MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: January 4, 2013
EFFECTIVE DATE: February 4, 2013

Chromosomal Microarray, Congenital, Blood

Test ID: CMAC

USEFUL FOR:

- First-tier, postnatal test for individuals with multiple anomalies that are not specific to well-delineated genetic syndromes, apparently nonsyndromic developmental delay or intellectual disability, or autism spectrum disorders as recommended by the American College of Medical Genetics (ACMG).
- An appropriate follow-up test for individuals with unexplained developmental delay/intellectual disability, autism spectrum disorders, or congenital anomalies with a previously normal conventional chromosome study.
- Determining the size, precise breakpoints, gene content, and any unappreciated complexity of abnormalities detected by other methods such as conventional chromosome and FISH studies.
- Determining if apparently balanced abnormalities identified by previous conventional chromosome studies have cryptic imbalances, since a proportion of such rearrangements that appear balanced at the resolution of a chromosome study are actually unbalanced when analyzed by higher-resolution chromosomal microarray.
- Assessing regions of homozygosity related to uniparental disomy or identity by descent.

METHODOLOGY: Chromosomal Microarray (CMA)

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: **Specimens must arrive within 96 hours of draw.**

A whole blood EDTA and a whole blood sodium heparin specimen, as well as the reason for referral, are required.

Specimen Type: Whole blood
Container/Tube: Green top (sodium heparin) **and** lavender top (EDTA)
Specimen Volume: 3 mL for each tube
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tubes.

NOTE:

- **New York Clients-Informed consent is required.** Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
- Cytogenetics-Array CGH Testing Patient Information Sheet (Supply T665) in Special Instructions

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Refrigerated	

CAUTIONS:

- This test does not detect balanced chromosome rearrangements such as Robertsonian or other reciprocal translocations, inversions, or balanced insertions.
- This test does not detect all types and instances of uniparental disomy.
- This test is not designed to detect mosaicism, although it can be detected in some cases.
- This test does not detect point mutations, small deletions or insertions below the resolution of this assay, or other types of mutations such as epigenetic changes.
- The results of this test may be of uncertain clinical significance. In such cases, studies of additional family members may be required to help interpret the results.

CPT CODE: 81229-Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities

DAY(S) SET UP: Monday-Thursday 6:30am-1:30pm
Friday 6:30am-5:00pm

ANALYTIC TIME: 8 days

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
SignatureChipOS	ZW229	2005	Signature Genomics
Chromosomal Microarray Analysis - HR + SNP Screen (Comprehensive)	ZW152	8665	Baylor Cytogenetics Laboratory

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