# Clinical Policy: Genetic Testing

Reference Number: TX.CP.MP.531
Last Review Date: 11/20

Coding Implications
Revision Log

**See Important Reminder at the end of this policy for important regulatory and legal information.**

## Description

Genetic testing identifies changes in chromosomes, genes, or proteins that may cause illness or disease. The results of a genetic test can confirm or rule out a suspected genetic condition.

Carrier screening is performed to identify members at risk of having offspring with a genetic disease. Carriers are usually not at risk of developing the disease, but have a risk of passing a pathogenic gene mutation to their offspring.

This policy applies to the following products: STAR, STAR+PLUS, STAR Health, STAR Kids, CHIP, and CHIP Perinate.

## Policy/Criteria

**I.** It is the policy of Superior HealthPlan that genetic testing is **medically necessary** when **all** of the following criteria are met:

A. The member displays clinical features, or is at direct risk of inheriting the mutation in question, *and*

B. The test results will be used to develop a clinically useful approach or course of treatment, or to cease unnecessary monitoring or treatments for the individual being tested. Clinically useful test results allow providers to do at least one of the following:
   1. Inform interventions that could prevent or delay disease onset,
   2. Detect disease at an earlier stage when treatment is more effective,
   3. Manage the treatable progression of an established disease,
   4. Treat current symptoms significantly affecting a member's health,
   5. Guide decision making for the member's current or planned pregnancy; *and*

C. The genetic disorder could not be diagnosed through completion of conventional diagnostic studies, pedigree analysis and genetic counseling consistent with the community standards; *and*

D. The member has not previously undergone genetic testing for the disorder, unless significant changes in testing technology or treatments indicate that test results or outcomes may change due to repeat testing.

- *Note: When using testing panels, including but not limited to, multiple genes or multiple conditions, and in cases where a tiered approach/method is clinically available, testing would be covered ONLY for the number of genes or tests deemed medically necessary to establish a diagnosis.*

**II.** It is the policy of Superior HealthPlan that all other requests for genetic testing not meeting the above stated criteria, including direct-to-consumer testing and genetic banking/DNA storage, are considered **not medically necessary**.

**III.** It is the policy of Superior HealthPlan that Cystic Fibrosis (CF) carrier screening (CPT® code 81220) is **medically necessary** for women who are pregnant meeting the following criteria:
A. No prior CF screening results are available, *and*
B. Pregnancy ≤ 22 weeks gestation, *and*
C. Underwent pretest counseling.

- *Note: CF carrier screening anytime other than during pregnancy and for requests for CPT® codes 81221 – 81224 during pregnancy is **medically necessary** when meeting the most current version of the relevant nationally recognized decision support tools.*

**IV.** It is the policy of Superior HealthPlan that Spinal Muscular Atrophy (SMA) carrier screening (CPT® code 81329) is **medically necessary** for women who are pregnant meeting the following criteria:
A. No prior SMA screening results are available, *and*
B. Pregnancy ≤ 22 weeks gestation, *and*
C. Underwent pretest counseling.

- *Note: SMA carrier screening anytime other than during pregnancy is **medically necessary** when meeting the most current version of the relevant nationally recognized decision support tools*.

**V.** It is the policy of Superior HealthPlan that Cell-Free Fetal DNA testing or noninvasive prenatal testing (CPT® code 81420 and 81507) is **medically necessary** for members meeting **all** of the following criteria:
A. Underwent pretest counseling, *and*
B. Current pregnancy is a singleton or twin gestation; *and*
C. At least 10 weeks gestation at the time the blood was drawn

**VI.** Non-Covered Benefits:
A. Whole exome sequencing (CPT 81415, 81416, 81417).
B. Mitochondrial genome sequencing (CPT 81440, 81460, 81465).
C. Genetic testing of members when performed for the benefit of family members is not covered by Superior HealthPlan.
D. Measurement of cell-free DNA for screening for micro-deletion syndromes, micro-duplication syndromes, and rare autosomal trisomies other than 13, 18 and 21 (CPT 81422) has not been clinically validated and is not covered.

