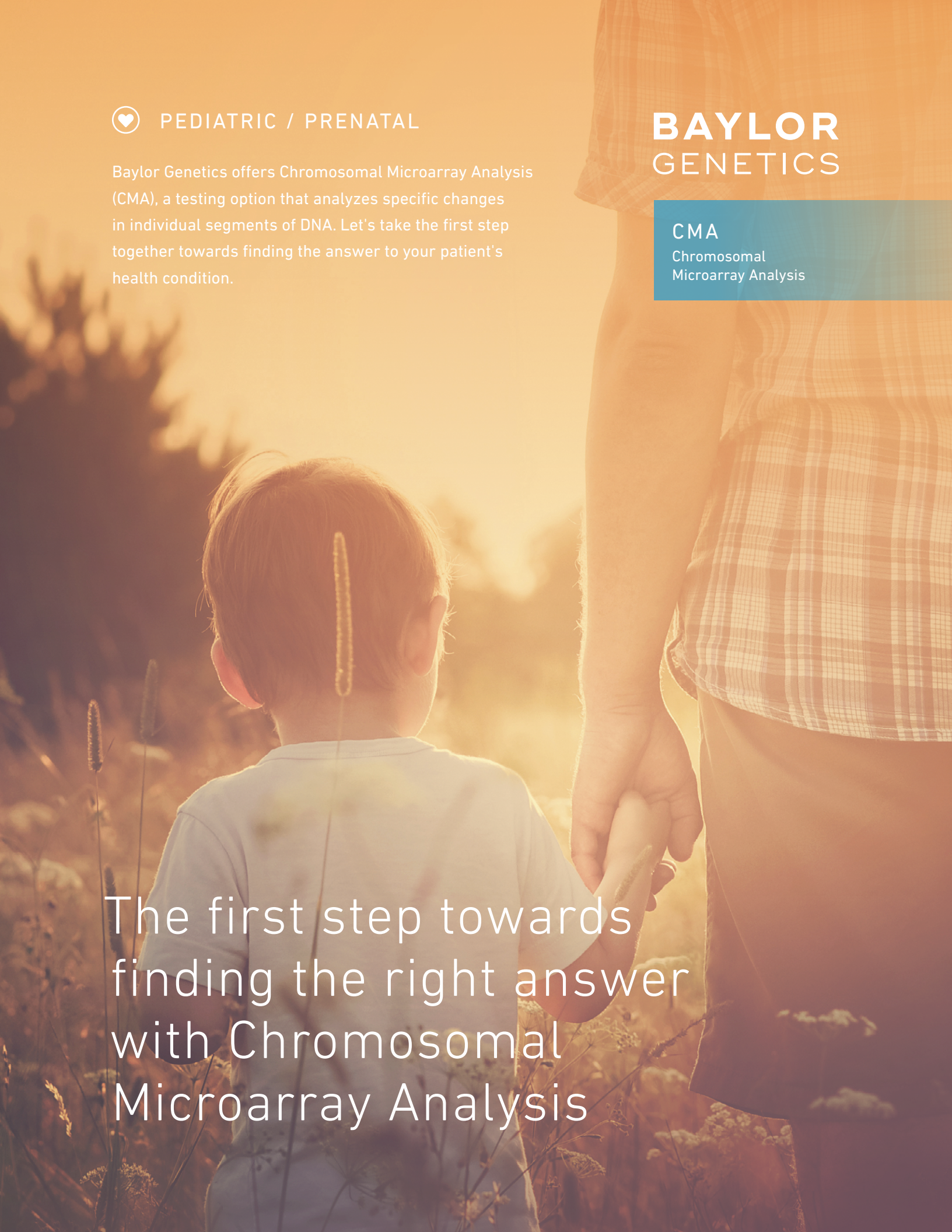


PEDIATRIC / PRENATAL

Baylor Genetics offers Chromosomal Microarray Analysis (CMA), a 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

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)

When an SNV is detected in only one allele, this test may detect potential copy number changes in an autosomal recessive disease gene

Custom In-house Design: Baylor Genetics' 400K Agilent array provides enhanced coverage for more than 5,000 genes associated with autosomal dominant, autosomal recessive X-linked disorders, and best candidate disease genes.

BENEFITS

- Maximum sensitivity for detection of gains and losses
- Exon-by-exon coverage of over 5,000 clinically relevant genes
- Whole genome backbone coverage at a 30 Kb resolution
- 60,000 SNP probes used for the detection of absence of heterozygosity (AOH) associated with uniparental disomy (UPD) or consanguinity

LIMITATIONS

- AOH less than 10 Mb in size may not be reported
- The uniparental 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

As one of the first laboratories to offer CMA testing, Baylor Genetics has performed over 100,000 CMA tests since 2004

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 examines 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.

