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

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 *Patient Consent Required

CYTOGENETICS

TEST(S)

☐ (25789) Chromosome Analysis (Karotype)

☐ REFLEX to MicroArray

☐ (16807) FISH N* (Specify_____)

RESTRICTED TEST
(REQUIRES AUTHORIZED PHYSICIANS' 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 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 and results shared with other parties, if needed: _____

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 ontained 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 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.
   * Chromosome genome mapping identify DNA structural variants (SVs), namely, insertions, deletions, and duplications as well as inversrion and translocations (balanced and unbalanced), losses and gains of whole chromosomes or segments of chromosomes. These constitutional chromosomal abberations 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) Provde prognostic information that influences patient management/treatment decions, (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). 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: _____

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: