

 PEDIATRIC / PRENATAL

Baylor Genetics offers Chromosomal Microarray Analysis (CMA), a cost-effective first-tier testing option that analyzes specific changes in individual segments of DNA. Let's take the first step together towards finding the answer to your patient's health condition.

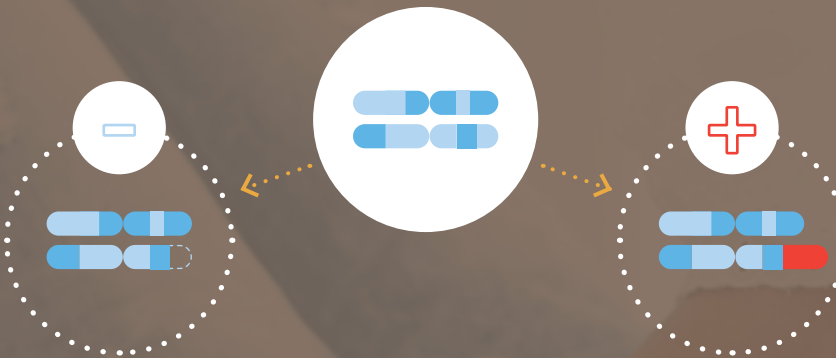
**BAYLOR**  
GENETICS

**CMA**  
Chromosomal  
Microarray Analysis

The first step towards  
finding the right answer  
with Chromosomal  
Microarray Analysis

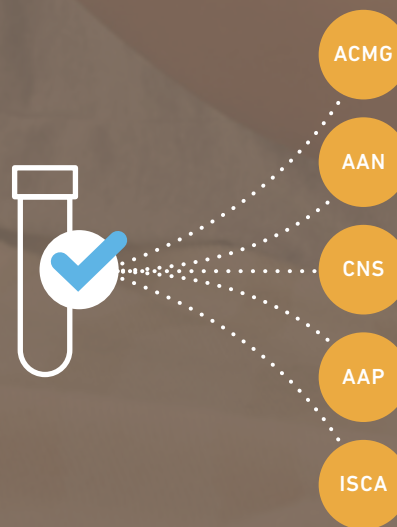
## Baylor Genetics was one of the first laboratories to offer CMA testing

Chromosomal microarray analysis (CMA) provides comprehensive genetic testing for the most common chromosomal conditions as well as a large number of severe genetic conditions not detected by traditional chromosome analysis. This test explores chromosomes in detail to help detect genetic conditions that cause significant disabilities. Baylor Genetics evaluates the entire human genome for regions that contain too many or too few copies of genetic material.



The microarray is like a grid covered with thousands of tiny probes consisting of small pieces of DNA from known locations on each of the 46 chromosomes. CMA looks for imbalances of chromosomal material between DNA from a control and your patient's DNA. When a patient's sample and the control sample are labeled and added to the microarray, our team can determine if there are any differences in copy number, also known as gains (duplications) or losses (deletions) in specific segments of DNA. If a difference is found, the location and type of change (gain or loss) will often determine the cause of your patient's health condition.

CMA is a first-tier diagnostic test recommended by the American College of Medical Genetics (ACMG), the American Academy of Neurology (AAN), the Child Neurology Society (CNS), the American Academy of Pediatrics (AAP), and the International Standard Cytogenomic Array (ISCA) Consortium. CMA should be considered for individuals who lack a sufficient specific history or features on physical examination to suggest a specific genetic (or non-genetic) cause for intellectual disability, developmental delay, autism spectrum disorder, or multiple congenital anomalies.





# Postnatal CMA Testing Options

## CMA-HR + SNP SCREEN (Comprehensive)

TEST CODE

8665

TURNAROUND TIME

14  
CALENDAR  
DAYS

High-resolution (HR) copy number analysis + SNPs for detection of absence of heterozygosity (AOH) & uniparental disomy (UPD)

Custom Baylor design – 400K Agilent

This level of detail is ONLY available on the Baylor Genetics CMA-HR + SNP screen

### BENEFITS

Maximum sensitivity for detection of gains and losses

Exon-by-exon coverage of over 4,200 clinically significant genes

Whole genome backbone coverage at a 30 Kb resolution

Tiling coverage of mitochondrial genome

57,000 oligos used for the detection of absence of heterozygosity (AOH) associated with uniparental disomy (UPD) or consanguinity

### LIMITATIONS

AOH less than 10 Mb in size will not be reported

The heterodisomy detection rate is not currently known for this assay

## CMA-HR

TEST CODE

8655

TURNAROUND TIME

14  
CALENDAR  
DAYS

High-resolution (HR) copy number analysis

Custom Baylor design – 180K Agilent

### BENEFITS

Lower cost array option

High sensitivity for detection of gains and losses

Exon-by-exon coverage of over 1,700 genes

Tiling coverage of mitochondrial genome

Whole genome backbone coverage at a 30 Kb resolution

### LIMITATIONS

Does not detect AOH, UPD, or consanguinity

Does not have the highest level of exon-by-exon coverage available

## CMA-SNP

TEST CODE

8650

TURNAROUND TIME

28  
CALENDAR  
DAYS

Excellent SNP coverage

Affymetrix-CytoScan® HD

### BENEFITS

Highly accurate detection of AOH associated with UPD or consanguinity

Copy number coverage for classical deletion/duplication syndromes and detection of novel variants over 100 Kb

