Quest Diagnostics™

# Spinal muscular atrophy (SMA) carrier screening

The insights you and your patients need to make more informed decisions

## Clear, actionable results to help determine an appropriate path

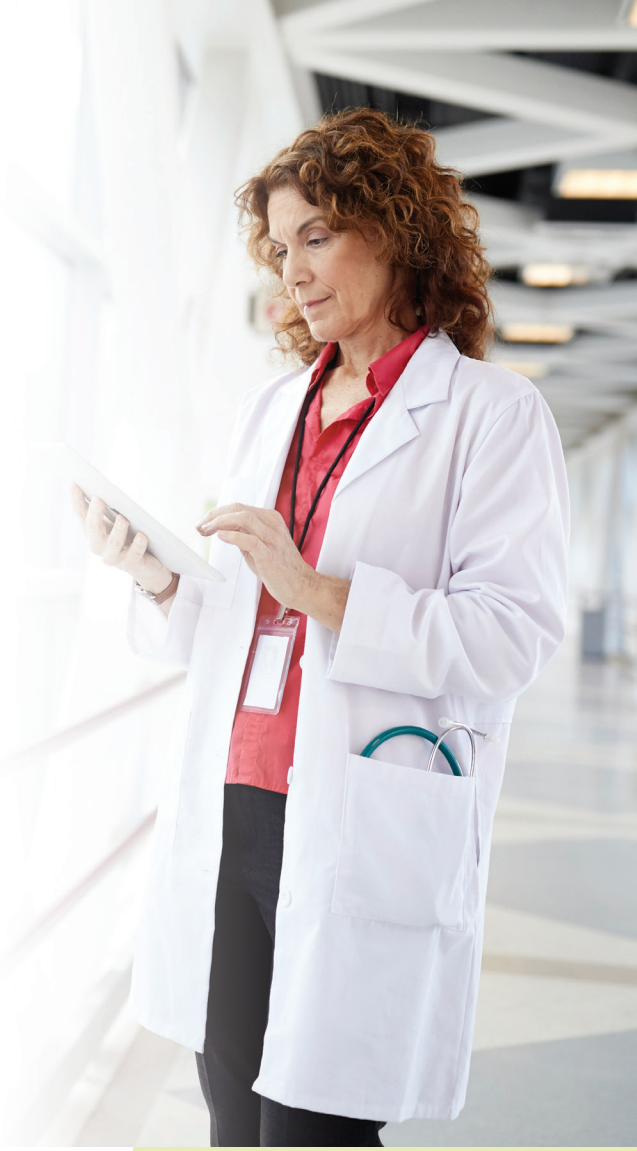
SMA is a progressive neuromuscular condition; it is the leading single-gene cause of death before the age of 2.[1]

Current guidelines, including those from the American College of Obstetricians and Gynecologists (ACOG), recommend SMA screening for women and couples who are considering conception, or who are early in pregnancy.[2,3]

SMA is relatively common—1 in 50 people in the US are carriers[4] and 1 in 4 children are born with SMA if both parents are carriers.[5]


Fortunately, it is easier to understand the risks of SMA with SMA Carrier Screen from Quest Diagnostics. Quest Diagnostics is the pioneering developer of SMA testing. In 1996, we offered the first commercial SMA Carrier Screen, and we've been evolving our test and processes ever since.

You can count on Quest Diagnostics for actionable insights into your patients' risk to have a child with SMA. We have the tests, support, and expertise you and your patients need to make more informed decisions.



## It's important to KNOW more

The Quest Diagnostics SMA Carrier Screen detects the copy number of survival motor neuron 1 *(SMN1)* and survival motor neuron 2 *(SMN2)* genes for a better understanding of carrier status. Typically, a higher number of *SMN2* gene copies is associated with a less severe presentation of SMA in affected individuals.[6,7] Assessing the *SMN2* copy number in carriers may provide additional information for genetic counseling in at-risk couples.[8]

## A clear picture to help you and your patients make informed decisions

The SMA Carrier Screen from Quest Diagnostics uses proven, advanced testing technologies to deliver timely, actionable results.

