Clinical Policy: Carrier Screening in Pregnancy

Reference Number: PA.CP.MP.83
Effective Date: 01/18
Last Review Date: 2/26/2021

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 ≤ 23 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 10 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 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

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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 reviewed	06.30.18	
Updated authorization protocol to allow for 10 business days retro review instead of 5. For clarification, revised I.B to state Pregnancy < 23 weeks gestation rather than < 22 weeks gestation. References reviewed and updated.	10/19	12/20/2019
Annual review completed. Codes reviewed. References reviewed and updated. Specialist reviewed.	2/26/2021	

References

1. The American College of Obstetricians and Gynecologists. Carrier screening for genetic conditions. Committee Opinion, Committee on Genetics, No 691, March 2017. Reaffirmed 2019.
2. The American College of Obstetricians and Gynecologists. Frequently asked questions FAQ171 Pregnancy. Cystic Fibrosis: Prenatal Screening and Diagnosis. June 2017. 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/
5. The American College of Obstetricians and Gynecologists. Frequently asked questions FAQ179. Pregnancy. Carrier Screening. Dec 2018. Accessed at: https://www.acog.org/Patients/FAQs/Carrier-Screening
6. The American College of Obstetricians and Gynecologists. Frequently asked questions FAQ197. Pregnancy. Carrier Screening for Spinal Muscular Atrophy. Oct 2018. Accessed at: https://www.acog.org/Patients/FAQs/Carrier-Screening-for-Spinal-Muscular-Atrophy
7. The American College of Obstetricians and Gynecologists. Carrier Screening in the Age of Genomic Medicine. No.690. March 2017. Reaffirmed 2019. Accessed April 26, 2020.