Prenatal/Preconception Genetic Testing to Determine Carrier State of a Parent or Prospective Parent

Last Revision/Review Date: November 18, 2020 **P&P # C.6.18**

Policy

The Medical Management Department reviews referral requests for prior authorization of prenatal genetic testing of parents or prospective parents for determination of carrier states for inheritable diseases or disorders.

This Medical Policy does not constitute medical advice. When deciding coverage, the enrollee's specific plan document must be referenced. The terms of an enrollee's plan document (Certificate of Coverage (COC) or Summary Plan Description (SPD)) may differ from this Medical Policy. In the event of a conflict, the enrollee's specific benefit plan document supersedes this Medical Policy. All reviewers must first identify enrollee eligibility, any federal or state regulatory requirements, and the plan benefit coverage prior to use of this Medical Policy. Other Policies and Coverage Determination Guidelines may apply. Quartz reserves the right, in its sole discretion, to modify its Policies and Guidelines as necessary.

Procedure

A. Documentation Required:

1. Name of the test to be performed
2. Characteristics of the requested genetic test
3. Potential impact of genetic test on patient's medical decision making
4. Patient's risk of having the suspected mutation
5. Clinical records of genetic counseling
6. Genetic counseling must be performed by a qualified, appropriately trained practitioner with **ONE** of the following backgrounds:
    a. Board certified/Board eligible (BC/BE) genetics counselor;
    b. BC/BE medical geneticist;
    c. Genetic nurse credentialed as either a genetic clinical nurse or advanced practice nurse in genetics;
    d. Physician specialist or advanced practice professional in obstetrics, gynecology or reproductive medicine with experience or documented training and expertise in prenatal genetics.
    e. The person providing the counseling cannot be employed by a commercial genetic testing laboratory except those employed by/contracted by a laboratory that is part of an integrated health system which routinely delivers health care services beyond just the laboratory test itself.
7. A general description of each specific disease or condition to be tested
8. The ordering providers statement of how the patient's medical treatment plan will be directed by the requested genetic testing whether testing results are positive or negative

B. General Criteria for Medical Necessity: (Used if specific genetic test is not listed in Section C) Prenatal /Preconception Genetic Testing of Inherited Disorders is medically necessary when **ALL** the following criteria are met:

1. The parents or prospective parents are at high risk of a specific genetic disorder with an autosomal recessive pattern of inheritance with an allele frequency of at least 1:100; **AND**

2. A specific mutation, or set of mutations, has been established in the scientific literature to be reliably associated with the disease, including both a clear association between the genotype and phenotype as well as a high allele detection rate; **AND**

3. The genetic disorder is associated with a potentially severe disability or has a lethal natural history; **AND**

4. Testing is accompanied by genetic counseling performed by a qualified, appropriately trained practitioner (see Definitions section above).

C. Medical Necessity Criteria for Specific Genetic Tests:

Prenatal /Preconception Genetic Testing of the following identified inherited disorders are considered medically necessary when the following criteria are met:

1. **Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutation**:
   American College of Medical Genetics (ACMG) 25-mutation panel for CFTR is medically necessary for **ANY** of the following indications:
   a. Couples seeking prenatal care; **OR**
   b. Couples who are planning a pregnancy; **OR**
   c. Persons with a family history of cystic fibrosis; **OR**
   d. Persons with a first degree relative identified as a cystic fibrosis carrier; **OR**
   e. Reproductive partners of persons with cystic fibrosis.

   Note: CFTR full gene sequencing will only be covered if the patient's partner is positive for a deleterious CFTR mutation and the patient has a family history of CF with a negative 25-mutation CFTR panel.

2. **Ashkenazi Jewish (see DEFINITIONS) descent carrier screening**:
   Carrier screening those of Ashkenazi Jewish descent for common inherited conditions is medically necessary using a limited gene panel (<20 genes).

3. **Duchenne Muscular Dystrophy and Becker Muscular Dystrophy**
   Carrier screening for Duchenne Muscular Dystrophy or Becker Muscular Dystrophy sequencing using DMD gene testing is medically necessary when the individual is an asymptomatic female and has an affected blood relative (test for known mutation if possible).

4. **Fragile X:**
   Screening for Fragile X gene mutation is medically necessary when **BOTH** of the following criteria are met:
   a. Individuals planning a pregnancy who have **EITHER** of the following:
      i. A family history of Fragile X related disorders, **OR**
      ii. A family history of intellectual disability suggestive of Fragile X syndrome;
   b. The results of the test will affect the patient's reproductive decisions.

