CHROMOSOMAL MICROARRAY
TESTING TO IDENTIFY CLINICALLY SIGNIFICANT CHROMOSOMAL ABNORMALITIES
CHROMOSOMAL MICROARRAY TESTING

Chromosomal microarray (CMA) uses the latest technology to provide expanded genetic testing for a large number of serious genetic disorders not detected by routine chromosome analysis. By analyzing more than 1.9 million copy number probes and 750,000 SNP probes, we can assess deletions and duplications, determine their precise breakpoints and gene content, and detect regions of homozygosity.

WHY USE MAYO MEDICAL LABORATORIES?

EXPERTISE IN ANALYSIS AND INTERPRETATION

CMA testing generates large amounts of complex data that require a high-level of expertise to accurately assess and classify each abnormality detected. Our laboratory directors review thousands of CMA tests each year and are actively involved in the field of cytogenetics, helping to create testing standards and shape practice guidelines. Every CMA test performed at Mayo Clinic is supported by the experience and expertise of our laboratory directors, ensuring we not only provide test results but clinically actionable interpretations.

Benefits of Mayo Clinic Testing

- Manual review of software output to eliminate common errors such as misidentification, background noise, and failure to detect mosaicism
- Examination of every abnormality regardless of size to determine the importance and relevance of gene content rather than a strict reliance on reporting size criteria
- Integration of historical data on common benign polymorphisms and previously reported abnormalities to improve accuracy of interpretation
- Extensive literature and database searches to ensure accurate and up-to-date classification of abnormalities
- Performance of parental studies, when necessary, to determine whether the chromosomal abnormality is inherited or de novo to help determine if it is benign or pathogenic and if future pregnancies are at risk
- Determination of maternal cell contamination to assess if fetal (not maternal) DNA is available for analysis from prenatal specimens
WHEN SHOULD I ORDER CMA?

AS A REPLACEMENT FOR FETAL KARYOTYPE

The American Congress of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine recommend CMA as a replacement for the fetal karyotype in patients with a pregnancy demonstrating one or more major structural abnormalities on ultrasound when undergoing invasive prenatal diagnosis.

IN CASES OF INTRAUTERINE FETAL DEMISE OR STILLBIRTH

ACOG also recommends CMA analysis of fetal tissue (e.g., amniotic fluid, placenta, or products of conception) when further cytogenetic analysis is desired in cases of intrauterine fetal demise or stillbirth. CMA testing increases the likelihood of obtaining results and improves detection of causative abnormalities. This testing is also available on both fresh and formalin-fixed paraffin-embedded tissue expanding the clinical situations in which this testing can be used.

IN CASES OF DEVELOPMENTAL DELAY AND BIRTH DEFECTS

The American College of Medical Genetics recommends CMA as a 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.

CMA is also an appropriate follow-up test for individuals with congenital anomalies with a previously normal conventional chromosome study.

CONSULT WITH MAYO CLINIC GENETIC COUNSELORS ABOUT TESTING OPTIONS

Genetic counselors bring real value to proper test utilization by making sure the most appropriate test has been ordered. On average, Mayo Clinic genetic counselors modify 8% of all reviewed genetic test orders, reducing health care costs and benefiting patient care.¹

Mayo Clinic genetic counselors are available to:

- Support the ordering process.
- Provide additional information about testing options.
- Offer results interpretation.
- Assist with case review and coordination.

REFERENCES

1. Wapner RJ et al., Chromosomal Microarray versus Karyotyping for Prenatal Diagnosis. NEJM. 2012; 367(23): 2175-84.
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FOR MORE INFORMATION ABOUT CMA TESTING, VISIT
MayoMedicalLaboratories.com/CMA