CLINICAL MEDICAL POLICY	
Policy Name:	Chromosomal Microarray Analysis: Comparative Genomic Hybridization (CGH) and Single Nucleotide Polymorphism (SNP)
Policy Number:	MP-036-MC-PA
Responsible Department(s):	Medical Management
Provider Notice/Issue Date:	10/26/2026; 07/01/2025; 07/01/2024; 07/01/2023; 08/01/2022; 07/16/2021; 07/20/2020; 08/12/2019; 09/01/2018
Effective Date:	12/26/2026;08/01/2025; 08/01/2024; 08/01/2023; 09/01/2022; 08/16/2021; 08/17/2020; 08/12/2019; 09/01/2018; 12/01/2017
Next Annual Review:	12/2027
Revision Date:	06/25/2026;06/18/2025; 06/19/2024; 06/21/2023; 06/15/2022; 06/16/2021; 06/17/2020; 06/11/2019; 06/20/2018
Products:	Pennsylvania Medicare Assured
Application:	All participating and nonparticipating practitioners and facilities unless contractually precluded
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Policy History

Date	Activity
12/26/2026	Provider Effective Date
07/15/2026	QIUM Committee
06/25/2026	Annual Review: No clinical changes.
08/01/2025	Provider Effective date
06/18/2025	QI/UM Committee review
06/18/2025	Annual Review: No changes to clinical criteria. Updated 'Reference Sources' section.
08/01/2024	Provider Effective date
06/19/2024	QI/UM Committee review
06/19/2024	Annual Review: No changes to clinical criteria. Updated 'Summary of Literature' and 'Reference Sources'
08/01/2023	Provider Effective date
06/21/2023	QI/UM Committee review

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06/21/2023	Annual Review: No changes to clinical criteria. Updated 'Summary of Literature' and 'Reference Sources' sections.
09/01/2022	Provider Effective date
06/15/2022	QI/UM Committee review
06/15/2022	Annual Review: Removed the statement "*CGH is considered experimental/investigational when used to determine a single congenital anomaly (i.e., developmental delay/intellectual disability, autism spectrum disorder, without other diagnoses)"* from the Procedures section. Per AMA guidance, updated the code descriptions for CPT codes 81228 & 81229; updated code description for F78.A9. Removed the following unspecified coding: F79, F80.9, F81.9, F84.9, F89, P02.9, Q01.9, Q03.9, Q04.9, Q05.9, Q06.9, Q07.9, Q89.9, Q92.9, Q93.9, & Q99.9. Added the code P02.8. Updated the Summary of Literature and Reference Sources sections.
08/16/2021	Provider effective date
06/16/2021	QI/UM Committee review
09/15/2017	Initial policy developed

Disclaimer

Highmark Wholecare℠ medical policy is intended to serve only as a general reference resource regarding coverage for the services described. This policy does not constitute medical advice and is not intended to govern or otherwise influence medical decisions.

Policy Statement

Highmark Wholecare does not provide coverage under the medical benefits of the Company's Medicare products for chromosomal microarray analysis which includes Comparative Genomic Hybridization (CGH) and Single Nucleotide Polymorphism (SNP) laboratory procedures.

This policy is designed to address medical necessity guidelines that are appropriate for the majority of individuals with a particular disease, illness or condition. Each person's unique clinical circumstances warrant individual consideration, based upon review of applicable medical records.

Definitions

Autism Spectrum Disorder – Per the PA Act 62, autism is defined as any of the pervasive developmental disorders defined by the most recent edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM), or its successor, including autistic disorder, Asperger's disorder and pervasive developmental disorder not otherwise specified.

Comparative Genomic Hybridization Microarray testing – a laboratory test performed to detect unbalanced genomic copy number of variations such as microdeletions and/or microduplications at a higher resolution level than conventional genetic evaluation (e.g., karyotype analysis or fluorescence in situ hybridization [FISH]). The test can be performed on blood, body fluid or tissue specimen

Developmental Delay – A term used to describe children younger than five years of age who present with delays in the attainment of development milestones at the expected age.

