MAYO CLINIC
Mayo Medical Laboratories

SPECIMEN REQUIREMENT REMINDER
NOTIFICATION DATE: June 9, 2010
EFFECTIVE DATE: immediately

Array Comparative Genomic Hybridization (aCGH), Whole Genome, Constitutional #88898

EXPLANATION: 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 and to ensure the best interpretation of patient results, the "Cytogenetics-Array CGH Testing Patient Information Sheet" (Supply T665) is required for all orders.

WHY THIS INFORMATION IS NEEDED:
Not only is this information used in the actual result interpretation for your patient, the Mayo Clinic Cytogenetics laboratory is an active member of the International Standard Cytogenomic Array (ISCA) Consortium, an organization of more than 100 laboratories working together to gather the information needed to understand the meaning of rare chromosomal microarray results. The Mayo Clinic Cytogenetics Laboratory contributes the microarray test result, along with the reason for testing (such as autism or heart defect), to the central ISCA Consortium database for every microarray test performed.

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

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 this information to improve the reporting of rare results and new research may be sparked. Patients with an uncertain result may then learn the true meaning of their microarray result and optimize their medical care.

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710

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Cytogenetics – Array CGH Testing Patient Information Sheet

Instructions: 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, supply the information requested below and **send paperwork with the specimen or return by fax to the Cytogenetics Laboratory, attn: Genetic Counselors, 507-284-0043 (phone 507-538-2952).**

Patient Name *(First, Middle, Last)*	Birth Date *(Month DD, YYYY)*	Sex ☐ Male ☐ Female

Clinical Information – 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
- ☐ Mental retardation
  List IQ/DQ, if known:_____
- ☐ Other: _____

Behavioral
- ☐ Asperger syndrome features
- ☐ Autism
- ☐ Oppositional-defiant disorder
- ☐ Obsessive-compulsive disorder
- ☐ Pervasive developmental delay
- ☐ 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 (explain below)

Clinical Descriptions – include any additional relevant clinical information not provided above

As a participant in the ISCA (International Standard Cytogenomic Array) Consortium, Mayo Clinic Cytogenetics Laboratory contributes submitted clinical information and test results to a HIPAA-compliant, de-identified public database as part of the NIH's effort to improve our understanding of the relationships between genetic changes and clinical symptoms. Confidentiality is maintained. Patients may request to opt-out of this scientific effort by: 1) checking the box below, 2) calling the laboratory at **1-800-533-1710, extension 8-2952** and asking to speak with a laboratory genetic counselor. Please call with any questions.

☐ Refusal for inclusion in these efforts may be indicated by checking this box. (If the box is not marked, the data will be anonymized and submitted.)

MML Supply T665
MC1235-166

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Chromosomal Microarray Testing and the ISCA Consortium Database

Your doctor has ordered a chromosomal microarray as part of a 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 their specific result and make informed decisions for medical and/or educational interventions.

The chromosomal microarray is a way for the laboratory to look at all of a person's genetic material 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 well understood patterns of associated medical concerns. However, some microarray results are rare and have not been seen before. In some of these cases, it is not possible to know if a particular microarray result will cause 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!

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To Opt-Out of the ISCA Database

If you do not want your results to be submitted to the ISCA Consortium database, you can "opt-out" of participation. If you opt-out, you can still have microarray testing and it will not affect your results.

There are three ways to opt-out:

1. Ask your doctor to check the opt-out box on the test requisition form or patient information sheet.

2. Call the laboratory and ask to speak with a laboratory genetic counselor: 507-538-2952 (800-533-1710, extension 8-2952).

3. Check the opt-out box on your final microarray report and fax to 877-749-3332, Attn: Cytogenetics laboratory genetic counselors.

Please call if you have questions about the use of your information.

To learn more about the ISCA Consortium visit:

https://isca.genetics.emory.edu

or call 800-533-1710 extension 8-2952

200 First Street SW • Rochester, MN 55905 • 507.538.2952 • 800.533.1710 ext. 8.2952 • FAX 877.749.3332 • GCCytogenetics@mayo.edu

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