Baylor Genetics provides two postnatal CMA options: CMA-HR+SNP Screen and CMA-HR. When we receive your patient's sample, it is analyzed against a control to determine differences in copy number variations (deletions or duplications). The location and type of change will often determine the cause of your patient's health condition.

CMA should be considered for individuals with unexplained intellectual disability, developmental delay, autism spectrum disorder, or multiple congenital anomalies.

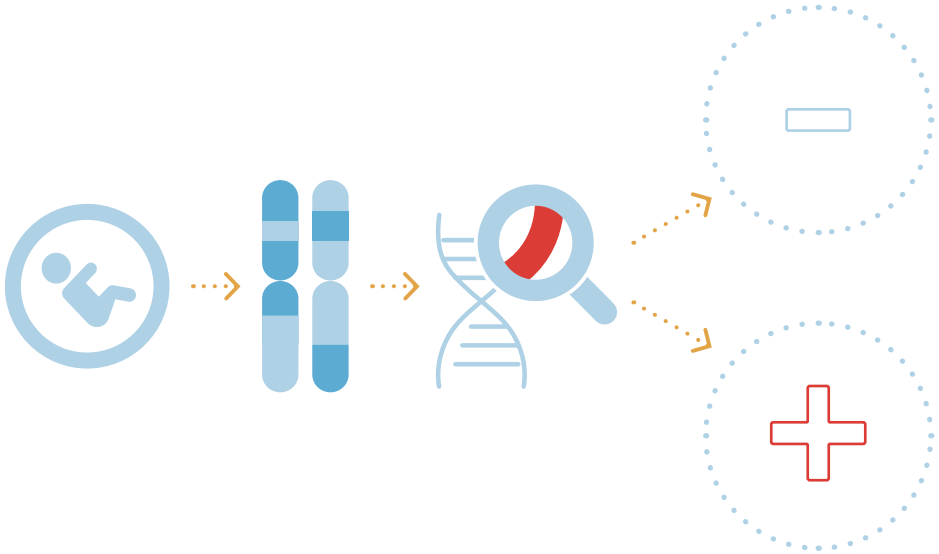
REFERENCES

Kearney HM, Thorland EC, Brown KK, Quintero-Rivera F, South ST. American College of Medical Genetics standards and guidelines for interpretation and reporting of postnatal constitutional copy number variants. Genet Med. 2011;13:680–5.

Manning M, Hudgins L. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genet Med. 2010;12:742–5.

Miller DT, Adam MP, Aradhya S, Biesecker LG, Brothman AR, Carter NP, Church DM, Crolla JA, Eichler EE, Epstein CJ, Faucett WA, Feuk L, Friedman JM, Hamosh A, Jackson L, Kaminsky EB, Kok K, Krantz ID, Kuhn RM, Lee C, Ostell JM, Rosenberg C, Scherer SW, Spinner NB, Stavropoulos DJ, Tepperberg JH, Thorland EC, Vermeesch JR, Waggoner DJ, Watson MS, Martin CL, Ledbetter DH. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010;86:749–64.

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 should be considered for fetuses with abnormal findings on imaging methodologies, NIPT, or serum screening. It should also be considered for advanced maternal age or further characterization of a previously identified chromosomal abnormality.

The American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) recommend CMA for prenatal diagnosis in cases with abnormal ultrasound findings.

REFERENCES

ACOG Practice Bulletin No. 163: Screening for Fetal Aneuploidy. American Obstetricians and Gynecologists. Obstet Gynecol. 2016; 127(5):e12337.

Hay SB, Sahoo T, Travis MK, Hovanes K, Dzidic N, Doherty C, Strecker MN. ACOG and SMFM guidelines for prenatal diagnosis: Is karyotyping really sufficient? Prenat Diagn. 2018 Feb;38(3):184-189. doi: 10.1002/pd.5212. Epub 2018 Feb 6. PMID: 29315677; PMCID: PMC5900922.