- Allele-specific, real-time polymerase chain reaction (rtPCR) technology
- *SMN1* copy number count for carrier status
- *SMN2* copy number count for further risk characterization for carriers
- Our easy-to-read results report includes *SMN1* and *SMN2* copy number counts and residual carrier risk by ethnicity
- In the case of a positive result, partner carrier screening* is available and easy to order

* Prenatal diagnosis for SMA is available for couples at 25% risk.

## Current guidelines and recommendations support SMA screening

SMA carrier screening is recommended by many of the world's leading medical organizations.

- Recently updated guidelines from ACOG recommend screening for SMA be offered to all women who are considering pregnancy or are currently pregnant[2]
- The American College of Medical Genetics recommends that carrier screening be offered to all couples, before conception or early in pregnancy, regardless of race or ethnicity[3]

## Trust Quest Diagnostics when you and your patients need to KNOW

Since 1996, Quest Diagnostics has been providing screening solutions for SMA, and was the first commercial laboratory to do so. When you have questions, rely on our experienced and trusted team of MDs, PhDs, genetic counselors, and geneticists for:

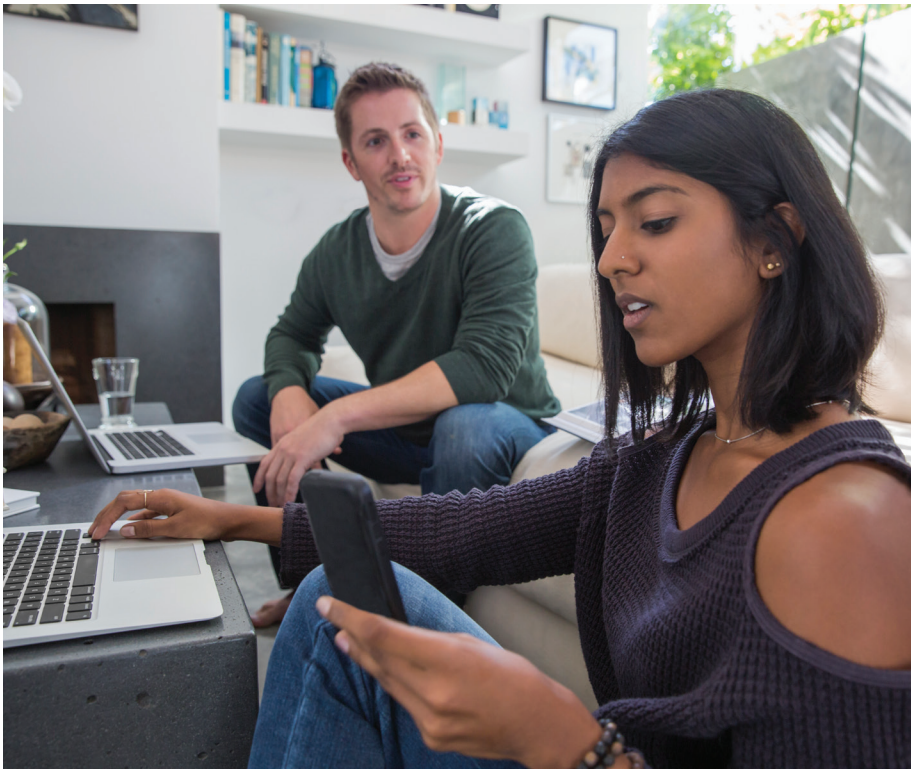
- Expert review and consultation to support decision-making when you need it
- One-on-one consultations and review of results interpretation

**Dedicated support at 1.866.GENE.INFO (1.866.436.3463), M-F, 8 AM-8 PM EST, or email GeneInfo@QuestDiagnostics.com**

## Balanced carrier screening tests and panels developed with you and your patient in mind

With the expansion of genetic information about extremely rare conditions, the choice of which tests to order has become harder for physicians and patients alike.

Quest Diagnostics carrier screening offerings strike the right balance. You can maximize the clinical insight for your patient while minimizing the uncertainty that arises from identifying carriers of extremely rare disorders or variants of unknown significance.



## New therapies for SMA offer potential treatments

In the past, a diagnosis offered little hope for patients and their families. However, new treatment options are available that have the potential to deliver improved quality of life for babies diagnosed with SMA, and even the possibility of a cure in some cases.[9]

With the potential for treating SMA, it's more important than ever that couples understand their SMA carrier status prior to, or early in pregnancy, so they can make informed decisions and plans.