Intellectual Disability – An intellectual disability (previously referred to as mental retardation) may be used to describe persons five years of age and older (when standardized measures of intelligence become reliable and valid) who exhibit deficits in intelligence (IQ), adaptive behavior, and systems of support (American Association on Mental Retardation, 2002).

Karyotype – A term that defines the number of chromosomes in a given cell. In normal human beings there are 46 chromosomes (23 pairs). The first 22 pairs are called the autosomes and are numbered from one to twenty-two according to length, longest to shortest. The 23rd pair is the sex chromosomes (X or Y).

Microdeletions - The loss of a minute piece of chromosome and microduplications are the gain of a minute piece of a chromosome. To detect the microdeletions or microduplications high resolution techniques such as DNA analysis is required.

Next-Generation Sequencing – A method of DNA sequencing genome technology at high speed. Also known as second generation sequencing or massively parallel sequencing.

Syndrome – A pattern of recognizable multiple malformations. The diagnosis of syndromes are often relatively straightforward and common enough to be clinically recognized without specialized testing. Syndrome examples would include Down syndrome and achondroplasia. In the very young or in the case of syndromes with variable presentation, confident identification may be difficult without additional testing.

Procedures

1. Chromosomal Microarray Analysis (CMA) testing is considered medically necessary when ALL of the following criteria is met :
    A. The child must be under the age of 21; AND
    B. The child's parents have been engaged in face-to-face genetic counseling with a healthcare professional; AND
    C. Targeted genetic testing (e.g., gene analysis for Fragile X) and biochemical testing for metabolic diseases are negative; AND
    D. Documentation that the genetic testing results will guide clinical decisions that would not otherwise be made in the absence of the testing.
    E. The child must exhibit ANY of the following conditions:
        1) Multiple congenital anomalies not specific to a well-delineated genetic syndrome; multiple congenital anomalies are defined as:
            - Two (2) or more major anomalies affecting different organ systems; OR
            - One (1) major and two (2) or more minor anomalies affecting different organ systems (**Note**: Major structural anomalies are generally serious enough as to require medical treatment, such as surgery, and are not minor developmental variations that may or may not suggest an underlying disorder; OR

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2) Apparent non-syndromic developmental delay/intellectual disability; OR
3) Autism Spectrum Disorder (e.g., Asperger's, autistic disorder, pervasive developmental disorder); AND

2. CMA of amniotic fluid, placenta, or products of conception (POC) for evaluation of pregnancy loss is considered medically necessary in EITHER ONE of the following conditions:
   A. In cases of pregnancy loss at 20 weeks of gestation or earlier, when there is a maternal history of recurrent miscarriage (history of two [2] or more failed pregnancies); OR
   B. In all cases of pregnancy loss after 20 weeks of gestation.

   Note: This policy does not address the use of CMA for preimplantation genetic diagnosis or preimplantation genetic screening.

3. Genetic Counseling

   Pre- and post-test genetic counseling is required to be performed by an independent genetic provider (not employed by a genetic testing lab) prior to genetic testing for mutations. This service is necessary in order to inform the patient being tested about the benefits and limitations of specific genetic tests. Genetic testing for mutations requires documentation of medical necessity from at least one of the following providers who has previously evaluated the patient, and intends to see the patient after genetic testing has been performed:
   - Board Eligible or Board Certified Genetic Counselor
   - Advanced Genetics Nurse
   - Genetic Clinical Nurse
   - Advanced Practice Nurse in Genetics
   - Board Eligible or Board Certified Clinical Geneticist
   - A healthcare professional of appropriate expertise or other obstetrical provider specializing in the care for the indication(s) for genetic testing

4. When the laboratory services are considered not medically necessary
   - CMA is considered not medically necessary when the diagnosis is readily apparent and can be confirmed on clinical evaluation alone.
   - CMA is unproven and not medically necessary for all other patient populations and conditions not listed in this policy.
   - Panel testing using next-generation gene sequencing is considered experimental/investigational in all cases of suspected genetic abnormality in children with developmental delay/intellectual disability, autism spectrum disorder, or congenital anomalies.
   - CMA of fetal tissue for the evaluation of pregnancy loss when the patient selection criteria is not met are considered not medically necessary.
   - Any requests for CMA approval that does not meet the guidelines listed above will require a review by a Medical Director on a case-by-case basis.

