

LASTNAME, FIRSTNAME

DOB: mm/dd/yyyy

Account Number: 00000000



Patient ID:

Specimen ID: 000-000-0000-0

Age: 00

Ordering Physician:

Sex: Female

Date Collected: mm/dd/yyyy

Date Received: mm/dd/yyyy

Date Reported: mm/dd/yyyy

Date Entered: mm/dd/yyyy

Specimen Type: Chorionic Villi

Ethnicity: Not Provided

Indication: Prenatal Test / Family history - previous affected child

Spinal Muscular Atrophy (SMA), Fetal Analysis

Summary: ■ POSITIVE

SAMPLE REPORT

Variants Detected

Disorder (Gene)	Result	Interpretation
Spinal muscular atrophy (SMN1) NM_000344.3	POSITIVE: PREDICT AFFECTED SMN1 copy number: 0 SMN2 copy number: 3	This fetus has no copies of SMN1 and is therefore predicted to be affected with spinal muscular atrophy, a disease of variable age of onset and severity. An increase in the number of copies of the SMN2 gene correlates with reduced disease severity. Genetic counseling is recommended.

Comparison of maternal and fetal DNA markers indicates that maternal cell contamination is unlikely to have interfered with the reported fetal result (maternal specimen # 00000000000). The result obtained from a multiple gestation pregnancy depends on the successful sampling of the intended fetus.

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 SMN1 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

Maternal Cell Contamination Analysis: DNA is isolated and amplified by the polymerase chain reaction (PCR). Fifteen polymorphic markers from 14 chromosomes are analyzed by capillary gel electrophoresis and fluorescence detection. Markers analyzed include TPOX, D3S1358, FGA, D5S818, CSF1PO, D7S820, D8S1179, THO1, vWA, D13S317, Penta E, D16S539, D18S51, D21S11, and Penta D. The analytical sensitivity of the assay is approximately 10%; maternal cell contamination present at a lower percentage may not be detected.

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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Age: 00

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Sex: Female

Spinal Muscular Atrophy (SMA), Fetal Analysis

Information Table

Spinal muscular atrophy detection rates for prenatal testing

Population	Detection rate
African American	90.3%
Ashkenazi Jewish	92.8%
Asian	93.6%
Caucasian	95.0%
Hispanic	92.6%
Mixed or other ethnic background	For counseling purposes, consider using the ethnic background with the most conservative risk estimates.

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.

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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: 0000000000

Control ID:

Alternate Control Number:

Date Collected: mm/dd/yyyy 0000 Local

Date Received: mm/dd/yyyy 1428 ET

Date Entered: mm/dd/yyyy 1203 ET

Date Reported: mm/dd/yyyy 2002 ET

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