**Background**

*Carrier Screening for Cystic Fibrosis:*

- Cystic Fibrosis (CF or mucoviscidosis) is an autosomal recessive genetic disorder that causes the body to make thick, sticky mucus that clogs the lungs and other organs such as the kidneys and digestive system. It has a prevalence of 1 in 2500 to 3300 live births or Caucasians of Northern European heritage. More than 10 million Americans are carriers of a defective CF gene and show no symptoms of the disease. CF is a recessive disorder;

therefore an abnormal gene must be inherited from both parents for the disease to manifest. Carrier testing can provide an early indication of the chances for a fetus to develop CF.

- If the maternal screening test is positive, the father of the fetus must also be tested to determine if he is a carrier. If negative, no further testing is needed. If both the mother and father test positive for a defective CF gene, the fetus will most likely be a carrier like the parents, however there is a 25% chance the fetus will have CF. If both parents are carriers, chorionic villus sampling or amniocentesis can be performed to determine if the fetus is affected.

*Carrier Screening for Spinal Muscular Atrophy:*

- Spinal Muscular Atrophy is an autosomal recessive disorder that causes degeneration of spinal cord motor neurons which leads to atrophy of the skeletal muscles. People with SMA experience overall weakness in the voluntary muscles, as well as trouble breathing and swallowing. Approximately 6 million Americans are carriers for SMA, with 1 in 6,000 to 1 in 10,000 live births being diagnosed with SMA. SMA is the leading genetic cause of infant mortality. Most cases of SMA result from both parents being carriers, but approximately 2% of cases are the result of a new gene mutation.

*Cell-Free Fetal DNA Testing or Noninvasive Prenatal Testing:*

- Cell-Free Fetal DNA testing is a screening test of the woman's blood taken after 10 weeks of pregnancy. It measures the relative amount of free fetal DNA and indicates if the fetus is at increased risk of having Down syndrome (trisomy 21), Edward syndrome (trisomy 18) and Patau syndrome (trisomy 13).

**Coding Implications**

This clinical policy references Current Procedural Terminology (CPT®). CPT® is a registered trademark of the American Medical Association. All CPT codes and descriptions are copyrighted 2020, American Medical Association. All rights reserved. CPT codes and CPT descriptions are from the current manuals and those included herein are not intended to be all-inclusive and are included for informational purposes only. Codes referenced in this clinical policy are for informational purposes only. Inclusion or exclusion of any codes does not guarantee coverage. Providers should reference the most up-to-date sources of professional coding guidance prior to the submission of claims for reimbursement of covered services.

| CPT® Codes | Description |
|---|---|
| 81220 | CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; common variants (e.g., ACMG/ACOG guidelines) |
| 81329 | Spinal muscular atrophy (SMA) carrier screening |
| 81420 | Fetal chromosomal aneuploidy (e.g., trisomy 21, monosomy X) |

| CPT® Codes | Description |
|---|---|
| 81422 | Fetal chromosomal microdeletion(s) genomic sequence analysis (e.g., DiGeorge syndrome, Cri-du-chat syndrome), circulating cell-free fetal DNA in maternal blood |
| 81507 | Fetal aneuploidy [trisomy 21, 18 and 13] DNA sequence analysis |
| 81415 | Exome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis |
| 81416 | Exome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator exome (e.g., parents, siblings) |
| 81417 | Exome (e.g., unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained exome sequence (e.g., updated knowledge or unrelated condition/syndrome) |
| 81440 | Nuclear encoded mitochondrial genes (e.g., neurologic or myopathic phenotypes), genomic sequence panel, must include analysis of at least 100 genes |
| 81460 | Whole mitochondrial genome (e.g., Leigh syndrome, mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes [MELAS], myoclonic epilepsy with ragged-red fibers [MERFF], neuropathy, ataxia, and retinitis pigmentosa [NARP], Leber hereditary optic neuropathy) |
| 81465 | Whole mitochondrial genome large deletion analysis panel (e.g., Kearns-Sayre syndrome, chronic progressive external opthalmoplegia), including heteroplasmy detection, if performed |

