Genetic Testing to Diagnose Non-Cancer Conditions

Description

Diagnostic testing is performed in patients with clinical signs or symptoms of a non-cancer genetic condition. The genetic test may confirm or rule out a clinical diagnosis. In some cases, genetic testing is the gold standard for making a diagnosis based on evidence- or consensus-based guidelines. In others, it may be used to confirm a clinical diagnosis, offer prognostic information that impacts management, or rule out a diagnosis in the differential. Often, diagnostic testing of an affected individual will offer results that are relevant to the testing of other family members.

- This guideline does not include risk assessment or predictive testing for at-risk, asymptomatic individuals. Please refer to Genetic Testing to Predict Disease Risk for that purpose.
- Diagnostic testing of a pregnancy or an embryo is addressed by guidelines on Genetic Testing for Prenatal Screening and Diagnostic Testing and Preimplantation Genetic Screening and Diagnosis, respectively.
- In addition, testing for hereditary cancer syndromes is addressed separately under Genetic Testing for Cancer Susceptibility and Hereditary Cancer Syndromes.

Criteria

Criteria: General Coverage Guidance

Individuals may be considered for diagnostic genetic testing when ALL of the following conditions are met:

- **Clinical signs and symptoms** in the individual are consistent with the diagnosis in question.
- **Technical and clinical validity**: The test must be accurate, sensitive and specific, based on sufficient, quality scientific evidence to support the claims of the test.
- **Clinical utility**: Healthcare providers can use the test results to provide significantly better medical care for the individual.
- **Reasonable use**: The usefulness of the test is not significantly offset by negative factors, such as expense, clinical risk, or social or ethical challenges.

Limits:
• Testing will be considered only for the number of genes or tests necessary to establish mutation status. A tiered approach to testing, with reflex to more detailed testing and/or different genes, will be required when clinically possible.

• Diagnostic genetic testing will be allowed once per lifetime per condition. Exceptions may be considered if technical advances in testing demonstrate significant advantages that would support a medical need to retest.

Criteria: Test-specific Guidelines

Test-specific guidelines are available for some tests designed to diagnosis non-cancer conditions. For tests without a specific guideline, use the General Coverage Guidance.

Benefit exclusion

Exclusions and other considerations

Testing unaffected individuals (e.g. carrier testing, predictive testing, presymptomatic testing, etc) is a BCBSAZ benefit exclusion and, therefore, not eligible for reimbursement.