



Chromosomal Microarray (CMA)

What is Chromosomal Microarray (CMA)?

Chromosomal Microarray (CMA) is a high resolution genomic test that detects submicroscopic chromosomal imbalances, including copy number variations (CNVs) and regions of loss of heterozygosity, which may not be identified by conventional karyotyping.

Prenatal

Analysis of fetal samples (CVS / amniotic fluid) to detect clinically significant micro deletions/ duplications, and numerical aberrations enabling early identification of chromosomal abnormalities.

Recurrent Pregnancy Loss

Evaluation of products of conception (POC) to identify chromosomal causes of miscarriage, supporting accurate diagnosis and future pregnancy planning.

Postnatal

Assessment of individuals with developmental delay, intellectual disability, autism spectrum disorders, or congenital anomalies for precise detection of genomic imbalances.

What genetic conditions can CMA diagnose?

Microdeletion syndromes (e.g., DiGeorge, Williams, Prader-Willi/Angelman)

Microduplication syndromes (e.g., 16p11.2, 22q11.2, 17q12 duplications)

Telomeric imbalances

Aneuploidies (e.g., Trisomy 21, Turner syndrome)

Who can consider testing ?

- Children with developmental delay, intellectual disability or Autism spectrum disorder
- Patients with multiple congenital anomalies
- Individuals with unexplained dysmorphic features
- Couples with recurrent pregnancy loss
- Prenatal cases with abnormal ultrasound findings

Available in both 315K and 750K array resolutions to suit varying clinical requirements and depth of analysis.

Technical Specifications

- Genome-wide analysis for detection of chromosomal imbalances
- High-resolution platform enabling identification of submicroscopic genomic alterations



Technology

- 750K BeadChip
- ~750,000 markers covering ~9,000 genes, including ~447 disease associated genes



Capabilities

- ~2.3 kb resolution for CNV detection
- CN-LOH detection



Coverage

- 324 cytogenetic hotspot regions
- ~495,000+ genomic variant



Turn Around Time

3 weeks



Sample type

AF/CVS/POC/Blood in EDTA

Also covers: Pericentromeres & telomeres, Sex chromosomes, PAR regions (PAR1 & PAR2), Known syndrome-associated regions.

*Test Performed at NABL and CAP Accredited Labs

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