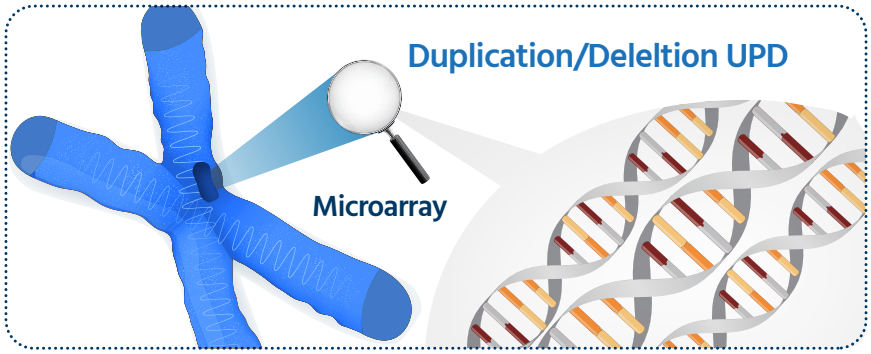
Genome-wide cytogenetic analysis
for early and precise answers

Chromosomal Microarray

What is CMA?



Chromosomal Microarray (CMA) can detect the most known cytogenetic disease among the existing genetic tests.
It can identify Chromosomal Abnormalities that cause developmental disabilities, mental retardation, autism, and multiple congenital malformations that could not be detected using previous techniques such as Karyotyping.

ACMG American College of Medical Genetics and Genomics

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN®

Several famous groups (ACMG, AAP)guideline recommend

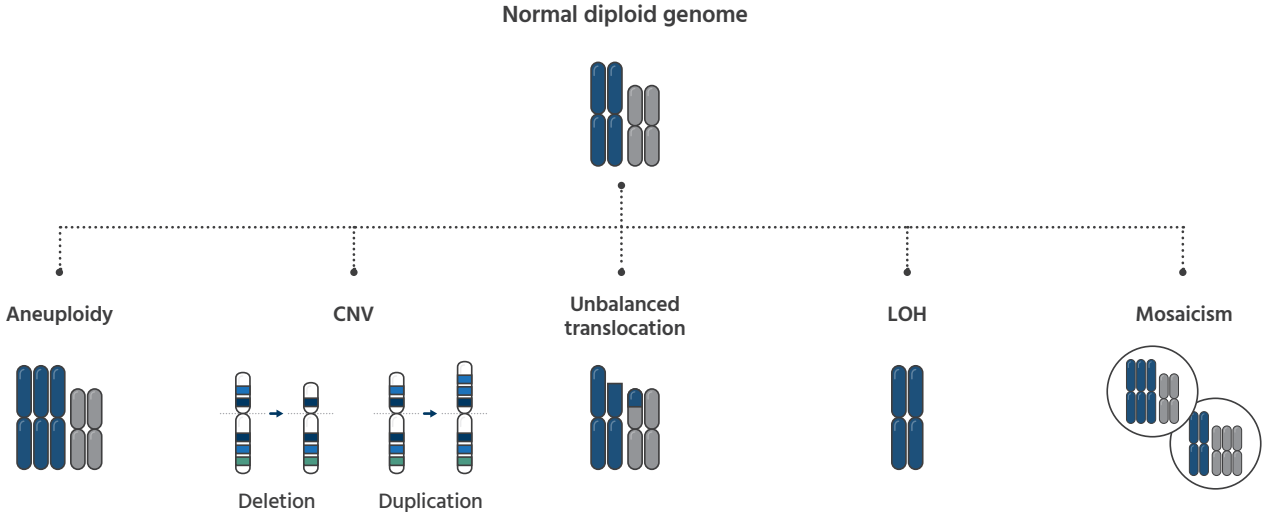
CMA as the first genetic test to order for a patient with autism, developmental delay, intellectual disabillity and/or multiple congenital anomalies.

- Accurate and comprehensive analysis of chromosomal abnormalities
- Can lead to direct diagnosis of chromosomal disease.
- Accurate location, size, and genetic information of chromosomal abnormalities
- Genetic Counseling required by family members

According to **ACMG guideline**,
CMA is recommended as a 1st-tier test for developmental disorders, mental retardation, autism, multiple congenital malformations due to small size of DNA deletion/duplication which cannot be detected by conventional karyotyping test.

Manning, M., & Hudgins, L. (2010, November). Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genetics in Medicine, 12(11), 742–745. https://doi.org/10.1097/gim.0b013e3181f8baad

What can we know from CMA?

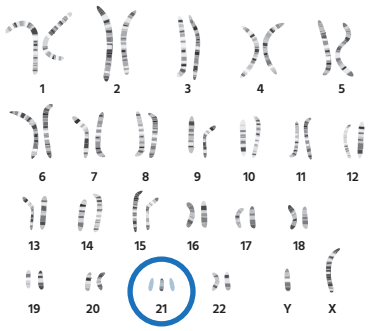
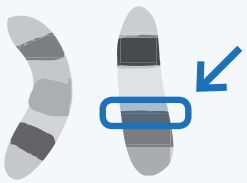


- Aneuploidy
- CNV (Copy Number Variation): Deletion, Duplication
- Unbalanced translocation
- LOH (Loss of Heterozygosity): LOH is only reported when the size is sufficiently suspicious for UPD (Uniparental Disomy).
- Mosaicism: Low level mosaicism may not be detected.

Genome-wide cytogenetic analysis
for early and precise answers

Chromosomal Microarray

How is it different from Karyotyping?

	Karyotyping	CMA
Resolution	Low (5~10Mb)	High (25~400kb)
Cell cultured	Required	Not required
Diagnosis rate	2~3%	15~20%
Target diseases	Down syndrome, Edward syndrome, Patau syndrome	Development disorders, Mental retardation, Autism, Multiple congenital anomalies etc.
Diagnosis example	Detect numerical or structural changes  Down Syndrome : Chromosome 21 Trisomy	Observe the microscopic duplication or deletion of chromosomes at higher resolution  22q11 Deletion Syndrome : Chromosome 22 partial deletion
Analytical time	Long for cell culture	Fast

CMA can detect small-sized chromosomes that are difficult to identify through conventional karyotyping, allowing for the detection of microdeletion/duplication of the copy number variation (CNV).
It enables precise identification of the exact location of mutations, leading to more accurate predictions of the severity of genetic disorder using software for objective result analysis.

Service features

Test	Chromosomal Microarray (CMA)	**Code**	OM001
Specimen	EDTA WB 3 ml	**TAT**	9 days
Method	Microarray	**Sample Storage**	Room temperature (Refrigerated is recommended.)
Test description	This test can detect copy number variation (CNV) (deletion >25kb, duplication >50kb) and loss of heterozygosity (LOH) (>3Mb) across the entire genome. It reports all CNVs of >400kb and clinically significant CNVs of any size. Also, LOH is also considered to be reported when associated with clinically significant situations such as uniparental disomy (UPD) or consanguinity.		
Caution & Limitation	• Low levels of mosaicism, balanced translocation, inversion, and point mutations cannot be detected. • If the LOH area is small or is a heterodisomic UPD, UPD may not be detected on the test principle. • Undetectable CNV area: 13p, 14p, 15p, 21p, 22p, Yq11.23, Yq12 and pericentric heterochromatin regions of all chromosomes		

GC Genome
Cell Center Bldg 1F, 107, Ihyeon-ro 30beon-gil, Giheung-gu, Yongin-si, Gyeonggi-do, Republic of Korea (16924)
Online : www.gc-genome.com| **Contact** : +82) 31-280-9910

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS