

SAMPLE REPORT



SMN1 Copy Number Analysis

Patient Name: LASTNAME, FIRSTNAME
Referring Physician: REF. PHYSICIAN NAME
Specimen #: 00000000-00
Patient #: 00000000
Client #: 000000
Case #: 00000000

CLIENT ADDRESS

DOB: MM/DD/YYYY
Sex: Female
Lab ID:
Hospital ID:
Specimen Type: Peripheral Blood
Date Collected: MM/DD/YYYY
Date Received: MM/DD/YYYY

Ethnicity: Ashkenazi Jewish
Indication: Screening

Disorder (Gene)	Results	Interpretation
Spinal muscular atrophy (SMN1)	POSITIVE	1 copy of SMN1. Predicted to be a carrier. Genetic counseling is recommended.

Genetic counseling services are available. To access Integrated Genetics Genetic Counselors please visit www.integratedgenetics.com/genetic-counseling or call (855) GC-CALLS (855-422-2557).

ADDITIONAL CLINICAL INFORMATION

Spinal muscular atrophy: 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, 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) (Luo, PMID 23788250; Feng, PMID 28125085). Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of these results, as well as recommendations for testing family members and, when applicable, this individual's partner. If this individual's reproductive partner is also a carrier, the risk for an affected fetus is 25%.

METHOD / 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. When no copies of SMN1 are detected, the primer and probe binding sites are sequenced to rule out variants that could interfere with copy number analysis and SMN2 copy number is assessed by digital droplet PCR analysis relative to an internal standard reference gene. For carrier screening, when two copies of SMN1 are detected, allelic discrimination qPCR targeting c.*3+80T>G in SMN1 is performed.

Limitations: False positive or false negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships.

Under the direction of:

Date Reported:

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Patient Name:

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INFORMATION TABLE

SMA risk reductions for individuals with no family history						
Disorder (Gene) Reference Sequence	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	
Spinal muscular atrophy (SMN1) NM_000344	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

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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