CLINICAL MEDICAL POLICY	
Policy Name:	Chromosomal Microarray Analysis (CMA): Comparative Genomic Hybridization (CGH) and Single Nucleotide Polymorphism (SNP)
Policy Number:	MP-012-MD-PA
Responsible Department(s):	Medical Management
Provider Notice/Issue Date:	03/01/2024; 02/01/2023; 04/01/2022; 03/19/2021; 02/17/2020; 05/06/2019; 04/15/2018; 10/01/2016
Effective Date:	04/01/2024; 03/01/2023; 05/01/2022; 04/19/2021; 03/16/2020; 05/06/2019; 04/15/2018; 10/01/2016
Next Annual Review:	11/2024
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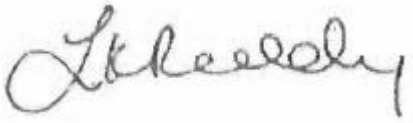
Products:	Highmark Wholecare℠ Medicaid
Application:	All participating hospitals and providers
Page Number(s):	1 of 14
Senior Medical Director/Medical Director Of Medical Policy:*	
Related Departments:*	Clinical Operations (UM); Claims; Systems Configuration; Appeals & Grievances
Copied Departments:*	Provider Relations; Clinical Pharmacy; Regulatory; Compliance; Legal; Fraud, Waste & Abuse

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Policy History

Date	Activity
04/01/2024	Provider Effective date
02/20/2024	PARP Approval
11/15/2023	QI/UM Committee review
11/15/2023	Annual Review: No changes to clinical criteria. Reformatted 'Procedures' section. Updated 'Summary of Literature' and 'Reference Sources' sections.
03/01/2023	Provider Effective date
01/10/2023	PARP Approval

11/16/2022	QI/UM Committee review
11/16/2022	Annual Review: No changes to clinical criteria. Updated 'Summary of Literature' and 'Reference Sources' sections. Updated code descriptions for CPT codes 81228 & 81229. Updated code descriptions for the following ICD-10 codes: P02.9, Q01.1, Q01.2, Q01.8, Q01.9, Q03.9. Removed deleted ICD-10 code F78, replaced with code F78.A9.
05/01/2022	Provider Effective date
03/09/2022	PARP Approval
11/17/2021	QI/UM Committee review
11/17/2021	Annual Review: Removed the phrase "*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 #4 under Procedures section. Added Chromosomal Microarray Testing definition. Added TAG determination information. Updated Summary of Literature and Reference sections.
04/19/2021	Provider effective date

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℠ provides coverage under the medical benefits of the Company's Medicaid products for medically necessary 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.

(Current applicable PA HealthChoices Agreement Section V. Program Requirements, B. Prior Authorization of Services, 1. General Prior Authorization Requirements.)

Definitions

Prior Authorization Review Panel (PARP) – A panel of representatives from within the PA Department of Human Services who have been assigned organizational responsibility for the review, approval and denial of all PH-MCO Prior Authorization policies and procedures.

Chromosomal Microarray Analysis (CMA) - a genome-wide assay that examines the chromosomes for tiny, sub-microscopic deletions or duplications of DNA sequences, known as copy-number variants.

Autism Spectrum Disorder (ASD) – 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 (CGH) 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 specimens.

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 – a disability characterized by significant limitations in both intellectual functioning and in adaptive behavior, which covers many everyday social and practical skills. This disability originates before the age of 18.

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. 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 is 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

Program Exception

Chromosomal Microarray Analysis (CMA) (CPT codes 81228 & 81229) requires a Program Exception, the ordering physician must provide ALL of the following:

- A supporting statement indicating why a requested therapy or device is medically necessary, and the alternative options have been or are likely to be ineffective, adversely affect patient compliance, or cause an adverse reaction., AND
- Documentation that medical necessity criteria are met.

1. Chromosomal Microarray Analysis (CMA) is considered medically necessary when ALL of the following criteria is met:
   A. The individual must be under the age of 21; AND
   B. The individual's parents have been engaged in face-to-face genetic counseling with a qualified 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 CMA testing results will guide clinical decisions that would not otherwise be made in the absence of the testing; AND
   E. The individual must have previously exhibited ANY of the following conditions:
      1) Multiple congenital anomalies not specific to a well-delineated genetic syndrome. Multiple congenital anomalies are defined as:
         a) Two (2) or more major anomalies affecting different organ systems; OR
         b) 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
      2) Apparent non-syndromic developmental delay/intellectual disability; OR
      3) Autism Spectrum Disorder (e.g., Asperger's, autistic disorder, pervasive developmental disorder).

