ATTENTION
THIS PATIENT RESULT HAS MULTIPLE PAGES

Marrujo, Jazmyne Josephine DOB: **03/19/1991** Account Number: **88029331**
Patient ID: **64060508** Age: **32** Ordering Physician: **S Isquick**
Specimen ID: **040-364-0223-0** Sex: **Female**

Date Collected: **02/06/2024**	Date Received: **02/09/2024**	Date Reported: **02/22/2024**	Date Entered: **02/09/2024**

Specimen Type: **Whole Blood** Ethnicity: **Not Provided**
Indication: **Carrier Test / Screening**

Inheritest® CF / SMA Panel

2 genes

Summary: ● NEGATIVE

Negative Results

Disorder *(Gene)*	Result	Interpretation
Cystic fibrosis *(CFTR)* NM_000492.4	NEGATIVE	This result reduces, but does not eliminate, the risk to be a carrier. **Risk:** NOT at an increased risk for an affected pregnancy.
Spinal muscular atrophy *(SMN1)* NM_000344.4	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).

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

Next-generation Sequencing: Genomic regions of interest are selected using the Twist Biosciences® hybridization capture method and sequenced via the Illumina® next generation sequencing platform. Sequencing reads are aligned with the human genome reference GRCh37/hg19 build. Regions of interest include coding exons, intron/exon junctions (+/- 20 nucleotides) and additional genomic regions with known significant pathogenic variants. Analytical sensitivity at 30X coverage is estimated to be >99% for single nucleotide variants, >99% for insertions/deletions less than six base pairs and >96% for insertions/deletions between six and forty-five base pairs. Variant detection is performed by QIAGEN CLC Genomics and in-house algorithms. Expected minimum size resolution for CNVs in *CFTR* is 60 bp of coding sequence. Precise breakpoints are not reported. Single-exon deletions or duplications are not detected in some cases due to CNV size limitations, or due to isolated data quality variation or intrinsic sequence properties. Confirmatory testing by orthogonal technologies includes Sanger sequencing, MLPA analysis.

Reported variants: Pathogenic and likely pathogenic variants are reported for all tests. Benign and likely benign variants are typically not reported. Variants of uncertain significance are reported when included in the test specification. Variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, http://www.hgvs.org/). Variant classification and confirmation are consistent with ACMG standards and guidelines (Richards, PMID:25741868; Rehm, PMID:23887774). Detailed variant classification information and reevaluation are available upon request.

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.

References

Gregg AR, Aarabi M, Klugman S *et al.* Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American

Electronically released by Jennifer Reiner, PhD, FACMG

Marrujo, Jazmyne Josephine DOB: **03/19/1991** Account Number: **88029331**
Patient ID: **64060508** Age: **32** Ordering Physician: **S Isquick**
Specimen ID: **040-364-0223-0** Sex: **Female**

Inheritest® CF / SMA Panel

2 genes

References (Cont.)
College of Medical Genetics and Genomics (ACMG). *Genet Med* 23, 1793 (2021). PMID: 34285390

Disorders Tested
Cystic fibrosis (1 gene). Autosomal recessive: *CFTR*
Spinal muscular atrophy (1 gene). Autosomal recessive: *SMN1*

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. Inheritest® and GeneSeq® are registered service marks of Laboratory Corporation of America Holdings.

Patient Details
Marrujo, Jazmyne Josephine

Phone:
Date of Birth: **03/19/1991**
Age: **32**
Sex: **Female**
Patient ID: **64060508**
Alternate Patient ID:

Physician Details
S Isquick
Santa Clara Valley Medical Ctr
7475 Camino Arroyo, Gilroy, CA 95020

Phone: **(408) 852-2258**
Account Number: **88029331**
Physician ID:
NPI: **1386987493**

Specimen Details
Specimen ID: **04036402230**
Control ID: **64593936**
Alternate Control Number: **64593936**
Date Collected: **02/06/2024 0000 Local**
Date Received: **02/09/2024 1959 ET**
Date Entered: **02/09/2024 1431 ET**
Date Reported: **02/22/2024 1402 ET**

Electronically released by Jennifer Reiner, PhD, FACMG