Genetic Testing for Carrier Status

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

Criteria: General Coverage Guidance

Testing unaffected individuals (e.g. carrier testing, predictive testing, presymptomatic testing, etc) is a BCBSAZ benefit exclusion and, therefore, not eligible for reimbursement.

Criteria: 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.
References

Introduction

This guideline cites the following references.


2. ACOG Committee Opinion 691: Carrier screening for genetic conditions. March 2017. Available at: https://www.acog.org/Clinical-Guidance-and-Publications/Committee-Opinions/Committee-on-Genetics/CARRIER-Screening-for-Genetic-Conditions