Genetic Testing for the Screening, Diagnosis, and Monitoring of Cancer

Description

Genetic testing for screening, diagnosis and monitoring of cancer refers to molecular diagnostic tests whose purposes include identifying the possible presence of cancer in asymptomatic, average risk individuals; confirming the absence or presence of cancer; and monitoring the absence or presence of cancer after a prior diagnosis and treatment.

Screening

The goal of cancer screening is to identify the possible presence of cancer before symptoms appear. Screening tests cannot diagnose cancer, but typically determine if there is an increased chance cancer is present, and triages individuals for more invasive, diagnostic testing. Most cancer screening does not include genetic testing, but instead relies on physical exam, radiological exams, or non-genetic laboratory tests. Advances in human genetics, however, have identified several molecular diagnostic tests that may provide clues for early cancer detection.

Diagnosis

When cancer is suspected because of an abnormal screening test or symptoms, blood tests for tumor markers or molecular testing on tissue samples can aid in confirming a diagnosis of cancer. These tests may contribute information to helping the clinician understand prognosis and treatment options.

Monitoring

During treatment, or after an apparently successful treatment, active monitoring is often recommended to identify if the cancer is responding to treatment or has returned or spread, before any symptoms appear. Monitoring may include increased surveillance or routine blood tests for tumor markers, and increasingly, molecular genetic tests.

- Tests used to determine hereditary cancer risk are covered separately as Genetic Testing for Cancer Susceptibility and Hereditary Cancer Syndromes.
- This policy does not address drug response to cancer, or testing to determine which therapies to use. Please refer to Pharmacogenomic Testing for Drug Toxicity and Response for that purpose.
- This policy does not address molecular tumor marker testing in solid tumors. Please refer to Somatic Mutation Testing–Solid Tumors and Liquid Biopsy Testing – Solid Tumors for that purpose.
• This policy does not address diagnostic or predictive testing for conditions other than non-inherited cancer. Refer to *Genetic Testing to Diagnose Non-Cancer Conditions* and *Genetic Testing to Predict Disease Risk* for those purposes.

**Criteria**

**Criteria: General Coverage Guidance**

Individuals may be considered for genetic testing for screening, diagnosing, or monitoring cancer when *ALL* of the following conditions are met:

- *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. A tiered approach to testing, with reflex to more detailed testing and/or different genes, will be required when clinically possible.

- For tests that look for changes in germline DNA (i.e., not tumor DNA or viral DNA), testing will be allowed once per lifetime per gene. 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 screen for, diagnose, or monitor cancer. For tests without a specific guideline, use the General Coverage Guidance in Section 1.