MEDICAL REVIEW GUIDELINE

Genetic Testing

Adopted by Medical Care Management
Committee on May 26, 2011
Revised June 5, 2023

Title: Genetic Testing

This Medical Review Guideline (Guideline) is provided for informational purposes only and does not constitute medical advice. It is intended solely for the use of Community Health Choice, Inc. (Community) clinical staff as a guideline for use in determining medical necessity of requested procedures and therapy. This Guideline does not address eligibility or benefit coverage. Other Policies and Coverage Determination Guidelines may apply. All reviewers must first identify enrollee eligibility, any federal or state regulatory requirements and the plan benefit coverage prior to use of this Medical Review Guideline. If there is a discrepancy between this Guideline and a member's benefit plan, summary plan description or contract, the benefit plan, summary plan description or contract will govern. Community reserves the right, in its sole discretion, to modify its Policies and Guidelines as necessary.

PURPOSE:

Genetic disorders are often diagnosed based on symptoms, physical findings and biochemical tests. The field of genetic testing is rapidly evolving. In some situations, genetic testing may be used to arrive at a diagnosis when other tests are not available or are not helpful. However, there are also limitations. A specific genetic abnormality may be observed but this may or may not reliably identify a disorder and may or may not result in improved care or treatment of the individual.

GUIDELINE:

Community considers genetic testing to be medically necessary for diagnostic purposes when **all** of the following criteria are met:

1. The member displays clinical features, or is at direct risk of inheriting the mutation in question (pre-symptomatic); ***and***
2. The results of the test are essential to the medical management of the member (e.g. will result in initiation of new course of therapy, alter an existing or proposed therapy, or determine future care); ***and***
3. A definitive diagnosis remains uncertain after history, physical examination, pedigree analysis, genetic counseling, and the suspected genetic disorder cannot be diagnosed by biochemical or other laboratory test; ***and***
4. The test has been established by clear and convincing evidence in the scientific literature to be reliably associated with the specific disease or disorder.

Situations where genetic testing is specifically considered not medically necessary:

1. Testing for screening purposes, or without a reasonable likelihood of confirming a specific genetic disorder.
2. When the diagnosis can be made clinically.

3. Testing of family members.
4. Repeat genetic testing.
5. Direct-to-Consumer (at-home) genetic testing.

SPECIFIC CONDITIONS:
The following are guidelines for genetic testing for specific conditions:

Autism Spectrum Disorder (ASD) or Pervasive Developmental Disorder (PDD):

- Chromosomal Microarray Analysis (CMA) is a first line test for children with ASD/PDD.
- Karyotyping may be warranted if a balanced translocation is suspected.
- FMR1 gene testing for fragile X syndrome is appropriate.
- Additional genetic testing (beyond CMA, fragile X, and possibly karyotype) is indicated only for individuals with dysmorphic features, microcephaly, macrocephaly, or cognitive impairment. Specific testing should be guided by the member's individual clinical profile.

Genetic Testing in Intellectual Disability (ID)/Global Developmental Delay (GDD):

- If a specific diagnosis is suspected it should be confirmed using appropriate diagnostic studies including single-gene tests if necessary or CMA.
- If diagnosis is unknown or not clinically suspected the following evaluation is recommended:
    - CMA
    - Fragile X gene testing
    - Consider specific metabolic testing based on history and physical examination findings, and should include serum total homocysteine, acyl-carnitine profile, amino acids; and urine organic acids, glycosaminoglycans, oligosaccharides, purines, pyrimidines, GAA/creatine metabolites.

Genetic Testing in Major Congenital Anomalies:

- CMA and additional genetic testing may be indicated when a major malformation or multiple clinical findings and laboratory values suggest a genetic etiology but when the clinician is unable to give a clear diagnosis for which the results would be essential to medical management.

Multigene panel testing for genetic mutations in cancer:
Community considers a multigene panel testing in cancer to be medically necessary when **all** of the following criteria are met:

- The member has a diagnosis of cancer; and
- The member has pretest genetic counseling with a health professional who does not work for a testing laboratory; and

- All the genetic tests in the panel are essential and will affect the cancer treatment options; and
- All the genetic tests in the panel meet NCCN or InterQual guidelines' genetic testing criteria and management recommendations.