

## Policy

EMHS Employee Health Plan (powered by Beacon Health) considers **Chromosomal Microarray Testing** medically necessary for the following indications:

- A. Prenatally, when the fetus has one or more major structural abnormalities identified on fetal ultrasound or magnetic resonance imaging (MRI)
- B. Stillborn fetuses with congenital anomalies present
- C. Members with multiple congenital anomalies not specific to a well-defined genetic syndrome
- D. Members with apparently non-syndromic developmental delay/intellectual disability
- E. Members with a clinical diagnosis of autism spectrum disorder (ASD) of sufficient severity that symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning

And

Chromosome Microarray (CMA)/Chromosome Genomic Hybridization (CGH) testing is only considered medically necessary when all of the following criteria are met:

1. The test must be ordered after completion of a three generation pedigree, or documentation that there is insufficient familial information available to complete the pedigree.
2. The signs and symptoms of the member do not suggest a classic condition for which there is a validated specific test.
3. Consultation with a BC/BE genetics counselor or a medical geneticist is to be completed before and after testing with documentation of the benefits/limitations of genetic testing and the potential to identify:
  - findings of uncertain significance,
  - misattributed paternity,
  - consanguinity, and
  - adult-onset disease;
4. Informed consent must be obtained prior to testing and kept on file;
5. Financial consult or counseling as appropriate
6. The results of the molecular/genetic test will specifically determine medication, treatment, and/or clinical management of the patient, or family member covered by EMHS Employee Health Plan (powered by Beacon Health).

### A. Limitations

CMA/CGH testing is considered not medically necessary and therefore not covered for any of the following:

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- A. Evaluation of first and second trimester pregnancy losses without congenital anomalies
- B. Members with multiple miscarriages, infertility, or who are suspected to have sex chromosome abnormalities, such as Turner or Klinefelter syndromes;
- C. Members with any symptoms, conditions, or diagnoses not included in the indications section of this policy;
- D. Members with suspected balanced chromosome rearrangements, such as balanced translocations and inversions;
- E. Members without documentation of informed consent completed prior to testing;
- F. Members who have not participated in counseling with a BC/BE genetics counselor or a medical geneticist before and after testing;
- G. Members for whom there is not a high index of suspicion of conditions due to a copy number variant;
- H. Members who present with signs and/or symptoms classic for a specific condition (a specific test should be ordered in lieu of a CMA).

### Background

The American College of Obstetricians and Gynecologists (ACOG) define chromosomal microarray analysis as a technique that identifies chromosomal abnormalities, including submicroscopic abnormalities that are too small to be detected by conventional karyotyping. More specifically, it is a high-resolution whole-genome screening that can identify major chromosomal aneuploidy as well as the location and type of specific genetic changes that are too small to be detected by conventional karyotyping. ACOG recommends CMA as a first-line genetic test in pregnancies to detect fetal abnormalities on an ultrasound screen.

Two types of chromosomal microarrays are used in clinical prenatal testing:

1. Comparative genomic hybridization (CGH)
2. Single-Nucleotide Polymorphism (SNP) arrays

### Codes:

CPT HCPCS Codes	
Code	Description
81228	Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (e.g. bacterial artificial chromosome (BAC) or oligo-based comparative genomic hybridization (CGH) microarray analysis)
81229	Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants for chromosomal abnormalities

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83870	Comparative genomic hybridization (CGH) microarray testing for developmental delay, autism spectrum disorder (ASD), and/or intellectual disability
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### Disclaimer:

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