Quest Advanced™
Women's Health

Spinal Muscular Atrophy (SMA)
with silent carrier screening

Increased detection for greater insights

Spinal muscular atrophy (SMA) is a progressive, neuromuscular, genetic disease that affects motor nerve cells in the spinal cord and is a leading cause of infant mortality.[1] Silent carriers have 2 normal copies of the *SMN1* gene on 1 chromosome and a deleted *SMN1* gene on the other chromosome.

Quest Diagnostics SMA with silent carrier screening:

- Detects the specific variant that helps to identify silent carriers
- Delivers timely, actionable results so you and your patients can make more informed decisions

Quest Diagnostics is a **pioneer in SMA screening,** bringing the first commercial test to market and continuing to innovate to help **minimize the uncertainty that arises** from identifying carriers of extremely rare disorders or variants of unknown significance.

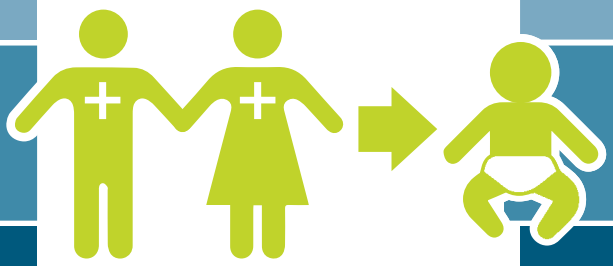
SMA is relatively common

Even if a couple already has a healthy child or children and no family history of SMA, 1 or both may still be a carrier of the disease. Carriers generally do not show signs and symptoms of SMA but could be at risk to have a child affected with the condition.

1 in 50 people in the US are carriers[2]

Family history a couple may still be carriers even if they already have a healthy child and no family history

Age is irrelevant, unlike with some other genetic disorders



If both parents are carriers...

25% chance any child will have SMA[3]

50% chance any child will not have the disease, but will be a carrier[3]

25% chance any child will not have the disease and will not be a carrier[4]

There is hope with early detection

SMA with silent carrier screening detection helps you and your patients make informed decisions. There is a 50% chance a silent carrier will pass the abnormal (missing *SMN1*) chromosome to the fetus,[4] so it's important that couples understand their SMA carrier status. New treatment options have the potential to deliver improved quality of life for babies diagnosed with SMA.[5]

Early screening is recommended by leading health organizations

ORGANIZATION	GUIDELINES
American College of Obsetricians and Gynecologists (ACOG)	Recommend screening for SMA be offered to all women who are considering pregnancy or are currently pregnant[5]
American College of Medical Genetics (ACMG)	Recommends that carrier screening be offered to all couples, before conception or early in pregnancy, regardless of race or ethnicity[6]

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Genetic insights that help you and your patients make informed decisions

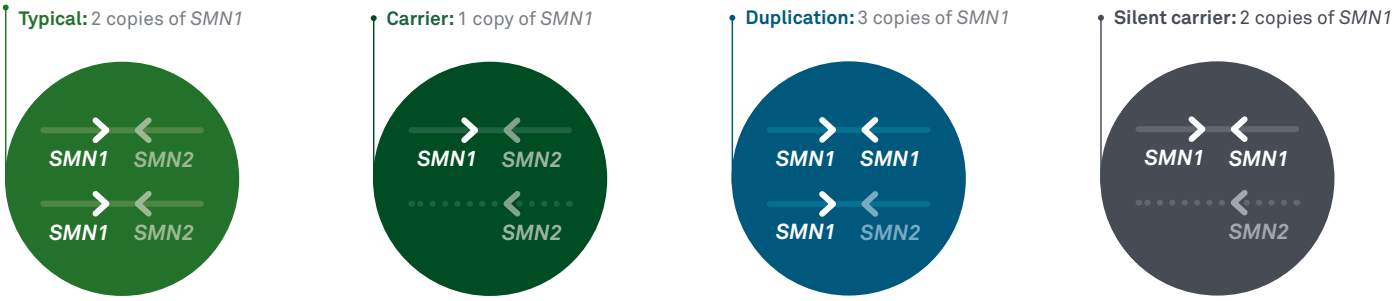
Typically, a higher number of *SMN2* gene copies is associated with a less severe presentation of SMA in affected individuals[4]

Assessing the *SMN2* copy number in carriers may provide additional information for genetic counseling in at-risk couples

In the case of a positive result, partner carrier screening is available and easy to order

Schematic of *SMN1* and *SMN2* gene configuration

SMA is caused by mutations in the *SMN1* gene generally involving its deletion or gene conversion with the tightly linked *SMN2* gene. While conventional screening for SMA typically determines total *SMN1* copy number, it does not include any information about how those copies are arranged, which limits the ability to identify silent (2+0) carriers.



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Supporting your **patients and your practice**

Quest Diagnostics is committed to helping you ensure the healthiest possible outcomes for your patients. With our broad range of tests, tools, and educational materials, we can help you manage their care more effectively.

Accessible testing

- **In-network with most major health plans,** providing coverage to over 90% of all commercially insured patients[7]
- **Financial assistance** for those who qualify

Simplified processes

- **Quanum® Lab Services Manager—** order tests and supplies, access results, or track an order
- **MyQuest™ patient mobile app and portal—**test results anytime, anywhere, plus online appointment scheduling

A leader in genetic testing

- **Comprehensive testing solutions for the complete pregnancy journey;** carrier screening and prenatal diagnosis that align with guidelines from leading health organizations
- **A team of experts** are available for consultation and results interpretation

Test ordering information

Test name	Test code	CPT code	Preferred specimen	Transport
SMA Carrier Screen	18041	81329	4 mL (2 mL minimum) whole blood collected in EDTA (lavender-top) tube	Room temperature. Do not refrigerate or freeze

For more information, contact your sales representative or contact one of our genetic counselors at **1.866.GENE.INFO (1.866.436.3463)** or **GeneInfo@QuestDiagnostics.com**

References
1. Spinal muscular atrophy: genetic concepts and carrier screening. ObG Project website. Accessed April 30, 2020. https://www.obgproject.com/2017/04/18/spinal-muscular-atrophy-genetic-concepts-carrier-screening/
2. About SMA: frequently asked questions. SMA Foundation website. Accessed May 12, 2020. https://smafoundation.org/about-sma/faq/
3. The American College of Obstetricians and Gynecologists (ACOG). Carrier screening for spinal muscular atrophy; frequently asked questions. Accessed May 11, 2020. https://www.acog.org/patient-resources/faqs/pregnancy/carrier-screening-for-spinal-muscular-atrophy
4. What is spinal muscular atrophy (SMA)? Together in SMA with Biogen website. Accessed May 13, 2020. https://www.togetherinsma.com/en_us/home/introduction-to-sma/smn1-gene.html
5. The American College of Obstetricians and Gynecologists (ACOG). Committee on Genetics. Committee Opinion No. 691: carrier screening for genetic conditions. Accessed July 8, 2020. https://www.acog.org/clinical/clinical-guidance/committee-opinion/articles/2017/03/carrier-screening-for-genetic-conditions
6. Prior TW; Professional Practice and Guidelines Committee. Carrier screening for spinal muscular atrophy. *Genet Med*. 2008;10(11):840-842. doi:10.1097/GIM.0b013e318188d069
7. Date on file at Quest Diagnostics, 2019.

The CPT® codes provided are based on American Medical Association guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.

Test codes may vary by location. Please contact your local laboratory for more information.

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