LASTNAME, FIRSTNAME

Patient ID:

Specimen ID: **000-000-0000-0**

DOB: **mm/dd/yyyy**

Age: **00**

Sex: **Female**

Account Number: **00000000**

Ordering Physician:

labcorp

Date Collected: **mm/dd/yyyy**	Date Received: **mm/dd/yyyy**	Date Reported: **mm/dd/yyyy**	Date Entered: **mm/dd/yyyy**

Specimen Type: **Whole Blood**

Ethnicity: **Not Provided**

Indication: **Carrier Test / Screening**

Spinal Muscular Atrophy (SMA)

SAMPLE REPORT

Summary: NEGATIVE

Negative Results

Disorder *(Gene)*	Result	Interpretation
Spinal muscular atrophy *(SMN1)* NM_000344.3	NEGATIVE 2 copies of *SMN1*; c.*3+80T>G risk variant not present.	This result reduces, but does not eliminate, the risk to be a carrier. **Risk:** NOT at an increased risk for an affected pregnancy.

Recommendations

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of positive results, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Labcorp Genetic Counselors please visit https://womenshealth.labcorp.com/genetic-counseling or call (855) GC-CALLS (855-422-2557).

Additional Clinical Information

Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disorder with variable age at onset and severity, characterized by progressive degeneration of the lower motor neurons in the spinal cord and brain stem, leading to muscle weakness, and in its most common form, respiratory failure by age two. Complications of SMA may include poor weight gain, sleep difficulties, pneumonia, scoliosis, and joint deformities. In severely affected individuals, abnormal fetal ultrasound findings may include congenital joint contractures, polyhydramnios, and decreased fetal movement. (Korinthenberg, PMID:9307259). Treatment is supportive. Targeted therapies may be available for some individuals. Approximately 94% of affected individuals have 0 copies of the *SMN1* gene; in these individuals, an increase in the number of copies of the *SMN2* gene correlates with reduced disease severity (Feldkotter M, PMID:11791208). Individuals with one copy of the *SMN*1 gene are predicted to be carriers of SMA; those with two or more copies have a reduced carrier risk. For individuals with two copies of the *SMN1* gene, the presence or absence of the variant c.*3+80T>G correlates with an increased or decreased risk, respectively, of being a silent carrier (2+0).

Comments

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder(s) tested. Information about the disorder(s) tested is available at https://womenshealth.labcorp.com.

Methods/Limitations

Spinal muscular atrophy: The copy number of *SMN1* exon 7 is assessed relative to internal standard reference genes by quantitative polymerase chain reaction (qPCR). A mathematical algorithm calculates 0, 1, 2 and 3 copies with statistical confidence. In fetal specimens and specimens with 0 or 1 copies, the primer and probe binding sites are sequenced to rule out variants that could interfere with copy number analysis. *SMN2* copy number is assessed by digital droplet PCR analysis relative to an internal standard reference gene in samples with no copies of *SMN1*. For carrier screening, when two copies of *SMN1* are detected, allelic discrimination qPCR targeting c.*3+80T>G in *SMN1* is performed.

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants, or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships.

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Spinal Muscular Atrophy (SMA)

Information Table
Spinal muscular atrophy risk reductions for individuals with no family history

Population	Detection rate (Copy number + SNP)	Pre-test carrier risk	Post-test risk of being a carrier with 2 copies**		Post-test risk of being a carrier with 3 copies
			POSITIVE for the c.*3+80T>G SNP	NEGATIVE for the c.*3+80T>G SNP	
African American	90.3%	1 in 72	1 in 34	1 in 375	1 in 4200
Ashkenazi Jewish	92.8%	1 in 67	High risk	1 in 918	1 in 5400
Asian	93.6%	1 in 59	High risk	1 in 907	1 in 5600
Caucasian	95.0%	1 in 47	1 in 29	1 in 921	1 in 5600
Hispanic	92.6%	1 in 68	1 in 140	1 in 906	1 in 5400
Mixed or other ethnic background	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.				

** includes carriers who are silent carriers (2+0) and carriers with a pathogenic variant not detected in this assay
Feng. PMID 28125085; Luo. PMID 23788250; Sugarman. PMID 21811307

References
Deignan JL, Astbury C, Behlmann A *et al*. Addendum: Technical standards and guidelines for spinal muscular atrophy testing. *Genet Med* 23, 2462 (2021). [Addendum to PMID: 21673580]

Prior TW, Leach ME, Finanger E. Spinal Muscular Atrophy. 2000 Feb 24 (Updated 2020 Dec 30). In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. PMID: 20301526

Performing Labs

Component Type	Performed at	Laboratory Director
Technical component, processing	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG
Technical component, analysis	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG
Professional component	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG

For inquiries, the physician may contact the lab at 800-255-7357

This test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. It has not been cleared or approved by the Food and Drug Administration.

Esoterix Genetic Laboratories, LLC is a subsidiary of Laboratory Corporation of America Holdings, using the brand Labcorp.

Patient Details
LASTNAME, FIRSTNAME

Phone:
Date of Birth: **mm/dd/yyyy**
Age: **00**
Sex: **Female**
Patient ID:
Alternate Patient ID:

Physician Details
CLIENT NAME
CLIENT ADDRESS

Phone: **000000000**
Account Number: **00000000**
Physician ID:
NPI:

Specimen Details
Specimen ID: **00000000000**
Control ID:
Alternate Control Number:
Date Collected: **mm/dd/yyyy 0000 Local**
Date Received: **mm/dd/yyyy 1428 ET**
Date Entered: **mm/dd/yyyy 1157 ET**
Date Reported: **mm/dd/yyyy 0944 ET**

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