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The Department of Vermont Health Access Clinical Criteria

Subject: Chromosomal Microarray Analysis (CMA) Genetic Testing
Last Review: September 7, 2023*
Past Revisions: June 13, 2022; November 20, 2020; June 4, 2018; and June 1, 2018

*Please note: Most current content changes will be highlighted in yellow.

Description of Service or Procedure

Chromosomal microarray (CMA) testing is utilized for detection of duplicated or deleted chromosomal segments in infants and children with characteristics of developmental delay, intellectual disability, autism spectrum disorder, and/or congenital anomalies. There are two different CMA testing platforms: array comparative genomic hybridization (aCGH) and single nucleotide polymorphism (SNP) arrays. The higher yield of CMA testing compared with other lab techniques such as G-banded karyotype analysis or FISH is primarily because of its sensitivity for identifying submicroscopic extra or missing chromosomal segments, also known as copy-number variations (CNVs).

There are many designs of CMA with different levels of resolution (from approximately 1 megabase to several kilobases). The arrays might be whole-genome arrays, designed to cover the entire genome, or targeted arrays, which target known disease-causing regions. A SNP is a variation in a single nucleotide that occurs at a specific position in the genome. SNP arrays can detect copy-number changes including amplification (increases in the number of copies of a gene in a genome) and homozygous deletion (deletion of both alleles). Sometimes these results will aid to diagnose a well described genetic condition.

CMA does not detect point mutations (sequence variants where a single nucleotide change occurs) responsible for single-gene disorders. In addition, CMA will not identify balanced translocations (when a section from one chromosome of a particular chromosomal pair swaps places with a section from a chromosome of another pair) such as inversions and may not detect low-level mosaicism, where a smaller proportion of the cells have a different genetic makeup and are affected.

Disclaimer

Coverage is limited to that outlined in Medicaid Rule or Health Care Administrative Rules that pertains to the member's aid category. Prior Authorization (PA) is only valid if the member is eligible for the applicable item or service on the date of service.



Medicaid Rule

Medicaid and Health Care Administrative Rules can be found at <u>https://humanservices.vermont.gov/rules-policies/health-care-rules/health-care-administrative-rules-hcar/adopted-rules</u>

- 7102.2 Prior Authorization Determination
- 7405 Laboratory and Radiology Services
- 4.101 Medical Necessity for Covered Services
- 4.104 Medicaid Non-Covered Services
- 4.106 Early and Periodic Screening, Diagnostic and Treatment (EPSDT) Services

Coverage Position

Chromosomal microarray analysis may be covered for members:

- When the test is prescribed by a licensed medical provider, enrolled in the Vermont Medicaid program, operating within their scope of practice as described on the Vermont Office of Professional Regulation's website*, Statute, or rule who is knowledgeable regarding genetic testing and who provides medical care to the member AND
- When the clinical criteria below are met.

* Vermont's Office of Professional Regulation's website: https://sos.vermont.gov/opr/

Coverage Criteria

Chromosomal microarray analysis may be considered medically necessary as first-line testing in the initial postnatal evaluation of members age 17 years or younger with clinical documentation of a developmental delay, autism spectrum disorder (ASD), or intellectual disability or when multiple congenital anomalies are present since birth, when the following conditions are met:

- 1. If warranted by the clinical presentation, biochemical testing for metabolic diseases has been performed and is negative **AND**
- 2. Targeted genetic testing if warranted by the clinical presentation and family history is negative **AND**
- 3. If no clinical features or family history suggestive of Fragile X Syndrome, negative FMR is not required AND
- 4. The individual's clinical presentation is not specific to a well delineated genetic syndrome

In addition, chromosomal microarray analysis is not considered medically necessary when:

- 1. Biologic parents have known chromosomal rearrangements* **OR**
- 2. There is a maternal history of recurrent miscarriages**

*Karyotyping is the test of choice when the individual has a family history of balanced chromosomal rearrangement

**Balanced translocation is a common reason for recurrent miscarriages and cannot be detected on cytogenetic microarrays. Karyotyping is the test of choice.

Considerations: Providers requesting this test should provide pre- and post-test genetic counseling for the member and family, if applicable.

Early and Periodic Screening, Diagnostic and Treatment (EPSDT): Vermont Medicaid will provide comprehensive services and furnish all Medicaid coverable, appropriate, and medically necessary services needed to correct and ameliorate health conditions for Medicaid members under age 21.

Please note, Vermont Medicaid Clinical Criteria is reviewed based on available literature, evidence-based guidelines/standards, Medicaid rule and policy, and Medicare coverage determinations that may be appropriate to incorporate when applicable.

Clinical criteria for repeat service or procedure

This test is covered once per lifetime.

Type of service or procedure covered

Chromosomal microarray analysis testing.

Type of service or procedure not covered (this list may not be all inclusive)

Chromosomal microarray analysis testing is **investigational** for all other uses, including for the evaluation of all other conditions of delayed development, including but not limited to idiopathic growth or language delay.

Coding guidelines

Please see the Medicaid Portal at <u>http://vtmedicaid.com/#/feeSchedule</u> for fee schedules, code coverage, and applicable requirements.

References

- Centers for Medicare and Medicaid Services. (n.d.). Early and periodic screening, diagnostic, and treatment. Medicaid.gov. <u>https://www.medicaid.gov/medicaid/benefits/epsdt/index.html</u>
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