# Trust Quest Diagnostics for all your testing needs

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.

## Support for physicians

- A clinically relevant carrier testing menu that aligns with guidelines from leading health organizations
- Proven expertise in genetic testing, built on the latest technology, and including more genetic tests than any other laboratory
- Comprehensive testing solutions for the whole pregnancy: carrier screening and prenatal diagnosis
- Quest Quanum™ Lab Services Manager—order tests and supplies, access results, or track an order

Call **1.866.GENE.INFO (1.866.436.3463)** to speak directly with a genetic counselor about test selection and results interpretation

## Support for patients

- Financial assistance for those who qualify
- In-network status with all major health plans, providing coverage to over 90% of all commercially insured patients[10]
- Educational content in multiple languages on a wide range of healthcare topics
- Tools that help patients make better decisions and stay engaged in their own healthcare
- MyQuest™ patient mobile app and portal—test results anytime, anywhere, plus online appointment scheduling

### Test ordering information

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

### Other carrier screening solutions from Quest Diagnostics

| Test name | Test code | CPT code* |
|---|---|---|
| Prenatal Carrier Screen (CF, Fragile X, SMA) | 90949, 93349 | 81220, 81243, 81329 |
| QHerit® Expanded Carrier Screen | 94372 | 81443 |
| Ashkenazi Jewish Panel (11 Tests) | 90891 | 81412 |
| CFvantage® Cystic Fibrosis Expanded Screen | 92068 | 81220 |
| XSense®, Fragile X with Reflex | 16313 | 81243 |

**TestDirectory.QuestDiagnostics.com**

*CPT codes provided are based on AMA 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.

For more information, contact your sales representative or contact a genetic counselor at **1.866.GENE.INFO (1.866.436.3463)** or GeneInfo@QuestDiagnostics.com.


References

1. Lally C, Jones C, Farwell W, Reyna SP, Cook SF, Flanders WD. Indirect estimation of the prevalence of spinal muscular atrophy Type I, II, and III in the United States. *Orphanet J Rare Dis*. 2017;12(1):175. doi: 10.1186/s13023-017-0724-z
2. Committee on Genetics. Committee Opinion No. 691: carrier screening for genetic conditions. *Obstet Gynecol*. 2017;129(3):e41-e55. doi: 10.1097/AOG.0000000000001952
3. Prior TW; for Professional Practice and Guidelines Committee. Carrier screening for spinal muscular atrophy. *Genet Med*. 2008;10(11):840-842. doi: 10.1097/GIM.0b013e318188d069
4. FAQ test: overview. SMA Foundation web site. http://www.smafoundation.org/faq-test-2/. Accessed May 16, 2019.
5. Muralidharan K, Wilson RB, Ogino S, Nagan N, Curtis C, Schrijver I. Population carrier screening for spinal muscular atrophya position statement of the association for molecular pathology. *J Mol Diagno*. 2011;13(1):3-6. doi: 10.1016/j.moldx.2010.11.012
6. Darras BT, Jones RH Jr, Ryan MM, De Vivo DC, eds. *Neuromuscular Disorders of Infancy, Childhood, and Adolescence: A Clinician's Approach*. 2nd ed. San Diego: Elsevier; 2015.
7. Fang P, Li L, Zeng J, et al. Molecular characterization and copy number of *SMN1*, *SMN2* and *NAIP* in Chinese patients with spinal muscular atrophy and unrelated healthy controls. *BMC Musculoskelet Disord*. 2015;16(1):11. doi: 10.1186/s12891-015-0457-x
8. Arnold WD, Kassar D, Kissel JT. Spinal muscular atrophy: diagnosis and management in a new therapeutic era. *Muscle Nerve*. 2015;51(2):157-167. doi: 10.1002/mus.24497
9. Drugs in development: SMA therapeutics pipeline. SMA Foundation web site. http://www.smafoundation.org/development/pipeline/. Accessed May 15, 2019.
10. Quest Diagnostics, data on file, 2019.


Image content used for illustrative purposes only. Persons depicted in the content are models.

QuestDiagnostics.com