Genetic Testing to Predict Disease Risk

Description

Predictive genetic testing is performed in people known to be at increased risk of developing an inherited non-cancer condition (for the purposes of this guideline) based on their family history. For some conditions, a positive genetic test predicts with certainty that the person will eventually develop signs and symptoms of a condition. For other conditions, a positive genetic test result indicates an increased risk (susceptibility) for a condition. A negative result may rule out a condition, or lower the risk significantly. Having test results may improve medical management through improved screening, preventive measures, prophylactic medication, and other means.

- This guideline does not include testing of a symptomatic individual. Please refer to Genetic Testing to Diagnose Non-Cancer Conditions for that purpose.
- Predictive testing for hereditary cancer syndromes is addressed separately under Genetic Testing for Cancer Susceptibility and Hereditary Cancer Syndromes.
- Testing of minors is addressed separately under Genetic Presymptomatic and Predictive Testing for Adult-Onset Conditions in Minors.

Criteria

Criteria: General Coverage Guidance

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

- The individual is known to be at-risk for developing inherited condition because a parent, sibling, or child is affected by or known to be a carrier of a genetic disease.
- **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 carrier status. A tiered approach to testing, with reflex to more detailed testing and/or different genes, will be required when clinically possible.

• Predictive 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.

• Predictive testing will be considered only for adult individuals (age 18 and over). Exceptions may be considered if there are medical management and/or significant psychosocial benefits to testing prior to adulthood.\(^1,2,3\)

Criteria: Special circumstances

Testing for Known Familial Mutations

The genetic mutation(s) associated with a genetic disease can often be defined in an affected family member, allowing for testing of at-risk relatives for those specific mutations. Testing for known familial mutations may be considered when **ALL** of the following conditions are met:

- The mutations in the family have been **clearly defined** by previous genetic testing and **information about those mutations can be provided** to the testing lab.
- **Technical and clinical validity**: The test must be accurate, sensitive and specific to the familial mutations.
- **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 known familial mutations when clinically possible.
- Predictive genetic testing will be allowed once per lifetime per condition.
- Predictive testing will be considered only for adult individuals (age 18 and over). Exceptions may be considered if there are medical management and/or significant psychosocial benefits to testing prior to adulthood.\(^1,2,3\)

Criteria: Test-specific Guidelines

Test-specific guidelines are available for some tests designed to predict disease risk. For tests without a specific guideline, use the General Coverage Guidance.
References

