Chromosomal Microarray Analysis (CMA)

Last Review Date: October 12, 2018	Number: MG.MM.LA.36aC2

Definition

Chromosomal Microarray Analysis (CMA) is a high-resolution whole-genome diagnostic tool which detects genomic copy number variations (CNVs). CNVs are chromosomal imbalances that result from the deletion and/or duplication of one or more sections of DNA. CMA is used in postnatal settings to diagnose individuals with congenital anomalies, unexplained developmental delay (DD), autism spectrum disorder (ASD) or intellectual disability (intellectual developmental delay); and in prenatal settings for miscarried fetuses and still births.

CMA which is also known as cytogenomic microarray analysis encompasses two different laboratory techniques, comparative genomic hybridization (aCGH) and single nucleotide polymorphism (SNP) arrays.

Related Guidelines

Genetic Counseling and Testing

Guideline

1. CMA genetic testing is considered medically necessary for the evaluation of a fetus when **ANY** of the following criteria are met:
    a. Abnormal fetal anatomic findings characteristic of a genetic abnormality and detected on fetal ultrasound or fetal magnetic resonance imaging
    b. Analyses of stillbirths with congenital anomalies or in stillbirths in which karyotype results cannot be obtained
    c. In patients with a structurally normal fetus undergoing invasive prenatal diagnostic testing
    d. The individual is considered at high risk for fetal aneuploidy due to ANY of the following:
        i. The expectant mother has a history of a prior pregnancy with a trisomy
        ii. The expectant mother will be 35 years or older at the time of delivery
        iii. The expectant mother has a positive first or second-trimester standard biomarker screening test

2. CMA genetic testing is considered medically necessary for diagnosing developmental delay (DD), intellectual disability (ID) or autism spectrum disorder (ASD) in children when **ALL** of the following criteria are met:
    a. A negative biochemical test for metabolic diseases, if indicated by the clinical presentation
    b. A negative targeted genetic test, (for example: FMR1 gene analysis for Fragile X), if or when indicated by the clinical and family history
    c. The member's clinical presentation is not specific to a well-delineated genetic syndrome
    d. Congenital anomalies, birth defects and anatomic malformations not part of a recognizable genetic or non-genetic syndrome
3. The results of the genetic test have the potential to impact the clinical management of the member.

Limitations/Exclusions

CMA genetic testing is considered investigational and not medically necessary for all indications when the above criteria are not met.

Revision History

11/11/16 — added congenital anomalies, birth defects and anatomic malformations not part of a recognizable genetic or non-genetic syndrome to prerequisite criteria.

Applicable Procedure Codes

81228	Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, 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
81405	Molecular pathology procedure, Level 6 (eg, analysis of 6-10 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of 11-25 exons, regionally targeted cytogenomic array analysis) CPOX (coproporphyrinogen oxidase) (eg, hereditary coproporphyria), full gene sequence CTRC (chymotrypsin C) (eg, hereditary pancreatitis), full gene sequence PKLR (pyruvate kinase, liver and RBC) (eg, pyruvate kinase deficiency), full gene sequence (Revision eff. 01/01/2018)
S3870	Comparative genomic hybridization (CGH) microarray testing for developmental delay, autism spectrum disorder and/or intellectual disability
88230	Tissue culture for non-neoplastic disorders; lymphocyte
88262	Chromosome analysis; count 15-20 cells, 2 karyotypes, with banding

Applicable ICD-10 Diagnosis Codes

E78.71	Barth syndrome
E78.72	Smith-Lemli-Opitz syndrome
F70 - F90.9	Mental, behavioral and neurodevelopmental disorders
G31.81	Alpers disease
G31.82	Leigh's disease
G90.1	Familial dysautonomia [Riley-Day]
H93.25	Central auditory processing disorder
O09.511 - O09.529	Supervision of elderly gravida

O35.1xx0 - O35.1xx9	Maternal care for (suspected) chromosomal abnormality in fetus
O35.2xx0 - O35.2xx9	Maternal care for (suspected) hereditary disease in fetus
P02.8	Newborn affected by other abnormalities of membranes
P02.9	Newborn affected by abnormality of membranes, unspecified
P29.30	Pulmonary hypertension of newborn
P29.38	Other persistent fetal circulation
Q00.0 - Q99.9	Congenital malformations, deformations and chromosomal abnormalities
R48.0	Dyslexia and alexia
R62.0	Delayed milestone in childhood
R62.50-R62.59	Unspecified/other lack of expected normal physiological development in childhood
R89.8	Other abnormal findings in specimens from other organs, systems and tissues
Z13.4	~~Encounter for screening for certain developmental disorders in childhood~~ (deleted as of 10/01/2018)
Z13.71	Encounter for nonprocreative screening for genetic disease carrier status
Z13.79	Encounter for other screening for genetic and chromosomal anomalies
Z13.89	Encounter for screening for other disorder
Z36.0	Encounter for antenatal screening for chromosomal anomalies
Z36.1	Encounter for antenatal screening for raised alphafetoprotein level
Z36.2	Encounter for other antenatal screening follow-up
Z36.3	Encounter for antenatal screening for malformations
Z36.4	Encounter for antenatal screening for fetal growth retardation
Z36.5	Encounter for antenatal screening for isoimmunization
Z36.81	Encounter for antenatal screening for hydrops fetalis
Z36.82	Encounter for antenatal screening for nuchal translucency
Z36.83	Encounter for fetal screening for congenital cardiac abnormalities
Z36.84	Encounter for antenatal screening for fetal lung maturity
Z36.85	Encounter for antenatal screening for Streptococcus B
Z36.86	Encounter for antenatal screening for cervical length
Z36.87	Encounter for antenatal screening for uncertain dates
Z36.88	Encounter for antenatal screening for fetal macrosomia
Z36.89	Encounter for other specified antenatal screening
Z36.8A	Encounter for antenatal screening for other genetic defects
Z37.1	Single stillbirth
Z37.3	Twins, one liveborn and one stillborn
Z37.4	Twins, both stillborn
Z37.69	Other multiple births, some liveborn
Z37.7	Other multiple births, all stillborn

References

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