2. CMA of amniotic fluid, placenta, or products of conception (POC) for evaluation of pregnancy loss is considered medically necessary under ANY of the following conditions:
   A. In cases of pregnancy loss at twenty (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 twenty (20) weeks of gestation.

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

3. When CMA laboratory services are considered not medically necessary:
   - CMA services performed for any conditions other than those listed above is considered not medically necessary because the scientific evidence has not been established.
   - CMA is 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.
   - 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 is 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.

4. 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 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 physician of appropriate expertise or other obstetrical provider specializing in the care for the indication(s) for genetic testing
   -
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 CMA laboratory testing is outpatient.

7. Related Policies
   - MP-071-MD-PA Non-Oncologic Genetic Testing Panels
   - MP-003-MD-PA Fetal Aneuploidy Testing Using Noninvasive Cell-Free Fetal DNA
   - MP-013-MD-PA Whole Exome and Whole Genome Sequencing for Diagnosis of Genetic Disorders

Operational Guidelines *Do not include on external version*

- For both professional and facility providers, the diagnosis and procedure codes are applied on a pre-service, prepayment basis.
- Patients must be under the age of 21 to be eligible for CPT codes 81228, 81229 & S3870 with any of the covered diagnosis codes listed in the Coding Requirements section.
- Claims for services that do not meet the procedure codes and the diagnosis codes in the Coding Requirements section, should deny as experimental/investigational and therefore not medically necessary.
- Any approval requests for CMA laboratory testing that do not meet the guidelines listed above will require a review by a Medical Director on a case-by-case basis.
- CPT codes 81228 & 81229 require a Program Exception per DHS TAG determination.

Governing Bodies Approval

Genetic testing includes 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 a specific test, but rather there is procedural compliance.

The Pennsylvania Department of Human Services Technology Assessment Group (TAG) workgroup meets quarterly to discuss issues revolving around new technologies and technologies or services that were previously considered to be a program exception. During this meeting, decisions are made as to whether or not certain technologies will be covered and how they will be covered. TAG's decisions are as follow:

- Option #1: Approved - Will be added to the Fee Schedule
- Option #2: Approved as Medically Effective - Will require Program Exception
- Option #3: Approved with (or denied due to) Limited/Minimal Evidence of Effectiveness - Will require Program Exception
- Option #4: Denied - Experimental/Investigational

As of March 2014, the TAG workgroup assigned chromosomal microarray analysis/comparative genomic hybridization/single nucleotide polymorphism an Option # 3, specifically for CPT codes 81228 and 81229.

Program Exception
CMA, comparative genomic hybridization, and single nucleotide polymorphism requires a program exception, the ordering physician must provide a supporting statement indicating why the requested therapy or device is medically necessary, and the alternative options have been or are likely to be ineffective, adversely affect patient compliance, or cause an adverse reaction.

Other Insurers (not for Provider version)

Insurer	Coverage	Date
UPMC	Genetic Testing -Chromosomal Microarray (CMA) Covered when medically necessary for pre- & postnatal testing, as well as parental testing when karyotyping is not applicable	07/2023
AmeriHealth Commercial	Genetic Testing - Whole-genome chromosomal microarray analysis (CMA) is considered medically necessary and, therefore, covered as first-line testing in individuals with any of the listed conditions	09/2023
Aetna	Comparative Genomic Hybridization (CGH) – Aetna considers comparative genomic hybridization (CGH) medically necessary for the listed indications	10/2023
Cigna	Comparative Genomic Hybridization Prenatal testing is considered medically necessary in women undergoing invasive prenatal genetic testing, or intrauterine fetal loss at ≥20 weeks or stillbirth	06/2023

UHC Community Plan	Chromosome Microarray Testing - medically necessary for women undergoing invasive prenatal testing (i.e., amniocentesis, chorionic villus sampling or fetal tissue sampling)	10/2023
Unicare	Chromosomal Microarray Analysis (CMA) for Developmental Delay, Autism Spectrum Disorder, Intellectual Disability (Intellectual Developmental Disorder) and Congenital Anomalies - medically necessary for postnatal as well as an alternative to fetal karyotyping	06/2023
Highmark Commercial	Chromosomal Microarray for Prenatal Diagnosis Chromosomal Microarray Testing For Developmental Disorders	02/2023

Summary of Literature

According to the World Health Organization (WHO), a congenital anomaly can be defined as structural or functional anomalies that occur during intrauterine life. Also called birth defects, congenital disorders, or 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. 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 (WHO, 2020).

