

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

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## 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.

- For information on tests used to determine hereditary cancer risk, please refer to the guideline *Genetic Testing for Cancer Susceptibility and Hereditary Cancer Syndromes*, as this testing is not addressed here.
- For information on drug response to cancer or testing to determine which therapies to use, please refer to the guideline *Pharmacogenomic Testing for Drug Toxicity and Response*, as this testing is not addressed here.

- For information on molecular tumor marker testing in solid tumors, please refer to the guideline *Somatic Mutation Testing* and *Liquid Biopsy Testing*, as this testing is not addressed here.
- For information on diagnostic or predictive testing for conditions other than non-inherited cancer, please refer to the guideline *Genetic Testing to Diagnose Non-Cancer Conditions* and *Genetic Testing to Predict Disease Risk*, as this testing is not addressed here.

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**Note:**

This benefit/harm statement only applies to those jurisdictions that do not have Medicare guidance. Based upon the clinical policy, following EviCore's criteria for genetic testing for the screening, diagnosis, and monitoring of cancer will ensure that testing will be available to those members most likely to benefit from the information provided by the assays. For those not meeting criteria, it ensures alternate diagnostic/management strategies are considered. However, it is possible that some members who would benefit from the testing, but do not meet criteria, will not receive an immediate approval for testing.

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**Criteria****Criteria: General Coverage Guidance**

Genetic testing for screening, diagnosing, or monitoring cancer is medically necessary 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 is medically necessary 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 above.