# Genetic Testing for Carrier Status

**MOL.CU.110.A**
**v1.0.2026**

Genetic testing for carrier status is performed to identify genetic risks that could impact reproductive decision-making for parents or prospective parents. Carriers are generally not affected but have an increased risk to have a child with a genetic condition. Carrier testing may be performed when there is a known elevated risk for a condition based on an individual's family history or ancestry or as a means of screening for reproductive risks in the general population.

## Availability of genetic testing for carrier status

Carrier testing may be available for autosomal recessive conditions, X-linked conditions, and certain chromosome abnormalities. Ideally, carrier screening is performed prior to pregnancy so that a full range of reproductive options are available to an at-risk couple. However, in practice, it is often performed early in pregnancy when prenatal care is established.

### Other applications of carrier testing

For information on prenatal screening and diagnostic testing, please refer to the guideline *Genetic Testing for Prenatal Screening and Diagnostic Testing,* as this testing is not addressed here.

For information on preimplantation genetic screening, please refer to the guideline *Preimplantation Genetic Screening and Diagnosis,* as this testing is not addressed here.

For information on non-invasive prenatal testing, please refer to the guideline *Non-invasive Prenatal Screening*, as this testing is not addressed here.

This guideline does not include testing that may identify carriers who have clinical signs and symptoms, such as cystic fibrosis testing for men with congenital absence of the vas deferens or fragile X genetic testing for women with premature ovarian failure. For information on this, please refer to the test specific guideline or *Genetic Testing to Diagnose Non-Cancer Conditions.*

**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 carrier status 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





730 Cool Springs Blvd, Franklin, TN 37067 (800) 918-8924 www.EviCore.com

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.

## Criteria

Requests for genetic testing for carrier status are reviewed using these criteria.

### Criteria for general coverage guidance

Genetic testing for carrier status 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 and/or assist individuals with reproductive planning. |
| **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 is only medically necessary 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.
- Carrier testing will be allowed once per lifetime. Exceptions may be considered if technical advances in testing demonstrate significant advantages that would support a medical need to retest.
- Carrier testing is only medically necessary in adults. Carrier screening in minor children is not medically necessary, except in the case of a pregnancy of the minor child.

#### Carrier testing based on family history

Carrier testing based on a family history of a genetic condition is medically necessary when ALL of the following conditions are met in addition to the general criteria above:

- The diagnosis of a genetic condition in a family member is known.
- The parent(s) or prospective parent(s) are at-risk to be carriers of that condition based on the pattern of inheritance.

- The genetic condition is associated with potentially severe disability or has a lethal natural history.

**Partner testing of known carrier or affected individuals**

Carrier screening is medically necessary in individuals whose partners are known carriers or affected individuals when all of the following conditions are met in addition to the general criteria above:

- The diagnosis of a genetic condition or carrier status in the partner is known.
- The genetic condition is associated with potentially severe disability or has a lethal natural history.

**Test-specific guidelines**

Test-specific guidelines are available for some tests designed to predict carrier status. For tests without a specific guideline, use the General Coverage Guidance above.





730 Cool Springs Blvd, Franklin, TN 37067 (800) 918-8924 www.EviCore.com