5. Post-payment Audit Statement

   The medical record must include documentation that reflects the medical necessity criteria and is subject to audit by Highmark Wholecare at any time pursuant to the terms of your provider agreement.

6. Place of Service
   The proper place of service for chromosomal microarray laboratory testing is outpatient.

Governing Bodies Approval

Genetic testing are laboratory developed tests that do not require premarket approval by the FDA. These types of tests are regulated under the Clinical Laboratory Improvement Amendments (CLIA) Act of 1998. The regulations of the CLIA Amendments do not include validation of specific test but rather there is procedural compliance.

The use of the chromosomal microarray testing outside of listed FDA guidelines will require approval from a Medical Director on a case-by-case basis.

CMS
The Center for Medicare and Medicaid Services has not published any National Coverage Determination (NCD) or Local Coverage Determination (LCD) articles on this topic.

Summary of Literature

According to the World Health Organization, a congenital disorder can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital malformations, these conditions develop prenatally and may be identified before or at birth, or later in life. An estimated 6% of babies worldwide are born with a congenital anomaly, resulting in hundreds of thousands of associated deaths (WHO, 2024). The anomaly can be classified as a minor anomaly in which the defect is an unusual anatomic feature that is of no serious medical or cosmetic consequence. Examples of a minor anomaly can include protruding ears, ptosis, anteverted nostrils, hypotelorism, minor hypospadias, partial syndactyly between 2-3 toes, and plagiocephaly. A major anomaly is a defect that has serious medical and cosmetic consequences. Examples of a major anomaly can include cleft lip and palate, absence or limb deficiencies, hydrocephaly, hypoplasia or coarctation of the aorta, micrognathia severe, pectus excavatum, spina bifida, and Tetralogy of Fallot.

Chromosomal microarray analysis (CMA) is a method of measuring gains and losses of DNA throughout the human genome. It can identify chromosomal aneuploidy and other large changes in the structure of chromosomes as well as submicroscopic abnormalities that are too small to be detected by traditional modalities (ACOG, 2020). Postnatal detection of significant CNVs provides findings that would have been missed using conventional karyotyping alone, such as developmental delays and intellectual disability. An additional 12.2% – 19% pathogenic anomalies may be detected with the addition of microarray. CNVs can be performed on tissue that is no longer viable. If DNA is present and of sufficient quality, test can be run on stillbirth specimens or products of conception (ObG Project).

CMA can identify genomic abnormalities that are associated with a wide range of developmental disabilities, including cognitive impairment, behavioral abnormalities, and congenital abnormalities. CMA can detect copy number variants (CNVs), and the frequency of disease-causing CNVs is highest (20%-25%) in children with moderate to severe intellectual disability accompanied by malformations or dysmorphic features. Disease-causing CNVs have been identified in 5% to 10% of cases of autism, being more frequent

in severe phenotypes. CMA includes both comparative genomic hybridization (CGH) and single nucleotide polymorphism (SNP) arrays. CGH microarray testing, also known as array comparative genomic hybridization (CGH), is a technology that can be used for the detection of genomic CNVs.

CNVs are alterations that include deletion and/or duplication of one or more sections of DNA. This method allows the detection of chromosome imbalances that can provide more information than detected by conventional chromosome analysis [e.g., standard karyotype or fluorescence in situ hybridization (FISH)]. The array CGH approach compares patient DNA extracted from skin, blood, or fetal cells to a control or reference DNA from a normal individual. These are labelled separately with different colored fluorescent dyes and then mixed together and allowed to combine or hybridize to an array containing known DNA sequences called probes. The amount of hybridization is measured by the amount and color of light emitted from each spot.

