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.

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

<table>
<thead>
<tr>
<th></th>
<th>EXPANDED CMA</th>
<th>EXPANDED CMA + LIMITED CHROMOSOME ANALYSIS</th>
<th>TARGETED CMA</th>
<th>TARGETED CMA + LIMITED CHROMOSOME ANALYSIS</th>
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</thead>
<tbody>
<tr>
<td>Amniotic Fluid (AF)</td>
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<td>8675</td>
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<td>TEST CODE</td>
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<td>Chorionic Villi</td>
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<td>8657</td>
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<td>Sampling (CVS)</td>
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<td>TEST CODE</td>
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<tr>
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**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 Specimen Requirements**

Please call 1.800.411.4363 to discuss prenatal sample requirements with a genetic counselor.
# Postnatal Specimen Requirements

<table>
<thead>
<tr>
<th>TYPE</th>
<th>REQUIREMENTS</th>
<th>SHIPPING CONDITIONS</th>
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</thead>
<tbody>
<tr>
<td><strong>BUCCAL SWAB</strong></td>
<td>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.</td>
<td>Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.</td>
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<tr>
<td><strong>BLOOD IN EDTA</strong></td>
<td>Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/Children) and 2-3 cc (Infant&lt;2yrs). Blood in sodium heparin (green top) tubes, 3-5 cc (adults/children) and 1-2 cc (infants &lt;2yrs), are highly recommended.</td>
<td>Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.</td>
</tr>
</tbody>
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