5. Hemoglobinopathies and Thalassemia

Carrier screening for hemoglobinopathies and thalassemia using a DNA-based carrier screen for sickle cell, alpha or beta thalassemia common variants, and beta thalassemia full gene sequencing is medically necessary when **ONE** of the following is met:

a. Both members of a couple are known carriers based on initial testing with CBC and hemoglobin electrophoresis **AND** the couple plans to have prenatal testing with CVS or amniocentesis for the hemoglobinopathy.
b. Person is known carrier based on initial testing with CBC and hemoglobin electrophoresis **AND** the reproductive partner is unavailable for testing **AND** person plans to have prenatal testing with CVS or amniocentesis for the hemoglobinopathy.
c. Persons who are suspected/possible carriers based on initial testing with CBC and hemoglobin electrophoresis, but that testing is unclear, and DNA testing is necessary to determine carrier status.

6. Huntington Disease

Screening for CAD repeat length mutation in the HTT gene for Huntington disease is considered medically necessary in asymptomatic patients when **BOTH** of the following criteria are met:

a. Individuals planning a pregnancy who have **ONE** of the following:
    ii. A first-degree blood relative - parent or sibling - with documented Huntington disease, **OR**
    iii. Have a second-degree blood relative – grandparent, aunt or uncle - with documented Huntington disease and the first-degree blood relative status is unknown.
b. The results of the test will affect the patient's reproductive decisions.

7. Marfan syndrome

Screening for a FNB1 gene mutation by sequence analysis followed by deletion and duplication analysis testing if negative for Marfan syndrome is considered medically necessary when **BOTH** of the following criteria are met:

a. Individuals planning a pregnancy who have **ONE** of the following:
    i. Suspected, but not confirmed, Marfan syndrome in patients with BOTH of the following:
        1. Absence of a confirmed family history of Marfan syndrome, **AND**
        2. Diagnosis of ectopia lentis with any aortic dilation or significant aortic dilation or dissection; **OR**
    ii. Asymptomatic individual with a first-degree blood relative with Marfan Syndrome and a known genetic mutation (test for known familial mutation);
b. The results of the test will affect the patient's reproductive decisions.

8. Osteogenesis Imperfecta

Screening for COL1A1 or COL1A2 sequence mutations in couples in which one of the couple has osteogenesis imperfecta caused by COL1A1 or COL1A2 mutations.

9. **Spinal Muscular Atrophy (SMA):**

Screening for spinal muscle atrophy by testing for deletions in *SMN1* and *SMN2*, i.e., SMA Carrier testing, is medically necessary for any of the following conditions:

a. Couples seeking prenatal care; **OR**
b. Couples who are planning a pregnancy; **OR**
c. Persons with a family history of spinal muscle atrophy; **OR**
d. Persons with a first degree relative identified as a spinal muscle atrophy carrier; **OR**

e. Reproductive partners of persons with spinal muscle atrophy.

NOTE: Molecular testing reports of the affected individual and carrier testing of the related parent should be reviewed, if possible, before testing the patient. If the reports are not available, SMA Carrier testing should be recommended for the low-risk partner.

10. Tay-Sachs Disease:

Screening for Tay-Sachs disease is medical necessary for any of the following conditions:

a. Persons with a family history consistent with Tay-Sachs disease; **OR**
b. Persons of Ashkenazi Jewish, French-Canadian or Cajun descent or the reproductive partner of such a person; **OR**
c. Partner of a known carrier of Tay-Sachs disease.

11. Evaluation of a fetus

Cytogenetic/chromosomal microarray testing (a.k.a., comparative genomic hybridization (CGH)) of amniotic fluid, placenta or products of conception for evaluation of a fetus is medically necessary for prenatal or postnatal testing in the evaluation of **ANY** of the following:

a. In patients with a fetus with one or more structural abnormalities identified on ultrasound or magnetic resonance imaging (MRI) examination; **OR**
b. In patients with an intrauterine fetal demise (at or after 20 weeks pregnancy), stillbirth, or infant born alive and died after birth from indeterminate causes; **OR**
c. In patients with recurrent pregnancy loss and a clinically confirmed pregnancy loss in the first trimester.

D. Indications Considered Experimental and Investigational: (*Not all inclusive)*

1. Requests for whole gene panel testing.
2. Routine screening for SMA in the general public.
3. Prenatal testing for autism spectrum disorder.
4. Chromosomal microarray analysis testing to screen for prenatal gene mutation in fetuses without structural abnormalities.
5. Whole exome or whole genome sequencing for prenatal diagnosis.

HCPCS/CPT CODES:

81220	CFTR
81329	SMA Carrier Testing
81243	Fragile X
81260	Familial Dysautonomia
81200	Canavan Disease
81255	Tay-Sachs Disease
81271	Huntington Disease Mutation Analysis

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