Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4454

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 11/13/23

Donor Reported Ancestry: German, English Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by genotyping of 99 mutations in the CFTR gene	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	<1/500
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	<1/1500 for Beta-Thalassemia <1/500 for Sickle Cell
Special Testing		
Genes: ACADVL, ACSF3, DHCR7, HGSNAT, SLC12A3	Negative by gene sequencing	

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

Counsyl

Results Recipient	Male	Female
Fairfax Cryobank [redacted]	Name: DONOR 4454	Not tested
[redacted]	DOB: [redacted]	
Report Date: 06/22/2012	Ethnicity: Northern European	
	Sample Type: EDTA Blood	
	Date of Collection: 06/14/2012	
	Date Received: 06/18/2012	
	Barcode: [redacted]	
	Indication: No family history (screening)	

Counsyl Test Results

The Counsyl test (**Fairfax Cryobank Fundamental Panel**) uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for **128 variants** associated with **4 diseases**. This report indicates which mutations, if any, were detected for each mutation panel. Because only select mutations are tested, the percentage of carriers detected varies by ethnicity. A full list of mutations tested is given on page 2. A negative test result does not eliminate the possibility that the individual is a carrier. Interpretation is given as an estimate of the risk of conceiving a child affected with a disease, which is based on reported ethnicity, the test results, and an assumption of no family history.*

DONOR 4454

DONOR 4454's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical notes:

- Individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and may also benefit from carrier testing by CBC and hemoglobin electrophoresis or HPLC. *ACOG Practice Bulletin No. 78. Obstet Gynecol 2007;109:229-37.*
- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a free appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

Lab Directors:

ENTERED NP 03.10.12

Jessica Jacobson, MD

William Seltzer, PhD, FACMG

*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately. Other possible sources of diagnostic error include sample mix-up, trace contamination, and technical errors. The reproductive risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation. For the purposes of risk calculations, it is assumed that mutations within the same gene are on different chromosomes.

This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's workup. CLIA Number: **#05D1102604**.

Counsyl

Male	Female
Name: DONOR 4454 DOB: [redacted]	Not tested

Mutations Tested

Beta Thalassemia - **Gene:** HBB. **Variants (27):** K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab. **Detection rate**: Northern European 83%.

Cystic Fibrosis - **Gene:** CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, E92X, Y122X, G178R, R347H, Q493X, V520F, S549N, P574H, M1101K, D1152H, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K. **Detection rate**: Northern European 91%.

Sickle Cell Disease - **Gene:** HBB. **Variants (28):** Hb S, K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab. **Detection rate**: Northern European 70%.

Spinal Muscular Atrophy - **Gene:** SMN1. **Variants (1):** Exon 7 deletion. **Detection rate**: Northern European 95%.

Counsyl

Male	Female
Name: DONOR 4454 DOB: [redacted]	Not tested

Risk Calculations

Below are the full test results for all diseases on the panel. Listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that his future children could inherit each disease. A negative result does not rule out the possibility of being a carrier of untested mutations. Estimates of post-test carrier risk assume a negative family history.

Disease	Donor 4454 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
Beta Thalassemia	1 in 1,500	< 1 in 1,000,000	1 in 250,000
Cystic Fibrosis	1 in 310	1 in 34,000	1 in 3,000
Sickle Cell Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Spinal Muscular Atrophy	1 in 700	1 in 97,000	1 in 4,800

Report Status: Final

ID, 4454-120614

Patient Information	Specimen Information	Client Information
ID, 4454-120614 **DOB: Not Given** **AGE: Not Given** Gender: M Fasting: N Phone: NG Patient ID: NG	Specimen: NE600713G Requisition: 0524662 Collected: 06/14/2012 / 09:20 EDT Received: 06/15/2012 / 04:09 EDT Reported: 06/18/2012 / 14:05 EDT	Client #: 19104437 HO130000 STERN, HARVEY J FAIRFAX CRYO BANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.38		4.20-5.80 Million/uL	QHO
HEMOGLOBIN	16.6		13.2-17.1 g/dL	
HEMATOCRIT		**50.3 H**	38.5-50.0 %	
MCV	93.4		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
RDW	13.6		11.0-15.0 %	
HEMOGLOBIN A	96.7		>96.0 %	QHO
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.3		1.8-3.5 %	
INTERPRETATION				
	Normal phenotype.			
CHOLESTEROL, TOTAL	168		125-200 mg/dL	QHO
AST	18		10-35 U/L	QHO
ALT	14		9-60 U/L	QHO
CBC (INCLUDES DIFF/PLT)				QHO
WHITE BLOOD CELL COUNT	6.0		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.38		4.20-5.80 Million/uL	
HEMOGLOBIN	16.6		13.2-17.1 g/dL	
HEMATOCRIT		**50.3 H**	38.5-50.0 %	
MCV	93.4		80.0-100.0 fL	
MCH	30.9		27.0-33.0 pg	
MCHC	33.0		32.0-36.0 g/dL	
RDW	13.6		11.0-15.0 %	
PLATELET COUNT	158		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	4146		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1302		850-3900 cells/uL	
ABSOLUTE MONOCYTES	474		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	48		15-500 cells/uL	
ABSOLUTE BASOPHILS	30		0-200 cells/uL	
NEUTROPHILS	69.1		%	
LYMPHOCYTES	21.7		%	
MONOCYTES	7.9		%	
EOSINOPHILS	0.8		%	
BASOPHILS	0.5		%	
ABO GROUP AND RH TYPE				QHO
ABO GROUP	A			
RH TYPE	RH(D) POSITIVE			