Computer analysis of the fluorescent signals is used to read and interpret the findings. Areas of unequal hybridization, mostly large deletions and duplications, signify a DNA alteration. CNVs may be benign, with no effect on clinical phenotype, or may be pathogenic and result in a variety of phenotypic abnormalities (Kearney et al., 2011). If an unknown CNV is detected, a genomic database is used to determine if the abnormality has been previously reported and if it has been associated with a benign or proposed pathogenic condition. The disadvantages of array CGH testing include the detection of a large number of variants of unknown clinical significance, potential false positives results that will require further testing, and the inability to detect certain anomalies such as those with balanced rearrangements where there is no net gain or loss of the chromosomes (Fruhman and Van den Veyver 2010; Bui 2011).

The American Academy of Neurology and the Practice Committee of the Child Neurology Society have determined that CMA testing has the highest diagnostic yield in children with DD/ID (Michelson et al., 2011). In addition, the society determined that CMA should be considered the first-line test in children with DD/ID. The authors note that the assistance of a medical geneticist is necessary.

The American College of Medical Genetics and Genomics (ACMG) published recommendations on the array-based technologies and the clinical utilization for detecting chromosomal abnormalities (Manning, Hudgens, 2010).

1. CMA testing for CNV is recommended as a first-line test in the initial postnatal evaluation of individuals with the following:
    A. Multiple anomalies not specific to a well-delineated genetic syndrome.
    B. Apparently non-syndromic DD/ID.
    C. Autism spectrum disorders.
2. Further determination of the use of CMA testing for the evaluation of the child with growth retardation, speech delay, and other less well-studied indications is recommended, particularly by prospective studies and after-market analysis.
3. Appropriate follow-up is recommended in cases of chromosome imbalance identified by CMA, to include cytogenetic/FISH studies of the patient, parental evaluation, and clinical genetic evaluation and counseling.

The ACMG guideline states that ordering providers should be aware of cytogenomic aberrations not detectable by CMA, including those relevant to various microarray platforms (e.g., single-nucleotide polymorphism [SNP] versus oligonucleotide).

Next-generation sequencing (NGS) panel testing allows for simultaneous analysis of a large number of genes and the testing has been proposed as a way to identify single-gene causes of syndromes that have autism as a significant clinical feature in patients with normal CMA testing. To date, there are no peer-reviewed full length publications on the commercially available NGS panels related to the clinical and analytical validity or the clinical utility of the diagnostic test.

Most pregnancy losses happen in early pregnancy. Pregnancy loss occurring before the 20th week of gestation is referred to as spontaneous abortion, early pregnancy loss, or miscarriage. Fetal loss occurring after 20 weeks gestation is referred to as stillbirth or intrauterine fetal death (IUFD). Early pregnancy loss is defined as a nonviable intrauterine pregnancy with either an empty gestational sac or an embryo/fetus without cardiac activity at <13 weeks gestation (ACOG 2015). It is estimated that early pregnancy loss occurs commonly and affects 10% to 15% of recognized pregnancies under 20 weeks. The overall risk of miscarriage in the next pregnancy remains at 15% after one miscarriage, rises to 17% from 13% after two consecutive miscarriages, and climbs to 25% to 46% after three or more miscarriages (UpToDate, 2017). There is no preventative therapy for women with threatened early pregnancy loss and a work-up on the cause of the loss, is not recommended until after the second consecutive loss.

Genetic evaluation of the products of conception has traditionally been performed using karyotyping of metaphase cells after cells are culture in tissue. Using this method, only visible rearrangements are detected. There are risks for maternal cell contamination which can impact karyotyping. An alternative genetic testing method has been utilized, chromosomal microarray testing.

The American College of Obstetrics and Gynecology (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) recommend use of CMA when genetic analysis is desired because of fetal congenital anomalies or intrauterine fetal death or stillbirth (UpToDate, 2021). CMA is useful in the valuation of stillbirth because both chromosomal abnormalities and culture failure are common. Paula and colleagues (2018) reported the results of systematic review of twenty-three studies in which CMA and karyotyping were performed concurrently. The analysis revealed that CMA showed a significant increase in test success rate and incremental diagnostic yield in early pregnancy loss. CMA revealed informative results in 95% of the 5,507 pregnancy losses reviewed while karyotyping results were 68%.

