



CENTER FOR
GENOMIC
MEDICINE



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RapidSure
MICROARRAY

COST - EFFECTIVE
FIRST-TIER
Testing Option

LET'S TAKE THE FIRST STEP TOGETHER TOWARDS FINDING THE ANSWER FOR YOUR PATIENT'S HEALTH CONUNDRUM.

Many patients face important decisions regarding genetic testing options for their families. For parents who are considering Chromosomal Microarray Analysis (CMA) for their child, NCGM offers a cost-effective first-tier testing option that analyzes changes in chromosomes which are not detectable by karyotyping.

INDICATIONS FOR TESTING (PRENATAL)

Microarray has been recommended by the American College of Obstetrics and Gynecology (ACOG) for :

- XX Abnormal ultrasound findings
- XX Abnormal screening tests
- XX Family history of a genetic or chromosomal abnormality that is detectable by microarray technology
- XX History of pregnancy loss
- XX For any patient undergoing invasive testing



Maternal cell contamination testing is recommended for all prenatal arrays. 4 ml maternal blood in EDTA required for the same

FIRST TIER TESTING RECOMMENDED BY THE EXPERTS (POSTNATAL)

Chromosomal MicroArray (CMA) is a first-tier diagnostic test recommended by the American College of Medical Genetics (ACMG), the American Academy of Neurology (AAN), the American Academy of Pediatrics (AAP) for :

- XX Intellectual disability, development delay
- XX Autism spectrum disorder
- XX Multiple congenital anomalies
- XX Dysmorphism



TYPES OF ARRAY








Microarray RapidSure Optima (315K)

Microarray RapidSure Deepdive (750K)







ADVANTAGES

-  Detects aneuploidy (including trisomy and sex chromosome abnormalities) & triploidy
-  Identifies sub-microscopic deletions and duplications in regions known to be associated with well-characterized microdeletion & microduplication syndromes
-  Provides enriched coverage of subtelomeric regions, often undetectable by traditional chromosome analysis
-  More comprehensive and cost-effective than individual FISH tests
-  Circumvents cell culture in most cases with more efficient turn-around times.
-  Often allows for results on suboptimal specimens for which chromosome analysis is not feasible
-  SNP array allows detection of Triploidy, Uniparental disomy & LOH

LIMITATIONS

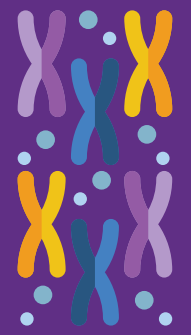
-  The minimum resolution required for the reporting of Microarray RapidSure Optima (315K) is 1 MB for losses, 2 MB for gains, and 5 MB for LOH / AOH for Microarray RapidSure Optima (315K).
-  The minimum resolution required for the reporting of Microarray RapidSure Deepdive (750K) is 200 KB for losses, 200 KB for gains, and 5 MB for LOH / AOH for Microarray RapidSure Deepdive (750K).

POTENTIAL TEST RESULTS



A NORMAL RESULT indicates no clinically-significant chromosome abnormalities were identified.

ABNORMAL RESULTS are reported by location in the genome, including chromosome and size.



DELETION : Part of a chromosome (genetic material) is missing. Some may be very small and only include one gene and others are bigger and may involve numerous genes.

DUPLICATION : Extra chromosome material is present in the patient's DNA.

AN ABNORMAL RESULT indicates that a clinically significant chromosome abnormality was identified that may provide an explanation for the indications.

Detected Copy Number Variants (CNV) are classified based on the American College of Medical Genetics and Genomics as

- Pathogenic
- Likely Pathogenic
- Variants of uncertain significance (VUS)

Benign and Likely Benign CNV's are not reported.

SAMPLE TYPE

- Amniotic fluid : 15 ml in a sterile tube(s)
- Blood : 4ml / EDTA
- Fetal product in normal saline with antibiotics

TURN AROUND TIME (TAT)

7 working days

REQUIRED FORM

The following form can be downloaded via our website

- CMA Consent form
- Form G (Prenatal)
- Form E (Prenatal)

REFERENCES

- XX The use of chromosomal microarray analysis in prenatal diagnosis. Committee Opinion No. 581. American College of Obstetricians and Gynecologists. ObstetGynecol 2013; 122:1374-1377.
- XX Richards et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med advance online publication 5 March 2015; 1-20.
- XX Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis Llui's Armengol Julia'n Nevado Clara Serra-Juhe' Alberto Plaja Carmen Mediano Fe Amalia Garcia- Santiago Manel Garcia-Aragone's Olaya Villa Elena Mansilla
- XX Abnormalities in spontaneous abortions detected by G-banding and chromosomal microarray analysis (CMA) at a national reference laboratory Boris T Wang, Thomas P Chong, Fatih Z Boyar, Kimberly A Kopita, Leslie P Ross, Mohamed M El-Naggar, Trilochan Sahoo, Jia-Chi Wang, Morteza Hemmat, Mary H Haddadin, Renius Owen and Arturo L Anguiano
- XX Comprehensive genetic analysis of pregnancy loss by chromosomal microarrays: outcomes, benefits, and challenges Trilochan Sahoo, MD, FACMG1, Natasa Dzidic, MS1, Michelle N. Strecker, MS1, Sara Commander, MS1, Mary K. Travis, MS1, Charles Doherty, MS1, R. Weslie Tyson, MD2, Arturo E. Mendoza, MD3, Mary Stephenson, MD4, Craig A. Dise, MD5, Carlos W. Benito, MD5, Mandolin S. Ziadie, MD6 & Karine Hovanes, PhD1

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