



## Donor 4355

### Genetic Testing Summary

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

Last Updated: 04/03/23

Donor Reported Ancestry: Irish, German

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	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/290
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
<b>Special Testing</b>		
Gene: GBA	Negative by gene sequencing	See attached results.

\*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.

**Results Recipient**

Fairfax Cryobank - Fairfax

3015 Williams Drive Suite 201  
Fairfax, VA 22031

Report Date: 08/16/2013

**Male**

Name: DONOR 4355

DOB [REDACTED]

Ethnicity: Northern European

Sample Type: EDTA Blood

Date of Collection: 08/12/2013

Date Received: 08/14/2013

Barcode [REDACTED]

Indication: Egg or Sperm Donor

**Female**

Not tested

**Counsyl Test Results Summary (Egg or Sperm Donor)**

The Counsyl test (Fairfax Cryobank Fundamental Panel) uses targeted genotyping and copy number analysis as described in the methods section on page 2 to determine carrier status associated with 3 diseases. Please refer to page 3 for a complete list of diseases and genes included in this panel.

**DONOR 4355**

DONOR 4355'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**

- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complimentary appointment to speak with a genetic counselor about these results, please visit [counsyl.com/counseling/](http://counsyl.com/counseling/).

**ENTERED**  
08/17/13



Male

Name: DONOR 4355

DOB [REDACTED]

Female

Not tested

## Methods and Limitations

### DONOR 4355: targeted genotyping and copy number analysis.

**Targeted genotyping:** Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

**Copy number analysis:** Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

**Limitations:** In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's workup. CLIA Number: #05D1102604.

Lab Director:

*Hyunseok Kang*

H. Peter Kang, MD



Male

Name: DONOR 4355

DOB: [REDACTED]

Female

Not tested

## Diseases Tested

**Cystic Fibrosis** - Gene: CFTR. Variants (99): G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1262X, 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%.

**Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and 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 83%.

**Spinal Muscular Atrophy (copy number analysis only)** - Gene: SMN1. Variant (1): SMN1 copy number. Detection rate: Northern European 95%.



Male

Name: DONOR 4355

DOB: [REDACTED]

Female

Not tested

## Risk Calculations

Below are the risk calculations for all diseases tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	DONOR 4355 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000