SNP microarrays are applications of microarray technology that also provide genome-wide copy number analysis. In addition to copy number changes, SNP arrays are able to detect so-called “copy number neutral” abnormalities such as segmental uniparental disomy and areas of long contiguous stretches of homozygosity that can give rise to disease, congenital anomalies, or cognitive impairment. SNP arrays are increasingly being used in the assessment of cognitive impairment or DD, with or without associated anomalies and are likely to be used in the diagnosis of these conditions (Manning, Hudgens, 2010).

A report published by the American Academy of Pediatrics (AAP) tested the hypothesis that chromosomal microarray analysis frequently diagnoses conditions that require specific medical follow-up and that referring healthcare professional’s respond appropriately to abnormal test results. A total of 46,298 postnatal patients were tested by chromosomal microarray analysis for a variety of indications, most commonly intellectual disability/developmental delay, congenital anomalies, dysmorphic features, and neurobehavioral problems. The frequency of detection of abnormalities associated with actionable clinical features was tallied, and the rate of healthcare professional’s response to a subset of abnormal tests results was monitored. The testing found that the disorders diagnosed by chromosomal microarray analysis frequently have clinical features that need medical attention, and healthcare professional’s

respond to the diagnoses with specific clinical actions, thus arguing that microarray testing provides clinical utility for a significant number of patients tested.

CMA limitations include the inability to detect rare balanced trans location where the break point might disrupt the coding region of a gene and inactivate it. CMA cannot determine the precise mechanism of a gain or loss, which may affect the recurrence risk relevant to future counselling for the same CNV disorder in other family members. In addition, most microarrays used for routine clinical CMA cannot detect single gene–level deletions or duplications unless the gene was specifically targeted in the array design (Audio). However, continued development of whole exome– and genome sequence–based analyses and new algorithms to identify copy-number variants from this data will likely overcome these limitations. It is anticipated that clinical genetic testing will become a sequencing-based test that can detect copy-number and sequence variants in a single assay. CMA testing can also detect abnormalities not previously described and of unclear clinical meaning (Martin, Ledbetter 2017).

CMA offers a powerful approach for detecting pathogenic copy-number changes in the genome. CMA should be offered when evaluating individuals diagnosed with otherwise unexplained developmental delay, intellectual disability, ASD, or congenital anomalies. CMA can be critical in these patient populations for providing etiologic diagnoses and to aid in directing medical management (Martin, Ledbetter 2017).

Coding Requirements

Procedure Codes

CPT Code	Description
81228	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number variants, comparative genomic hybridization [CGH] microarray analysis
81229	Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants, comparative genomic hybridization (CGH) microarray analysis

Diagnosis Codes

ICD-10 Code	Description
F70	Mild intellectual disabilities
F71	Moderate intellectual disabilities
F72	Severe intellectual disabilities
F73	Profound intellectual disabilities
F78.A9	Other genetic related intellectual disability
F80.0	Phonological disorder
F80.1	Expressive language disorder
F80.2	Mixed receptive-expressive language disorder
F80.4	Speech and language development delay due to hearing loss

