Genetic Testing for Prenatal Screening and Diagnostic Testing

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Description

Prenatal screening and diagnostic testing is performed during pregnancy to identify fetuses at increased risk for or affected with genetic conditions and birth defects. Screening with ultrasound and maternal serum markers is routinely offered. Prenatal diagnosis by chorionic villus sampling or amniocentesis for chromosome abnormalities is available to all women; however, it is usually offered specifically to those at higher risk because of maternal age, a positive screen result, abnormal ultrasound findings, or known risk of a genetic condition based on family history. Investigations for fetal infection and blood antigen incompatibility may also be performed in the prenatal period. Results of testing are used to guide reproductive decision-making, pregnancy management and anticipatory management of the infant at birth.

- For information on prenatal or preconception carrier screening or preimplantation genetic testing, please refer to the guidelines *Genetic Testing for Carrier Status* and *Preimplantation Genetic Screening and Diagnosis,* as this testing is not addressed here.

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 prenatal screening and diagnostic testing will ensure that testing will be available to those members most likely to benefit from a genetic diagnosis. 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.

Criteria

Criteria: General Coverage Guidance

Genetic testing for prenatal screening and diagnostic testing 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.

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- **Clinical utility**: Healthcare providers can use the test results to provide significantly better medical care and/or assist patients 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 covered for the number of genes or tests necessary to establish a prenatal diagnosis. A tiered approach to testing, with reflex to more detailed testing and/or different genes, will be required when clinically possible.
- Prenatal diagnostic testing is medically necessary once per pregnancy. Exceptions may be considered if ambiguous results require retesting for clarification.
- If prenatal samples are studied concurrently with a maternal DNA sample to rule out prenatal analytic errors due to maternal cell contamination, a single unit of CPT code 81265 is reimbursable.

Criteria: Special Prenatal Diagnosis Circumstances

Each of the following sections addresses a group of tests that are used for similar purposes in pregnancy. Because a variety of tests may be used, but the circumstances that justify testing are the same, individual test-specific policies are not necessary.

Prenatal diagnostic testing based on family history

Prenatal genetic testing, generally by amniocentesis or CVS, for the diagnosis of a genetic condition is medically necessary when the following conditions are met:

- The pregnancy is at an increased risk for a genetic disease based on the inheritance pattern of the disorder in question and ANY of the following:
  - At least one parent is known or suspected to be a carrier of a genetic condition based on the family history and/or previous carrier testing results; or
  - One or both parent(s) are affected with a genetic condition; or
  - A sibling is affected with a genetic condition; AND
- The genetic condition is associated with potentially severe disability or has a lethal natural history.

Fetal infectious disease testing

Genetic testing may be used for the diagnosis of an infectious disease (e.g., cytomegalovirus, toxoplasmosis, parvovirus B19, and varicella zoster) in a fetus according to current guidelines from the American College of Obstetricians and Gynecologists (ACOG).[1] Prenatal testing, generally by amniocentesis or CVS, is medically necessary when ANY of the following conditions are met:

- Clinical signs and symptoms of a current infection in the mother; OR
- Serologic evidence of a current or recent infection in the mother (with or without clinical signs); OR

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- Fetal abnormalities identified on ultrasound indicating an increased risk for a congenital infection

Criteria: Test-specific Guidelines

- Test-specific guidelines are available for some prenatal screening tests and diagnostic tests. For tests without a specific guideline, use the General Coverage Guidance above.

References

1. ACOG Practice Bulletin. Cytomegalovirus, Parvovirus B19, Varicella Zoster, and Toxoplasmosis in Pregnancy. Number 151, June 2015 (reaffirmed 2024). *Obstet Gynecol.* 2015;125(6):1510-1525.

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