HS 7/19/12

PERFORMING SITE:

QHO QUEST DIAGNOSTICS HORSHAM, 900 BUSINESS CENTER DRIVE, HORSHAM, PA 19044-3432 Laboratory Director: ANDREW S EDELMAN, MD PHD, CLIA: 39D0204404

LIST OF RESULTS PRINTED IN THE OUT OF RANGE COLUMN:

HEMATOCRIT	**50.3 H**	38.5-50.0 %	QHO
HEMATOCRIT	**50.3 H**	38.5-50.0 %	QHO

ENTERED NP 07.19.12

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted] **Email** N/A

Patient name/Donor Alias Donor # 4454 **Patient DOB** N/A

Donor # 4454-120614 **Specimen type** Peripheral Blood

Collection Date 06/14/2012 **Accession #** 12-074CG

Date Received 06/15/2012

RESULTS

CYTOGENETIC ANALYSIS

Cells counted 20

Cells analyzed 5

Cells karyotyped 2

Modal chromosome # 46

Type of banding GTG

Band resolution 550

KARYOTYPE 46,XY

FISH

Probe(s) N/A

Nuclei scored N/A

INTERPRETATION

Normal male karyotype

No clonal numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments

ENTERED NP 07.10.12

Wayne S. Stanley, Ph.D., FACMG
Clinical Cytogeneticist

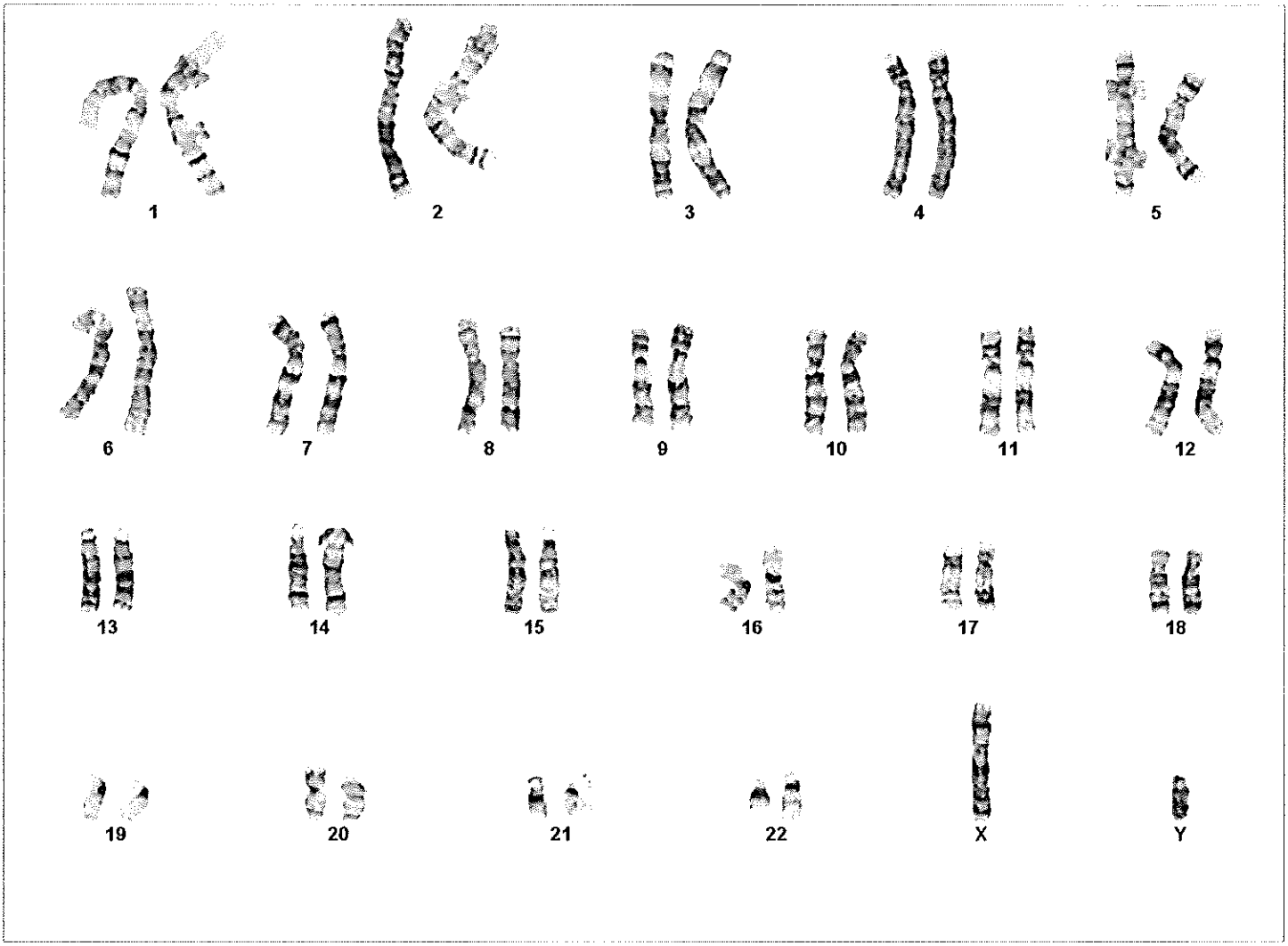
6/26/12
Date

Genetics and IVF Preimplantation Genetics Laboratory

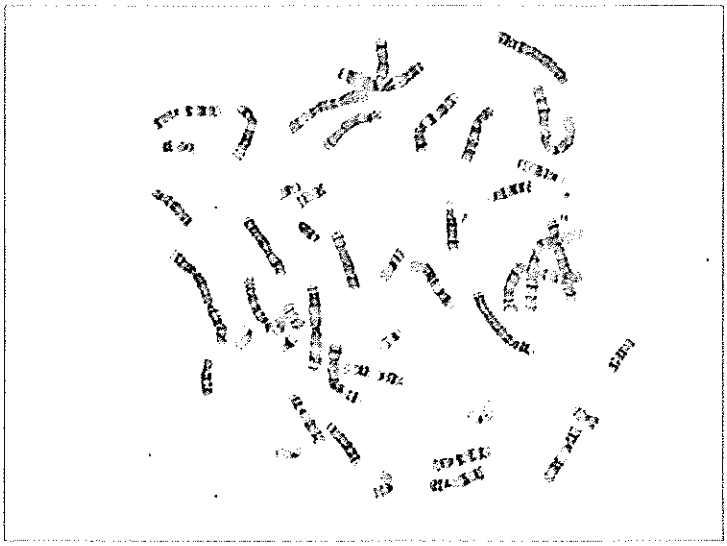
Patient name: DONOR # 4454

Case name: 12-074CG

46,XY



Case: 12-074CG Slide: A1 Cell: 4



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[redacted]

Patient Information:
4454, Donor
DOB: [redacted]
Sex: M
MR#: 4454
Patient#: [redacted]

Partner Information:
Not Tested

Physician:
Seitz, Suzanne
ATTN: Seitz, Suzanne
Fairfax Cryobank
3015 Williams Drive
Fairfax, VA 22031

Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Nov 09,2023**

Accession:
FT-5760037
Test#: FT-TS14695385
Specimen Type: DNA
Collected: Oct 24,2023

Accession:
N/A

FINAL RESULTS

No carrier mutations identified

TEST PERFORMED

Custom Beacon Carrier Screening Panel

(5 Gene Panel: *ACADVL, ACSF3, DHCR7, HGSNAT, and SLC12A3*; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- This report does not include variants of uncertain significance.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; https://www.nsgc.org)

