

Clinical Policy: Carrier Screening in Pregnancy

Reference Number: PA.CP.MP.83

Effective Date: 01/18

Last Review Date: 07/17

[Coding Implications](#)

[Revision Log](#)

Description

This policy outlines medical necessity criteria for cystic fibrosis (CF) and spinal muscular atrophy (SMA) carrier testing.

Policy/Criteria

- I. It is the policy of Pennsylvania Health and Wellness[®] (PHW) that CF carrier screening (CPT[®] code 81220) or SMA carrier screening (81401) is **medically necessary** for women who are pregnant and meet the following criteria:
 - A. No prior CF or SMA screening results are available, and
 - B. Pregnancy \leq 22 weeks gestation, and
 - C. Underwent pretest counseling.
- II. It is the policy of PHW that CF or SMA carrier screening anytime other than during pregnancy and for requests for CF screening CPT[®] codes 81221 – 81224 during pregnancy is **medically necessary** when meeting the most current version of the relevant nationally recognized decision support tools.

Authorization Protocols

Requests for prior authorization will be accepted up to 5 business days after specimen collection and reviewed for medical necessity based on the above stated criteria.

Background

CF is a genetic disorder that causes the body to make thick, sticky secretions that clog the lungs and other organs such as the digestive system. 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 in order for the child to develop CF. Carrier testing may provide an early indication as to whether a fetus might either be a carrier or might develop CF.

SMA 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.

For either CF or SMA, if the maternal screening test is positive, the father of the baby 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 gene, the infant will most likely be a carrier like the parents (50% chance). However, there is a 25% chance that the infant will have the disease. If both parents are carriers, chorionic villus sampling or amniocentesis can be performed to see whether the fetus has the disease. Since these are both invasive procedures that carry a slight

CLINICAL POLICY
Carrier Screening in Pregnancy



risk to the fetus, further testing should only be performed if and when the course of the pregnancy will be altered based on results of the testing.

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 2017, 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 (eg. Cystic fibrosis) gene analysis; common variants (eg. ACMG/ACOG guidelines)
81221	known family variants
81222	Duplication/deletion variants
81223	Full gene sequence
81224	Intron 8 poly-T analysis (eg, male infertility)
81401	Molecular pathology procedure level 2 (used for SMA carrier testing)

Reviews, Revisions, and Approvals	Date	Approval Date

References

1. The American College of Obstetricians and Gynecologists. Carrier screening for genetic conditions. Committee Opinion, Committee on Genetics, No 691, March 2017.
2. The American College of Obstetricians and Gynecologists. Frequently asked questions FAQ171 Pregnancy. Accessed online at: <http://www.acog.org/~media/For%20Patients/faq171.pdf?dmc=1&ts=20130624T1134297985>.
3. Cystic Fibrosis Foundation. CF Genetics. Accessed online at: <http://www.cff.org/AboutCF/Testing/Genetics/>.
4. Spinal Muscular Atrophy Foundation. Overview. Accessed online at: <http://www.smafoundation.org/about-sma/>