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. 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) (ACOG, 2020).

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).

Rationale

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 (ACMG) published recommendations on the array-based technologies and the clinical utilization for detecting chromosomal abnormalities:

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 (Manning, Hudgens, 2010).

ACMG guidelines also state 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. 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 (UpToDate, 2017).

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 ACOG and the Society for Maternal-Fetal Medicine (SMFM) recommend use of CMA of fetal tissue ((ie, amniotic fluid, placenta, or products of conception) in the evaluation of intrauterine fetal death or stillbirth when further cytogenetic analysis is desired because of the test's increased likelihood of obtaining results and improved detection of causative abnormalities (ACOG, 2016).

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 tested the hypothesis that chromosomal microarray analysis frequently diagnoses conditions that require specific medical follow-up and that referring physicians 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 physician 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 physicians 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. 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).

Hayes, Inc.

- Clinical Utility of Prenatal Genetic Testing for Autism Spectrum Disorder
    - **1 – Insufficient**: For use of genetic testing for autism spectrum disorder (ASD) during the prenatal period to independently predict risk in the fetus and improve postnatal outcomes. No peer-reviewed studies were identified that provide evidence for the clinical utility of genetic testing for ASD during the prenatal period to independently predict risk in the fetus and improve postnatal outcomes. Studies are needed that enroll pregnant women who elect prenatal genetic testing, clearly identify those whose results include fetal pathogenic variants associated with ASD risk, and report on prenatal and postnatal outcomes, following ASD-related outcomes for at least a few years after birth.

Coding Requirements

Procedure Codes

CPT / HCPCS 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
S3870	Comparative genomic hybridization (CGH) microarray testing for developmental delay, autism spectrum disorder and/or intellectual disability

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
F79	Unspecified intellectual disabilities
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
F80.9	Developmental disorder of speech and language, unspecified
F81.0	Specific reading disorder
F81.2	Mathematics disorder
F81.81	Disorder of written expression
F81.89	Other developmental disorders of scholastic skills
F81.9	Developmental disorder of scholastic skills, unspecified
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
F84.9	Pervasive developmental disorder, unspecified
F88	Other disorders of psychological development
F89	Unspecified disorder of psychological development
F90.8	Attention-deficit hyperactivity disorder, other type
H93.25	Central auditory processing disorder
P02.9	Newborn affected by abnormality of membranes, unspecified
Q00.0	Anencephaly
Q00.1	Craniorachischisis
Q00.2	Iniencephaly
Q01.0	Frontal encephalocele
Q01.1	Nasofrontal encephalocele
Q01.2	Occipital encephalocele
Q01.8	Encephalocele of other sites
Q01.9	Encephalocele, unspecified
Q02	Microcephaly
Q03.0	Malformations of aqueduct of Sylvius
Q03.1	Atresia of foramina of Magendie and Luschka
Q03.8	Other congenital hydrocephalus
Q03.9	Congenital hydrocephalus, unspecified
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
Q04.9	Congenital malformation of brain, unspecified
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
Q05.9	Spina bifida, unspecified
Q06.0	Amyelia
Q06.1	Hypoplasia and dysplasia of spinal cord
Q06.2	Diastematomyelia
Q06.3	Other congenital cauda equine malformations
Q06.4	Hydromyelia
Q06.8	Other specified congenital malformations of spinal cord
Q06.9	Congenital malformation of spinal cord, unspecified
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
Q07.9	Congenital malformation of nervous system, unspecified
Q89.7	Multiple congenital malformations, not elsewhere classified
Q89.8	Other specified congenital malformations
Q89.9	Congenital malformation, unspecified
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
Q92.9	Trisomy and partial trisomy of autosomes, unspecified
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
Q93.9	Deletion from autosomes, unspecified
Q95.2	Balanced autosomal rearrangement in abnormal individual
Q95.3	Balanced sex/autosomal rearrangement in abnormal individual
Q99.8	Other specified chromosome abnormalities
Q99.9	Chromosomal abnormality, unspecified
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.

Reference Sources

American Association on Intellectual and Developmental Disabilities. Intellectual Disability: Definition, Classification, and Systems of Supports. 12th Edition, 2021. Accessed on September 16, 2021.

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Fruhman G, Van den Veyver IB. Applications of array comparative genomic hybridization in obstetrics. Obstet Gynecol Clin North Am. March 2010. Accessed on September 16, 2021.

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