### LIMITATIONS

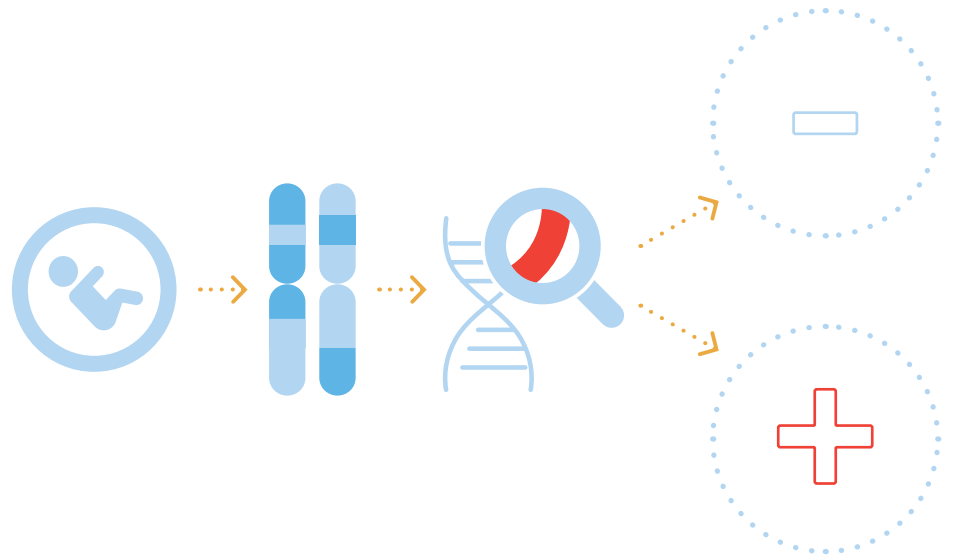
AOH less than 5 Mb in size will not be reported

The heterodisomy detection rate is not currently known for this assay

Does not detect small intragenic or exonic number changes with the accuracy and sensitivity of the CMA-HR + SNP SCREEN array



Prenatal CMA compares specific regions of an unborn baby's DNA to that of a normal genome.



The discovery of a genetic change may provide vital information to help manage your patient's pregnancy and prepare for the baby after delivery. If the ultrasound detects an abnormality, the CMA test might help to determine the cause.

Prenatal CMA compares specific regions of an unborn baby's DNA to that of a normal genome. CMA can detect chromosomal duplications or deletions—places where there are extra or missing pieces of DNA—that are not detected by standard karyotype testing. CMA identifies nearly all of the same information as a karyotype and more, including microduplication and microdeletion syndromes, all through the convenience of one test. Microduplication and microdeletion syndromes are genetic disorders caused by a small amount of DNA being duplicated or deleted on a chromosome. A person with an additional or missing copy of DNA may experience intellectual or physical abnormalities. Nearly everyone has the same risk to have a child with one of these disorders at any age.

# Prenatal CMA Comparison Chart

	EXPANDED CMA	EXPANDED CMA + LIMITED CHROMOSOME ANALYSIS	TARGETED CMA	TARGETED CMA + LIMITED CHROMOSOME ANALYSIS
Amniotic Fluid (AF) <b>TEST CODE</b>	8670	8675	8656	8673
Chorionic Villi Sampling (CVS) <b>TEST CODE</b>	8671	8676	8657	8672
DIRECT	✓	✓	✓	✓
CULTURED	✓	✓	✓	✓

## PRENATAL EXPANDED CMA

The expanded prenatal array offers exon-by-exon coverage of over 1,700 genes as well as SNP probes across the entire genome. It is recommended for providers and patients who want the highest level of detection possible.

## PRENATAL TARGETED CMA

The targeted prenatal array contains 180,000 oligonucleotides for copy number analysis and SNP probes targeted for chromosomes 6, 7, 11, 14, 15, 20 and X for detection of uniparental disomy (UPD). Comparable to what was used in the National Institute of Child Health and Human Development (NICHD) trial, this prenatal array is ideal for providers and patients who want detection of all well-characterized deletion/duplication syndromes.

## PRENATAL EXPANDED CMA + LIMITED CHROMOSOME ANALYSIS

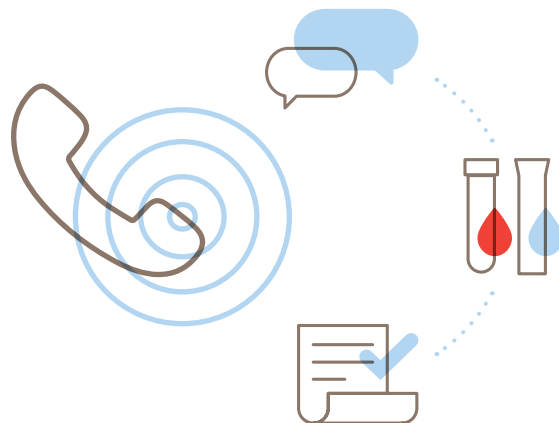
The combination of the expanded CMA and limited karyotype analysis provides a more comprehensive and cost-effective way to obtain the highest level of CMA information as well as detection of any balanced chromosomal rearrangements that could be missed by CMA.

## PRENATAL TARGETED CMA + LIMITED CHROMOSOME ANALYSIS



The combination of the targeted CMA and limited karyotype analysis provides a more comprehensive and cost-effective way to obtain targeted CMA information as well as detection of any balanced chromosomal rearrangements that could be missed by CMA.

## Prenatal Specimen Requirements

Please call 1.800.411.4363 to discuss prenatal sample requirements with a genetic counselor.



# Postnatal Specimen Requirements

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
 <b>BUCCAL SWAB</b>	Collect with ORAcollect•Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). We highly recommend the sample be collected by a healthcare professional.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
 <b>BLOOD IN EDTA</b>	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/Children) and 2-3 cc (Infant<2yrs). Blood in sodium heparin (green top) tubes, 3-5 cc (adults/children) and 1-2 cc (infants <2yrs), are highly recommended.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.