| HCPCS Codes | Description |
|---|---|
| N/A | |

**ICD-10-CM Diagnosis Codes that Support Coverage Criteria**

| ICD-10-CM Code | Description |
|---|---|
| N/A | |

| Reviews, Revisions, and Approvals | Date | Approval Date |
|---|---|---|
| Policy Created | 07/13 | 07/13 |
| Updated work process and reference. | 10/13 | 10/13 |
| Rephrase policy statement to “Diagnostic tests to check for genetic abnormalities must be performed only if the result of the test will aid in the diagnosis and/or prognosis **AND** influence the medical or surgical management of the member or family member at risk.” Added Cell-Free Fetal DNA Testing or Noninvasive Prenatal Testing criteria. Updated references. | 01/13 | 01/13 |

| Reviews, Revisions, and Approvals | Date | Approval Date |
|---|---|---|
| Updated Cell-Free Fetal DNA Testing definition and criteria. Updated references. Updated signatories. | 04/14 | 04/14 |
| Updated work process to change requests for PA will be accepted up to 5 business days after specimen collection instead of 7 days after specimen collection and reviewed for medical necessity based on the above stated criteria. Attached the Genetic Testing Reminder. | 08/14 | 08/14 |
| Updated general genetic testing medically necessity criteria to include members displaying clinical features or is at direct risk or inheriting the mutation in question, interpretation of test and genetic disorder could not be diagnosed through completion of conventional diagnostic studies based on CP.MP.89. Removed work process and imbedded under attachment. Updated References and Signatories. | 01/15 | 01/15 |
| Updated definition of genetic testing. Removal of work process imbedded under attachment. Added non-covered benefits to include whole exome sequencing and genetic testing of members when performed for the benefit of family members not covered by Superior HealthPlan. Changed baby to fetus. Added CPT codes 81420 and 81507. Added CHIP Perinate to Products. Updated references and signatories. | 01/16 | 01/16 |
| Added STAR Kids to product type. Added CPT code 81422 and description. Added measurement of cell-free DNA for screening for micro-deletion syndromes, micro-duplication syndromes, and rare autosomal trisomies *other than* 13, 18 and 21 (CPT 81422) has not been clinically validated and is not covered. Updated references and signatories. | 01/17 | 01/17 |
| Changed estimated gestational age to 22 weeks, added criteria for SMA carrier screening. Removed pharmacogenetic verbiage. | 07/17 | 07/17 |
| Policy updated to concur with intended 2013 change. | 09/17 | 09/17 |
| Removed requirement for increased risk of aneuploidy to align with ACOG recommendations that any patients may choose cell-free DNA as a screening strategy regardless of her risk status. | 02/18 | 02/18 |
| Annual revision. Policy amended to align with HHSC decision to not cover the newly-issued CPT code for SMA carrier screening. Added requests for SMA carrier screening for Medicaid members under age 21 should be referred to a medical director for a medical necessity review. Updated references and signatories. | 02/19 | 02/19 |
| Updated to new template from TX.UM.10.31 (TX.CP.MP.531 nomenclature implementation 09/14/19). | 07/19 | 07/19 |
| Policy amended to align with HHSC decision to cover CPT code 81329 for SMA carrier screening. | 11/19 | 11/19 |
| Annual review. Added criteria to Section V. regarding Cell-Free Fetal DNA testing or noninvasive prenatal testing that members current pregnancy is a | 10/20 | 10/20 |

| Reviews, Revisions, and Approvals | Date | Approval Date |
|---|---|---|
| singleton or twin gestation; and at least 10 weeks gestation at the time the blood was drawn. For clarification added whole exome sequencing CPT codes 81415, 81416, 81417 to section VI. Non-Covered Benefits A. Added criterion B. Mitochondrial genome sequencing (CPT 81440, 81460, 81465) to section VI. Non-Covered Benefits. Updated references. | | |

