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: Special Circumstances

Diagnostic testing of an individual to inform reproductive planning and testing for parents or testing for siblings.

Diagnostic genetic testing may be requested in a symptomatic individual with a known genetic condition. While diagnostic testing may not impact management of the affected individual, the information gained from genetic testing may be needed to perform accurate carrier testing in the parent(s), genetic diagnosis in a pregnancy, or genetic diagnosis in a sibling.*

In these circumstances, diagnostic genetic testing in a symptomatic individual may be considered when ALL of the following conditions are met:

• The diagnosis of the disease in the affected individual is certain or highly probable based on clinical signs and symptoms, history, imaging, and/or results of other laboratory testing.

• The results of the genetic test in the symptomatic individual must be required in order to perform accurate carrier testing in the parent(s), genetic diagnosis in a pregnancy, or genetic diagnosis in a sibling.

• 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 informative genetic testing for the sibling, parents, or for a current or future at-risk pregnancy.

• 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 indicated only for the number of genes or tests necessary to establish the familial mutation(s). 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.

*Parent or sibling must also be a covered member under the same health plan.
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 in Section 1.