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[redacted]

GENES TESTED:

Custom Beacon Carrier Screening Panel - 5 Genes

This analysis was run using the Custom Beacon Carrier Screening Panel gene list. 5 genes were tested with 100.0% of targets sequenced at >20x coverage. For more gene specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

ACADVL, ACSF3, DHCR7, HGSNAT, SLC12A3

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution

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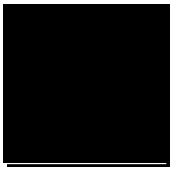

of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

SIGNATURE:

Dr. Harry Gao, DABMG, FACMG on **11/9/2023 10:53 PM PST**
Electronically signed

DISCLAIMER:

This test was developed and its performance characteristics determined by **Fulgent Genetics**. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or **info@fulgentgenetics.com**. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	General Population	1 in 118	93%	1 in 1,672	1 in 789,184
			Middle-Eastern Population	1 in 74	93%	1 in 1,044	1 in 309,024
			Native American Population	1 in 61	93%	1 in 858	1 in 209,352
			South Asian/Indian Population	1 in 73	93%	1 in 1,030	1 in 300,760
ACSF3	Combined malonic and methylmalonic aciduria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
DHCR7	Smith-Lemli-Opitz syndrome	AR	General Population	1 in 30	96%	1 in 726	1 in 87,120
			African/African American Population	1 in 138	96%	1 in 3,426	1 in 1,891,152
			Ashkenazi Jewish Population	1 in 36	96%	1 in 876	1 in 126,144
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population	1 in 434	98%	1 in 21,651	<1 in 10 million
			Caucasian / European Population	1 in 345	98%	1 in 17,201	<1 in 10 million
SLC12A3	Gitelman syndrome	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400

* For genes that have tested negative

Abbreviations: AR, autosomal recessive; XL, X-linked