**References**

1. CP.MP.36 Coverage of Experimental Technologies
2. CP.MP.83 Cystic Fibrosis Carrier Screening.
3. CP.MP.84 Cell-Free Fetal DNA Testing
4. Cystic Fibrosis Foundation. CF Genetics. Accessed online at: http://www.cff.org/AboutCF/Testing/Genetics/.
5. InterQual (IQ) 2018. Molecular Diagnostics Criteria.
6. The American College of Obstetricians and Gynecologists. (ACOG) Update on carrier screening for cystic fibrosis. Committee Opinion, Committee on Genetics, No. 486, April 2011.
7. The American College of Obstetricians and Gynecologists. Frequently asked questions FAQ171 Pregnancy. Accessed online at: https://www.acog.org/womens-health/faqs/cystic-fibrosis-prenatal-screening-and-diagnosis.
8. The Leeds Method of Management. April, 2008. Pregnancy and cystic fibrosis [online]. Leeds Regional Adult and Pediatric Cystic Fibrosis Units, St James's University Hospital, Leeds, UK. Available at http://www.cysticfibrosismedicine.com. Accessed June 3, 2013.
9. TX.UM.05 Timeliness of UM Decisions and Notifications.
10. Wenstorm MD, Katherine D., Wilkins-Haug MD, PhD, Louise, Barrs, MD, Vanessa A. UpToDate April 2013. Cystic Fibrosis: Prenatal Genetic Screening. Available at http://www.uptodate/com. Accessed June 3, 2013.
11. TX.UM.10.35 Physician's Peer to Peer Policy
12. CP.MP.89 Genetic Testing
13. Texas Medicaid Provider Manual, Gynecological, Obstetrics, and Family Planning Title XIX Services Handbook, Section 5 "Noninvasive Prenatal Testing (NIPT)," Sub-section 5.5, "Non-Covered Services." October 2020.
14. Texas Health and Human Services 2019 Healthcare Common Procedure Coding System (HCPCS) Special Bulletin, No. 15. December 31, 2018.
15. The American College of Obstetrics and Gynecologists Practice Bulletin, Screening for Fetal Chromosomal Abnormalities, No. 226. American College of Obstetrics and Gynecologists. October 2020.

**Important Reminder**

This clinical policy has been developed by appropriately experienced and licensed health care professionals based on a review and consideration of currently available generally accepted standards of medical practice; peer-reviewed medical literature; government agency/program

approval status; evidence-based guidelines and positions of leading national health professional organizations; views of physicians practicing in relevant clinical areas affected by this clinical policy; and other available clinical information. The Health Plan makes no representations and accepts no liability with respect to the content of any external information used or relied upon in developing this clinical policy. This clinical policy is consistent with standards of medical practice current at the time that this clinical policy was approved. "Health Plan" means a health plan that has adopted this clinical policy and that is operated or administered, in whole or in part, by Centene Management Company, LLC, or any of such health plan's affiliates, as applicable.

The purpose of this clinical policy is to provide a guide to medical necessity, which is a component of the guidelines used to assist in making coverage decisions and administering benefits. It does not constitute a contract or guarantee regarding payment or results. Coverage decisions and the administration of benefits are subject to all terms, conditions, exclusions and limitations of the coverage documents (e.g., evidence of coverage, certificate of coverage, policy, contract of insurance, etc.), as well as to state and federal requirements and applicable Health Plan-level administrative policies and procedures.

This clinical policy is effective as of the date determined by the Health Plan. The date of posting may not be the effective date of this clinical policy. This clinical policy may be subject to applicable legal and regulatory requirements relating to provider notification. If there is a discrepancy between the effective date of this clinical policy and any applicable legal or regulatory requirement, the requirements of law and regulation shall govern. The Health Plan retains the right to change, amend or withdraw this clinical policy, and additional clinical policies may be developed and adopted as needed, at any time.

This clinical policy does not constitute medical advice, medical treatment or medical care. It is not intended to dictate to providers how to practice medicine. Providers are expected to exercise professional medical judgment in providing the most appropriate care, and are solely responsible for the medical advice and treatment of members. This clinical policy is not intended to recommend treatment for members. Members should consult with their treating physician in connection with diagnosis and treatment decisions.

Providers referred to in this clinical policy are independent contractors who exercise independent judgment and over whom the Health Plan has no control or right of control. Providers are not agents or employees of the Health Plan.

**Note: For Medicaid members**, when state Medicaid coverage provisions conflict with the coverage provisions in this clinical policy, state Medicaid coverage provisions take precedence. Please refer to the state Medicaid manual for any coverage provisions pertaining to this clinical policy.

**Note: For Medicare members,** to ensure consistency with the Medicare National Coverage Determinations (NCD) and Local Coverage Determinations (LCD), all applicable NCDs, LCDs, and Medicare Coverage Articles should be reviewed prior to applying the criteria set forth in this clinical policy. Refer to the CMS website at http://www.cms.gov for additional information.