbox"/> VSD</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Craniofacial:</u></p> <p><input type="checkbox"/> Cleft lip +/- cleft palate</p> <p><input type="checkbox"/> Cleft palate alone</p> <p><input type="checkbox"/> Coloboma</p> <p><input type="checkbox"/> Craniosynostosis</p> <p><input type="checkbox"/> Dysmorphic facial features</p> <p><input type="checkbox"/> Ear malformation</p> <p><input type="checkbox"/> Macrocephaly</p> <p><input type="checkbox"/> Microcephaly</p> <p>List HC, if known: _____</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Cutaneous:</u></p> <p><input type="checkbox"/> Hyperpigmentation</p> <p><input type="checkbox"/> Hypopigmentation</p> <p><input type="checkbox"/> Other: _____</p>	<p><u>Musculoskeletal:</u></p> <p><input type="checkbox"/> Contractures</p> <p><input type="checkbox"/> Club foot</p> <p><input type="checkbox"/> Diaphragmatic hernia</p> <p><input type="checkbox"/> Limb anomaly</p> <p><input type="checkbox"/> Polydactyly</p> <p><input type="checkbox"/> Scoliosis</p> <p><input type="checkbox"/> Syndactyly</p> <p><input type="checkbox"/> Vertebral anomaly</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Gastrointestinal:</u></p> <p><input type="checkbox"/> Gastroschisis</p> <p><input type="checkbox"/> Hirschsprung disease</p> <p><input type="checkbox"/> Omphalocele</p> <p><input type="checkbox"/> Pyloric stenosis</p> <p><input type="checkbox"/> Tracheoesophageal fistula</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Genitourinary</u></p> <p><input type="checkbox"/> Ambiguous genitalia</p> <p><input type="checkbox"/> Hydronephrosis</p> <p><input type="checkbox"/> Hypospadias</p> <p><input type="checkbox"/> Kidney malformation</p> <p><input type="checkbox"/> Undescended testis</p> <p><input type="checkbox"/> Urethra/ureter obstruction</p> <p><input type="checkbox"/> Other: _____</p> <p><u>Family History:</u></p> <p><input type="checkbox"/> Parents with <math>\geq 2</math> miscarriages</p> <p><input type="checkbox"/> Other relatives with similar clinical history (please explain below)</p> <p><input type="checkbox"/> Parental Sample</p> <p>Proband _____</p>
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**Clinical descriptions - please include any additional relevant clinical information not provided above:**