



Guideline Change: Carrier Testing for Spinal Muscular Atrophy

In March 2017, The American College of Obstetricians and Gynecologists (ACOG) published Committee Opinion #691 "Carrier Screening for Genetic Conditions."¹ While providing guidance for routine carrier testing, the Committee Opinion expanded the recommendation for spinal muscular atrophy (SMA) carrier testing from only those individuals with a family history of the disease to any woman who is pregnant or considering pregnancy.

eviCore's Clinical Policy team and Medical Advisory Board reviewed this Committee Opinion along with the existing American College of Medical Genetics and Genomics (ACMG) Position Statement on expanded carrier screening.² As part of the evaluation process, our team took into account the impact ACOG's Committee Opinion could have on ordering providers and our health plan clients, the SMA carrier frequency of approximately 1 in 40 to 1 in 60 in most populations,¹ the potentially lethal nature of the condition, and the recently announced availability of an FDA-approved treatment for some cases of SMA.³

As a result, eviCore has concluded that effective July 3, 2017 we will change our recommended coverage determination for SMA carrier screening to include any woman who is pregnant or considering pregnancy. We are in the process of updating our SMA and Expanded Carrier Screening Panels policies to reflect this coverage revision. The draft policy includes the following criteria for carrier screening:

Carrier Screening (SMN1/SMN 2 Dosage Analysis):

- Be of reproductive age, and
- Have potential and intention to reproduce, or
- Have a reproductive partner who is a carrier of SMA, or
- Have a reproductive partner with SMA

Updated policies will be sent this fall for your review as part of our bi-annual policy update. Please note, however, that our clinical review practices can implement this coverage change even before the policies are officially published.

Given the expanded population who will be eligible for testing, this change will likely increase your molecular spend. Please keep the impact of this expanded population in mind when negotiating contracts with laboratories that perform SMA carrier screening, as there are many diagnostic companies offering such testing and the increased volume should promote more competitive rates. SMA carrier screening is also often included in expanded carrier screening panels. Coding and billing guidance will be added to the applicable policies in order to discourage inappropriate code stacking for expanded carrier tests.

SMA testing should only be performed once per lifetime. SMA is currently billed using non-specific Tier 2 CPT code 81401. Since many other tests may be billed with the same code, monitoring inappropriate re-testing will be challenging. Our hope is that SMA and/or expanded carrier screening will have a test-

specific code assigned by the AMA in the near future, but we have no information to suggest such code additions are imminent.

As you know, changes occur frequently in the fast-paced world of genetics; eviCore will continue to keep you advised of these changes whenever significant events arise.

References:

1. ACOG Committee Opinion. Number 691, March 2017. Carrier Screening for Genetic Conditions Obstet Gynecol. 2017 Mar;129 (3):pe41-e55. Available at: <https://www.acog.org/-/media/Committee-Opinions/Committee-on-Genetics/co691.pdf?dmc=1>
2. Grody WW, Thompson BH, Gregg AR, et al. ACMG position statement on prenatal/preconception expanded carrier screening. Genet Med. 2013;15(6):482-3. Available at: https://www.acmg.net/docs/Prenatal_Preconception_Expanded_Carrier_Screening_Statement_GiM_June_2013.pdf
3. FDA News Release: <https://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm534611.htm>