Prenatal CMA Comparison Chart

	EXPANDED CMA	EXPANDED CMA + LIMITED CHROMOSOME ANALYSIS	TARGETED CMA	TARGETED CMA + LIMITED CHROMOSOME ANALYSIS	PRODUCT OF CONCEPTION CMA
TEST CODE					
Amniotic Fluid (AF)	8670	8675	8656	8673	NA
TEST CODE					
Chorionic Villi Sampling (CVS)	8671	8676	8657	8672	NA
TEST CODE					
Tissue / Cord Blood	NA	NA	NA	NA	8639
TEST CODE					
Cord blood (for ongoing pregnancy)	8665	NA	NA	NA	NA
DIRECT	✓	✓	✓	✓	✓
Turnaround Time	7 - 10 DAYS	7 - 10 DAYS	7 - 10 DAYS	7 - 10 DAYS	21 DAYS
CULTURED	✓	✓	✓	✓	✓
Turnaround Time	21 - 28 DAYS	21 - 28 DAYS	21 - 28 DAYS	21 - 28 DAYS	21 DAYS

DISCLAIMER: CMA will be performed on direct whenever possible, chromosomes will require culturing.

PRENATAL EXPANDED CMA

The expanded prenatal array offers exon-by-exon coverage of over 1,700 clinically relevant 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, and 20 for detection of uniparental disomy (UPD). Comparable to the array 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 way to obtain the highest level of CMA information as well as detection of any balanced chromosomal rearrangements, triploidy, tetraploidy, and mosaicism diagnosed by cytogenetic analysis.

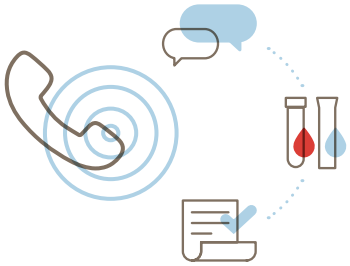
PRENATAL TARGETED CMA + LIMITED CHROMOSOME ANALYSIS

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, triploidy, tetraploidy, and mosaicism diagnosed by cytogenetic analysis.

Prenatal Specimen Requirements

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

For detailed specimen requirements, please visit: www.baylorgenetics.com/cma



Are Parental Samples Necessary?

While not mandatory, if received, we use the maternal samples to check for maternal cell contamination and parental samples to clarify variants of unknown significance.

Postnatal Specimen Requirements

Test Code: 8665, 8655

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
Blood in EDTA	Draw blood in an EDTA (purple-top) tube(s) and send 3-5cc (adults/children) and 2-3cc (infant <2 years). For clarification or follow-up of CMA results, sodium heparin (green top) tubes are highly recommended. Send 3-5cc (adults/children) and 1-2cc (infant<2 years).	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Extracted DNA	Send at least 20ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7-2.0).	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Skin Fibroblasts	Send 1-2 T25 flasks at approximately 80% confluency.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Skin Biopsy	Collect 5mm^3 millimeters of skin from a central location (e.g., buttock or upper thigh) rather than from a distal location (e.g., foot) to enhance cell viability. Place sample in a separate sterile container with RPMI media. In the absence of RPMI media, place the sample in a sterile container with either sterile saline, HBSS, or Ringer's solution. Unacceptable Conditions: Specimens placed in formalin or other fixatives.	Ship the sample at room temperature to the laboratory by overnight express. The specimen should arrive in the laboratory within 48 hours of sample date. The specimen cannot be frozen.
Buccal Swab	Collect with ORAcollect•Dx (OCD-100) self-collection kit (provided by Baylor Genetics with instructions). We highly recommend the sample to be collected by a healthcare professional.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.

Product of Conception Specimen Requirements

Test Code: 8639

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
Product of Conception (POC)	Collect 10 cubic cc of placenta from fetal side near the site of cord insertion or 5 cubic cc of identifiable fetal parts (cord, lung/kidney). Place sample in a separate sterile container with sterile: 1. Transport media provided by our laboratory, 2. Ringer's lactate, or 3. Hanks' balanced salt solution.	Ship the sample at room temperature to the laboratory by overnight express. The specimen should arrive in the laboratory within 48 hours of sample date. The specimen cannot be frozen.
Blood in EDTA	Draw cord blood or cardiac blood in an EDTA (purple-top) tube and send 2-3cc. For clarification or follow-up of CMA results, sodium heparin (green-top) tubes are highly recommended. Send 2-3 cc.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
Skin Fibroblasts	Send 1-2 T25 flasks at approximately 80% confluency.	Ship at room temperature in an insulated container by overnight courier. The specimen must arrive within 72 hours of sample date. The specimen cannot be frozen.
Cultured Tissue	Send 1-2 T25 flasks at approximately 80% confluency.	Specimen should arrive in the laboratory within 48 hours of sample date. Specimen cannot be frozen.

1.800.411.4363
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