Introduction

Carrier screening 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.

Availability of genetic testing for carrier status

Carrier screening 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

This guideline does not address prenatal or preimplantation genetic testing. Refer to guidelines on Genetic Testing for Prenatal Screening and Diagnostic Testing and Preimplantation Genetic Screening and Diagnosis for those purposes.

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, is addressed in test specific guidelines or Genetic Testing to Diagnose Non-Cancer Conditions.

Criteria

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Requests for carrier screening are reviewed using these criteria.

Criteria for general coverage guidance

Individuals may be considered for genetic testing for carrier screening 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 will only be considered 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 indicated only in adults. Carrier screening in minor children is not indicated, except in the case of a pregnancy of the minor child.

**Routine carrier screening**

Individuals may be considered for routine carrier screening when testing is supported by evidence-based guidelines from governmental organizations and/or well-recognized professional societies in the United States.¹²³

**Carrier screening based on family history**

Individuals may be considered for carrier screening based on a family history of a genetic condition 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**

Individuals may be considered for carrier screening if their partners are known carrier 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 in Section 1.

References

Introduction

This guideline cites the following references.