GENETICS & IVF  
Institute

ENTERED  
11/29/2013

Cytogenetic Report

Client Fairfax Cryobank - Fairfax

Address 3015 Williams Drive  
Fairfax, VA 22031

Reporting Phone #



Patient name/Donor Alias Donor # 4355

Patient DOB N/A

Donor #



Specimen type Peripheral Blood

Collection Date 08/12/2013

Accession #



Date Received 08/12/2013

RESULTS

CYTOGENETIC ANALYSIS

FISH

Cells counted 20

Type of banding GTG

Probe(s) N/A

Cells analyzed 5

Band resolution 550

Nuclei scored N/A

Cells karyotyped 2

Modal chromosome # 46

KARYOTYPE 46,XY

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

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

8/29/13

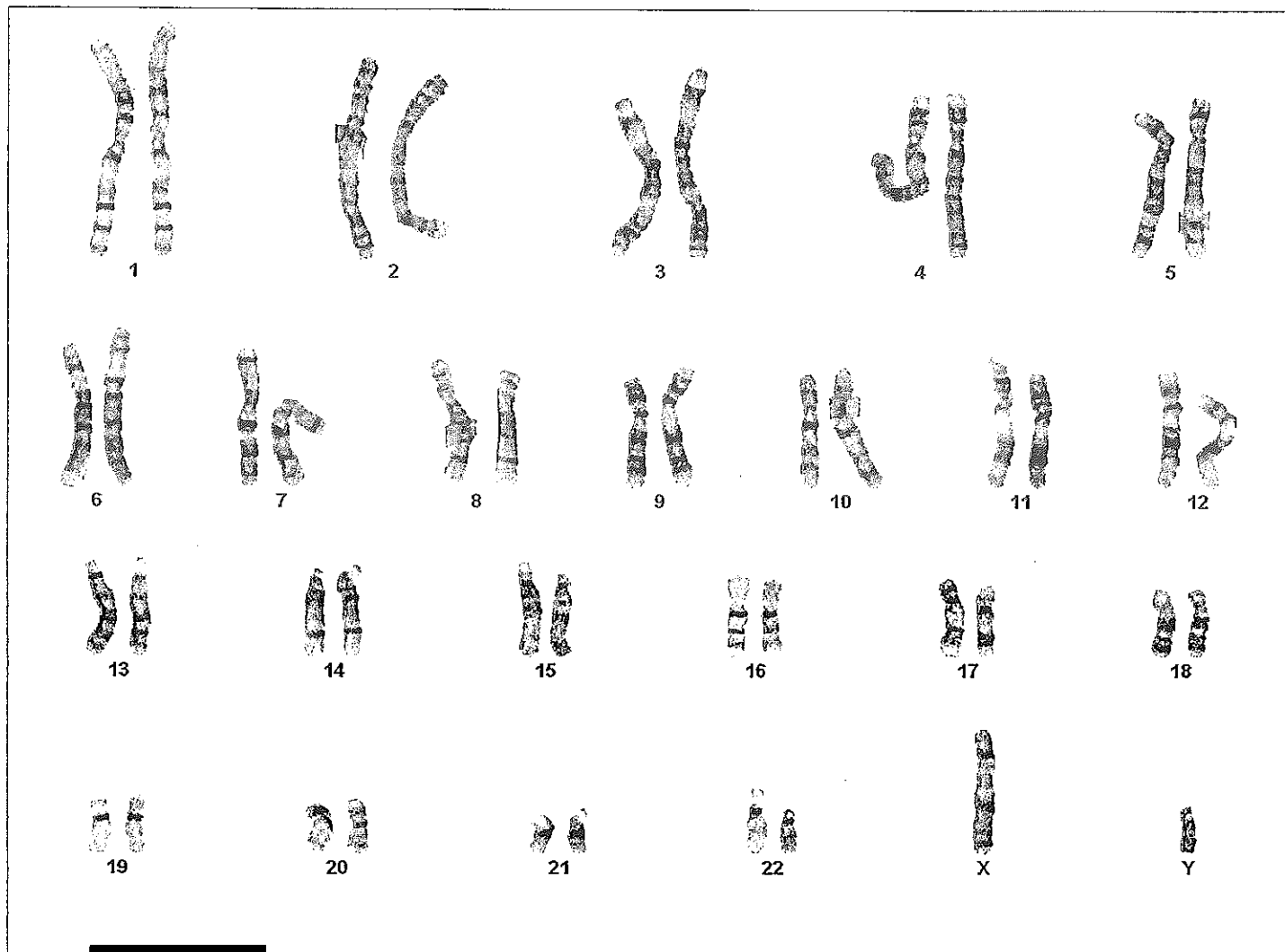
Date

# Genetics and IVF Preimplantation Genetics Laboratory

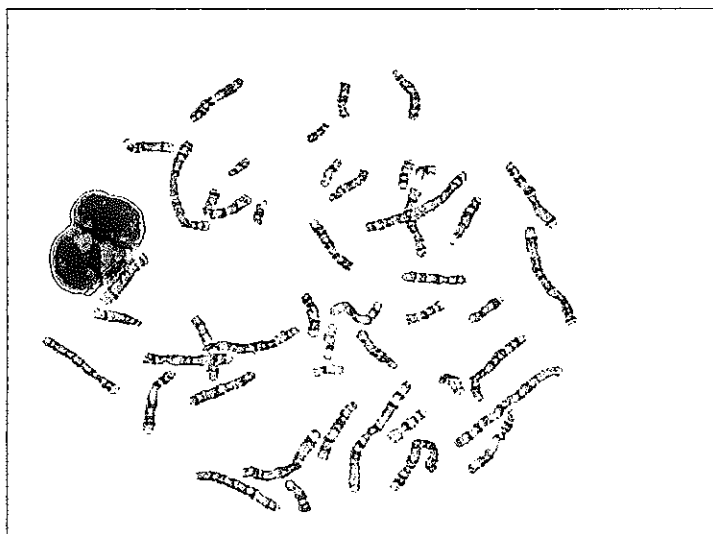
Patient name: DONOR # 4355

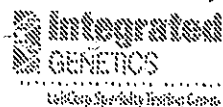
Case name: [REDACTED]

46,XY



Case: [REDACTED] Slide: C2 Cell: 13





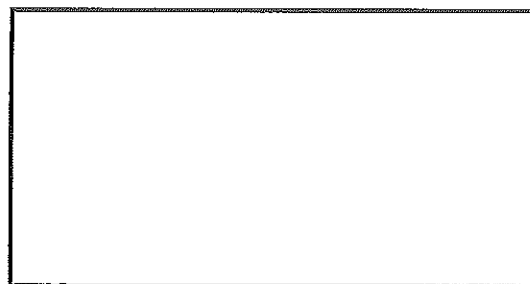
## Tay-Sachs Enzyme Analysis

Patient Name: Donor # 4355,  
Referring Physician: Steve Pool, MD  
Specimen #: ~~XXXXXXXXXX~~  
Patient ID: ~~XXXXXXXXXX~~

Client #: ~~XXXXXXXXXX~~

DOB: Not Given  
SSN: \*\*\*-\*\*-\*\*\*\*

Date Collected: 08/12/2013  
Date Received: 08/14/2013  
Lab ID:  
Hospital ID:  
Specimen Type: White Blood Cells



**RESULTS:** Hexosaminidase Activity : 1097 nmol/mg protein  
Hexosaminidase Percent A: 56.9

		Plasma/Serum	WBC
Expected Non-Carrier Range:	Hex A	≥54%	≥54%
Expected Carrier Range:	Hex A	20 - 49%	20 - 49%

**INTERPRETATION: NON CARRIER**

This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

ENTERED  
Aug 24 13

Under the direction of:



*Stanford Marenberg, PhD, MCC*  
Stanford Marenberg, Ph.D.

