



Chromosomal Microarray Analysis LAB-001

Iowa Medicaid Program	Prior Authorization	Effective Date	01/01/2021
Revision Number	5	Last Reviewed	01/17/2025
Reviewed By	Medicaid Medical Director	Next Review	01/16/2026
Approved By	Medicaid Clinical Advisory Committee	Approved Date	12/23/2020

Descriptive Narrative

Chromosomal microarray analysis (CMA) is a high-resolution whole-genome screening test, which detects small genetic alterations, including submicroscopic abnormalities that are too small to be identified by conventional karyotyping or FISH (fluorescence in situ hybridization) analysis. While conventional karyotypes primarily detect genetic abnormalities resulting from large changes in the structure or number of chromosomes (translocations, aneuploidy, large deletions or duplications), CMA identifies genomic copy number variations, which are deletions and/or duplications that are smaller than the resolution of karyotyping. CMA does not detect balanced chromosome rearrangements in which there is no gain or loss of DNA (balanced inversions or balanced translocations). CMA is also known as molecular karyotyping, microarray-based genomic copy-number analysis, or array-based comparative genomic hybridization.

CMA is a useful diagnostic tool for infants and children with unexplained developmental delay (DD), autism spectrum disorder (ASD) or intellectual disability (ID). The use of CMA leads to a genetic diagnosis in 15 to 20 percent of members with unexplained ID. If CMA fails to find a cause of ID, whole exome sequencing may be used to identify the causative mutation(s).

Definitions

Intellectual Disability is a neurodevelopmental disorder characterized by deficits in intellectual and adaptive skills, affecting at least one of three adaptive domains (conceptual, social, and practical) with varying severity. The older term "mental retardation" is no longer used in clinical practice. Standardized intelligence quotient testing is no longer used to classify ID severity. The prevalence of ID in the general population is approximately 1 percent. ID is mild in approximately 85 percent of affected individuals.

Global Developmental Delay (GDD) is the preferred term to describe intellectual and adaptive impairment in infants and children younger than 5 years of age who fail to meet expected developmental milestones in multiple areas of functioning. Not all children with GDD meet criteria for ID as they grow older. The prevalence of GDD is estimated to be 1 to 3 percent. GDD does not necessarily predict later ID, although there is a strong correlation.

Well-Delineated Genetic Syndrome is a collection of recognizable traits or abnormalities that tend to occur together and are associated with a specific disease. Distinguishing characteristics, such as specific facial features or other physical traits, lab tests, or family history can be used to identify a genetic syndrome.

Karyotype is a laboratory technique that produces an image of a member’s chromosomes. The karyotype is used to look for abnormal numbers or structures of chromosomes.

Criteria

Chromosomal microarray analysis is considered medically necessary when **ALL** the following are met:

1. Member is 17 years of age or younger; **AND**
2. Genetic counseling has been provided to the member and their family by a qualified health professional; **AND**
3. The member has **ONE** of the following:
 - a. Significant dysmorphic features or congenital anomalies not specific to a well delineated genetic syndrome; **OR**
 - b. Diagnosis of ASD; **OR**
 - c. Presentation of non-syndromic DD or ID; **AND**
4. The test results have the potential to impact clinical management.

Coding

The following list of codes is provided for reference purposes only and may not be all inclusive. The inclusion of a code does not imply any right to reimbursement or guarantee claim payment, nor does the exclusion of a code imply that its association to the HCPCS/CPT code is inappropriate.

CPT	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).
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Compliance

1. Should conflict exist between this policy and applicable statute, the applicable statute shall supersede.
2. Federal and State law, as well as contract language, including definitions and specific contract provisions or exclusions, take precedence over medical policy and must be considered first in determining eligibility for coverage.
3. Medical technology is constantly evolving, and Iowa Medicaid reserves the right to review and update medical policy on an annual and as-needed basis.

Medical necessity guidelines have been developed for determining coverage for member benefits and are published to provide a better understanding of the basis upon which coverage decisions are made. They include concise clinical coverage criteria based on current literature review, consultation with practicing physicians in the service area who are medical experts in the particular field, FDA and other government agency policies, and standards adopted by national accreditation organizations. Criteria are revised and updated annually, or more frequently if new evidence becomes available that suggests needed revisions.

References

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Shafqat I. Martinez-Agosto JA. The Role of Genetic Testing Among Autistic Individuals. American Academy of Pediatrics. Published in Pediatrics (2022) 149 (Supplement 4): e2020049437L.

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Development of utilization management criteria may also involve research into other state Medicaid programs, other payer policies, consultation with experts and review by the Medicaid Clinical Advisory Committee (CAC). These sources may not be referenced individually unless they are specifically published and are otherwise applicable to the criteria at issue.

Criteria Change History			
Change Date	Changed By	Description of Change	Version
[mm/dd/yyyy]			[#]
Signature			
Change Date	Changed By	Description of Change	Version
01/17/2025		Annual Review. References updated.	5
Signature			
William (Bill) Jagiello, DO 			
Change Date	Changed By	Description of Change	Version
01/19/2024		Annual Review.	4
Signature			
William (Bill) Jagiello, DO 			
Change Date	Changed By	Description of Change	Version
01/20/2023		Annual Review.	3
Signature			
William (Bill) Jagiello, DO 			
Change Date	Changed By	Description of Change	Version
01/21/2022		Annual Review.	2
Signature			
William (Bill) Jagiello, DO 			
Change Date	Changed By	Description of Change	Version
01/15/2021	CAC	Criteria implementation.	1
Signature			
William (Bill) Jagiello, DO 			

CAC = Medicaid Clinical Advisory Committee