Clinical Use Guidelines

Genetic Testing to Predict Disease Risk

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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 the following conditions are met:

- **Pre and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND**
- **No previous genetic testing of the requested gene that would have included the KFM, AND**
- **KFM is disease-causing (classified as pathogenic or likely pathogenic), AND**
- **Member is a 1st, 2nd, or 3rd degree biological relative of the family member with the KFM, AND**
- **Member is 18 years of age or older, AND**
- **Healthcare providers can use the test results to provide significantly better medical care for the individual, AND**
- **Rendering laboratory is a qualified provider of service per the Health Plan policy.**

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]**

Note **For medical necessity criteria for predictive testing of a known familial mutation in individuals younger than 18 years, see the guideline: *Genetic Presymptomatic and Predictive Testing for Adult-Onset Conditions in Minors*.**

Criteria: Test-specific Guidelines

Test-specific guidelines are available for some tests designed to predict disease risk.

References

1. **Ross LF, Saal HM, David KL, Anderson RR. Technical report: ethical and policy issues in genetic testing and screening of children. *Genet Med* 2013;15: 234–245. doi: 10.1038/gim.2012.176**
2. **National Society of Genetic Counselors Position Statement. Genetic testing of minors for adult-onset conditions. Adopted 2012. Updated 2018. Available at: https://www.nsgc.org/Policy-Research-and-Publications/Position-Statements/Position-Statements/Post/genetic-testing-of-minors-for-adult-onset-conditions**
3. **Botkin, JR, Belmont JW, Berg JS, et al. Points to consider: ethical, legal, and psychosocial implications of genetic testing in children and adolescents. *Am J Hum Genet*. 2015;97:6-21. doi: 10.1016/j.ajhg.2015.05.022**