GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL ANOMALIES

Coverage for services, procedures, medical devices and drugs are dependent upon benefit eligibility as outlined in the member’s specific benefit plan. This Medical Coverage Guideline must be read in its entirety to determine coverage eligibility, if any.

This Medical Coverage Guideline provides information related to coverage determinations only and does not imply that a service or treatment is clinically appropriate or inappropriate. The provider and the member are responsible for all decisions regarding the appropriateness of care. Providers should provide BCBSAZ complete medical rationale when requesting any exceptions to these guidelines.

The section identified as “Description” defines or describes a service, procedure, medical device or drug and is in no way intended as a statement of medical necessity and/or coverage.

The section identified as “Criteria” defines criteria to determine whether a service, procedure, medical device or drug is considered medically necessary or experimental or investigational.

State or federal mandates, e.g., FEP program, may dictate that any drug, device or biological product approved by the U.S. Food and Drug Administration (FDA) may not be considered experimental or investigational and thus the drug, device or biological product may be assessed only on the basis of medical necessity.

Medical Coverage Guidelines are subject to change as new information becomes available.

For purposes of this Medical Coverage Guideline, the terms "experimental" and "investigational" are considered to be interchangeable.

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GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL ANOMALIES (cont.)

Description:

Chromosomal microarray testing (CMA) is a laboratory test that increases the chromosomal resolution for detection of abnormalities. It has been used for detection of genetic imbalances in infants or children with characteristics of autism spectrum disorder (ASD), developmental delay/intellectual disability (DD/ID) and congenital anomalies. It has also been investigated as a prenatal test of a fetus that may be at high risk for a chromosomal abnormality. CMA was previously referred to as array comparative genomic hybridization (aCGH).

Tests include, but are not limited to:

- Affymetrix CytoScan® Dx Assay
- FirstStepDx PLUS (postnatal)
- GenomeDx whole genome array (postnatal)
- Reveal® SNP microarray-Pediatric
- SignatureChipOS® microarray (postnatal)
- Signature PrenatalChip®OS microarray (postnatal)

Next generation sequencing (NGS) panel testing allows for simultaneous analysis of multiple genes, and has been investigated as a way to identify single gene causes of syndromes that have autism as a significant clinical feature, in individuals with normal array-based testing.

Tests include, but are not limited to:

- devSEEK® and devSEEK Triome™
- devACT® Clinical Management Panel
- Mount Sinai School of Medicine Department of Genetics 30 Gene Sequencing Panel Autism
- XLID Next-Gen Panel™ (XLMR), Ambry Genetics
- X-Linked Intellectual Disability (XLID), Greenwood Genetic Center
- X-Linked Intellectual Disability Gene Panel, Emory Genetics Laboratory
GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL ANOMALIES (cont.)

Definitions:

Genetic Testing:
Analysis of DNA, RNA, chromosomes, proteins and certain metabolites in order to detect alterations related to an inherited disorder.

Gene:
A hereditary unit consisting of segments of DNA that occupies a specific location on chromosomes. Genes undergo mutation when their DNA sequence changes.

Genetic Counseling:
Instruction that provides interpretation of genetic tests and information about courses of action that are available for the care of an individual with a genetic disorder or for future family planning.

Affected Individual:
An individual displaying signs or symptoms characteristic of a suspected or specific inherited disorder.

Unaffected Individual:
An individual who displays no signs or symptoms characteristic of a suspected or specific inherited disorder.

Screening:
Genetic screening is the testing of an individual with no symptoms for a specific inherited disorder to determine if the individual carries an abnormal gene. Screening can be used to predict risk or potential risk for the individual or their offspring.
GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL (cont.)

Criteria:

Other BCBSAZ Medical Coverage Guidelines may exist for diagnoses not addressed in this guideline.

- Genetic testing and/or counseling of an unaffected individual, regardless of risk factors is considered screening and not eligible for coverage.

- Genetic testing and/or counseling of an affected individual to confirm a disease when confirmation of the diagnosis would not impact the care and/or management is considered not medically necessary and not eligible for coverage.

- Chromosomal microarray analysis of an affected individual is considered medically necessary as first-line testing in the initial postnatal evaluation with documentation of ANY of the following:
  1. Apparently nonsyndromic developmental delay/intellectual disability
  2. Autism spectrum disorder
  3. Multiple congenital anomalies not specific to a well-delineated genetic syndrome

- Chromosomal microarray analysis for all other indications not previously listed or if above criteria not met is considered experimental or investigational based upon:
  1. Insufficient scientific evidence to permit conclusions concerning the effect on health outcomes, and
  2. Insufficient evidence to support improvement of the net health outcome, and
  3. Insufficient evidence to support improvement of the net health outcome as much as, or more than, established alternatives, and
  4. Insufficient evidence to support improvement outside the investigational setting.
GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL ANOMALIES (cont.)

Criteria: (cont.)

- Genetic testing and/or counseling using panel testing with next generation sequencing in all cases of suspected genetic abnormality in children with developmental delay/intellectual disability or autism spectrum disorder or congenital anomalies is considered experimental or investigational based upon:
  1. Insufficient scientific evidence to permit conclusions concerning the effect on health outcomes, and
  2. Insufficient evidence to support improvement of the net health outcome, and
  3. Insufficient evidence to support improvement of the net health outcome as much as, or more than, established alternatives, and
  4. Insufficient evidence to support improvement outside the investigational setting.

Resources:

Literature reviewed 10/11/16. We do not include marketing materials, poster boards and non-published literature in our review.

The BCBS Association Medical Policy Reference Manual (MPRM) policy is included in our guideline review. References cited in the MPRM policy are not duplicated on this guideline.

Resources prior to 03/20/2013 may be requested from the BCBSAZ Medical Policy and Technology Research Department.


GENETIC TESTING FOR DEVELOPMENTAL DELAY/INTELLECTUAL DISABILITY, AUTISM SPECTRUM DISORDER AND CONGENITAL ANOMALIES (cont.)

Resources: (cont.)