Testing Performed At Esoterix Genetic Laboratories, LLC 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

Date: 08/16/2013

Page 1 of 1



PATIENT INFORMATION  
4355, DONOR

REPORT STATUS Final

QUEST DIAGNOSTICS INCORPORATED  
CLIENT SERVICE 410.247.9100

DOB: [REDACTED] Age: [REDACTED]  
SEX: M

ORDERING PHYSICIAN

STERN, HARVEY

CLIENT INFORMATION

507059

FAIRFAX CRYOBANK

3015 WILLIAMS DR STE 110

FAIRFAX, VA 22031

SPECIMEN INFORMATION

SPECIMEN: [REDACTED]

REQUISITION: [REDACTED]

LAB REF NO:

ID: [REDACTED]

ENTERED  
08.24.13

COLLECTED: 08/12/2013 00:00  
RECEIVED: 08/12/2013 22:32  
REPORTED: 08/14/2013 15:09

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				QBA
RED BLOOD CELL COUNT	4.85		4.20-5.80 Million/uL	
HEMOGLOBIN	13.8		13.2-17.1 g/dL	
HEMATOCRIT	41.3		38.5-50.0 %	
MCV	85		80-100 fL	
MCH	28.3		27-33 pg	
RDW	14.8		11.0-15.0 %	
HEMOGLOBIN A	97.5		>96.0 %	
HEMOGLOBIN F	NONE DETECTED		0.0-1.9	
HEMOGLOBIN A2	2.5		1.8-3.5 %	
HGB SCREEN INTERPRETATION				
	THE HEMOGLOBINOPATHY SCREEN IS NORMAL.			
ABNORMAL HEMOGLOBIN #1 %:	0.0		%	

Performing Laboratory Information:

QBA Quest Diagnostics Incorporated 1901 Sulphur Spring Road Baltimore MD 21227 Laboratory Director: Edgar G. Khalluf, M.D.



Patient Information:

**4355, Donor**

**DOB:** [REDACTED]

**Sex:** M

**MR#:** 4355

**Patient#:** [REDACTED]

Accession:

**Test#:** [REDACTED]

**Order#:** [REDACTED]

**Ext Test#:** [REDACTED]

**Ext Order#:** [REDACTED]

**Specimen Type:** DNA

**Collected:** Feb 28, 2023

**Received Date:** Mar 07, 2023

**Authorized Date:** Mar 09, 2023

Physician:

**Seitz, Suzanne**

**ATTN:** Seitz, Suzanne

**Fairfax Cryobank**

**3015 Williams Drive**

**Fairfax, VA 22031**

**Phone:**

**Fax:**

Laboratory:

**Fulgent Genetics**

**CAP#:** 8042697

**CLIA#:** 05D2043189

**Laboratory Director:**

**Dr. Hanlin (Harry) Gao**

**Report Date:** Mar 24, 2023

Final Report

TEST PERFORMED

**GBA Single Gene**

(1 Gene Panel: *GBA*; gene sequencing with deletion and duplication analysis)

RESULTS:

**No clinically significant sequence or copy-number variants 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 of the sort not queried by this test or in areas not reliably assessed by this test.**

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- 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>)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (<https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep>)

GENES TESTED:

**GBA Single Gene**

1 genes tested (100.00% at >20x).

GBA

Gene Specific Notes and Limitations

*GBA*: Significant pseudogene interference and/or reciprocal exchanges between the *GBA* gene and its pseudogene, *GBAP1*, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of this individual's clinical findings, biochemical profile, and family history. The current testing method cannot detect copy-neutral rearrangements between the pseudogene and the functional gene, which have been reported in very rare cases of Gaucher disease (PubMed: [21704274](https://pubmed.ncbi.nlm.nih.gov/21704274/)).

**Patient:** 4355, Donor; **Sex:** M;

**DOB:** [REDACTED] **MR#:** [REDACTED]

**Accession#:** [REDACTED]; **FD Patient#:** [REDACTED];

**DocID:** [REDACTED]; **PAGE 1 of 3**



## 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 ( $\pm$  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 identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. New York patients: diagnostic findings are confirmed by Sanger, MLPA, or qPCR; exception SNV variants in genes for which confirmation of NGS results has been performed  $\geq 10$  times may not be confirmed if identified with high quality by NGS. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

## 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 this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. 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 designed and validated for detection of germline variants only. It 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 (eg. trinucleotide or hexanucleotide repeat expansion). 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 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 are two or more contiguous exons in size; 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.

## SIGNATURE:



**Zhenbin Chen, Ph.D., CGMBS, FACMG** on 3/24/2023 04:39 PM PDT

Electronically signed



#### DISCLAIMER:

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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](mailto: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.