F80.81	Childhood onset fluency disorder
F80.82	Social pragmatic communication disorder
F80.89	Other developmental disorders of speech and language
F81.0	Specific reading disorder
F81.2	Mathematics disorder
F81.81	Disorder of written expression
F81.89	Other developmental disorders of scholastic skills
F82	Specific developmental disorder of motor function
F84.0	Autistic disorder
F84.3	Other childhood disintegrative disorder
F84.5	Asperger's syndrome
F84.8	Other pervasive developmental disorders
F88	Other disorders of psychological development
F90.8	Attention-deficit hyperactivity disorder, other type
H93.25	Central auditory processing disorder
P02.8	Newborn affected by other abnormalities of membranes
Q00.0	Anencephaly
Q00.1	Cranioarchischisis
Q00.2	Iniencephaly
Q01.0	Frontal encephalocele
Q01.1	Nasofrontal encephalocele
Q01.2	Occipital encephalocele
Q01.8	Encephalocele of other sites
Q02	Microcephaly
Q03.0	Malformations of aqueduct of Sylvius
Q03.1	Atresia of foramina of Magendie and Luschka
Q03.8	Other congenital hydrocephalus
Q04.0	Congenital malformations of corpus callosum
Q04.1	Arhinencephaly
Q04.2	Holoprosencephaly
Q04.3	Other reduction deformities of brain
Q04.4	Septo-optic dysplasia of brain
Q04.5	Megalencephaly
Q04.6	Congenital cerebral cysts
Q04.8	Other specified congenital malformations of the brain
Q05.0	Cervical spina bifida with hydrocephalus
Q05.1	Thoracic spina bifida with hydrocephalus
Q05.2	Lumbar spina bifida with hydrocephalus
Q05.3	Sacral spina bifida with hydrocephalus
Q05.4	Unspecified spina bifida with hydrocephalus
Q05.5	Cervical spina bifida without hydrocephalus
Q05.6	Thoracic spina bifida without hydrocephalus
Q05.7	Lumbar spina bifida without hydrocephalus
Q05.8	Sacral spina bifida without hydrocephalus
Q06.0	Amyelia
Q06.1	Hypoplasia and dysplasia of spinal cord

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Q06.2	Diastematomyelia
Q06.3	Other congenital cauda equine malformations
Q06.4	Hydromyelia
Q06.8	Other specified congenital malformations of spinal cord
Q07.00	Arnold-Chiari syndrome without spina bifida or hydrocephalus
Q07.01	Arnold-Chiari syndrome with spina bifida
Q07.02	Arnold-Chiari syndrome with hydrocephalus
Q07.03	Arnold-Chiari syndrome with spina bifida and hydrocephalus
Q07.8	Other specified congenital malformation of nervous system
Q89.7	Multiple congenital malformations, not elsewhere classified
Q89.8	Other specified congenital malformations
Q90.0	Trisomy 21, nonmosaicism (meiotic nondisjunction)
Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2	Trisomy 21, translocation
Q90.9	Down syndrome, unspecified
Q91.0	Trisomy 18, nonmosaicism (meiotic nondisjunction)
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.2	Trisomy 18, translocation
Q91.3	Trisomy 18, unspecified
Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction)
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation
Q91.7	Trisomy 13, unspecified
Q92.0	Whole chromosome trisomy, nonmosaicism (meiotic nondisjunction)
Q92.1	Whole chromosome trisomy, mosaicism (mitotic nondisjunction)
Q92.2	Partial trisomy
Q92.5	Duplications with other complex rearrangements
Q92.61	Marker chromosomes in normal individual
Q92.62	Marker chromosomes in abnormal individual
Q92.7	Triploidy and polyploidy
Q92.8	Other specified trisomies and partial trisomies of autosomes
Q93.0	Whole chromosome monosomy, nonmosaicism (meiotic nondisjunction)
Q93.1	Whole chromosome monosomy, mosaicism (mitotic nondisjunction)
Q93.2	Chromosome replaced with ring, dicentric or isochromosome
Q93.3	Deletion of short arm of chromosome 4
Q93.4	Deletion of short arm of chromosome 5
Q93.51	Angelman syndrome
Q93.59	Other deletions of part of a chromosome
Q93.7	Deletions with other complex rearrangements
Q93.81	Velo-cardio-facial syndrome
Q93.82	Williams syndrome
Q93.88	Other microdeletions
Q93.89	Other deletions from the autosomes
Q95.2	Balanced autosomal rearrangement in abnormal individual
Q95.3	Balanced sex/autosomal rearrangement in abnormal individual
Q99.8	Other specified chromosome abnormalities

R48.0	Dyslexia and alexia
R62.0	Delayed milestone in childhood
R62.50	Unspecified lack of expected normal physiological development in childhood
R62.51	Failure to thrive (child)
R62.59	Other lack of expected normal physiological development in childhood
R89.8	Other abnormal findings in specimens from other organs, systems and tissues

Reimbursement

Participating facilities will be reimbursed per their Highmark Wholecare℠ contract.

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