Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4939

Genetic Testing Summary

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

Last Updated:11/27/23

Donor Reported Ancestry: Austrian, Russian, English Jewish Ancestry: Yes

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 Hexosamindase-A activity	
Standard Panel attached 17 diseases by genotyping	Negative for mutations tested	
Special Testing		
Gene: TTN	**Positive** for variant associated with autosomal dominant dilated cardiomyopathy (c.61876C>T; p.Arg20626*) – see attached	Children from this donor are at 50% risk to inherit this variant. All children should be tested and if positive, followed by a cardiologist.
Carnitine Palmitoyltransferase II Deficiency	Negative by genotyping of 21 mutations in the CPT2 gene	<1/600
Retinitis Pigmentosa: DHDDS Related	Negative by genotyping of 1 mutation in the DNDDS gene	<1/9100

*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.

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El Monte, CA 91731
(p) 626-350-0537 (f) 626-454-1667
info@fulgentgenetics.com
www.fulgentgenetics.com

fulgent

[redacted]

Patient Information
4939, Donor
DOB: [redacted]
Sex: M
MR#: 4939
FD Patient#: [redacted]

Accession: [redacted]
FD Test#: [redacted]
Order#: [redacted]
Ext Test#: [redacted]
Ext Order#: [redacted]
Specimen Type: DNA
Collected: Not Provided
Received Date: Sep 19, 2023
Authorized Date: Sep 21, 2023

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

Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Oct 16, 2023**

FINAL Report

TEST PERFORMED

Known Mutation / Site-Specific Testing
(1 Variant)

RESULTS:

1 VARIANT DETECTED

Gene Info		Variant Info			
GENE	INHERITANCE	VARIANT	ZYGOSITY	CLASSIFICATION	RESULTS
TTN NM_001267550.2	Autosomal Dominant & Autosomal Recessive	c.61876C>T p.Arg20626*	Heterozygous	**Pathogenic**	**DETECTED**

INTERPRETATION:

Notes and Recommendations:

- **The *TTN* variant was detected in the submitted extracted DNA specimen (reported source: semen) at a level consistent with the variant being heterozygous. Approximately 49% of NGS sequence reads show the alternate allele at this position in the TTN gene (374x coverage). Testing on a peripheral blood specimen could be considered.**
- Genetic counseling is recommended.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.

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The targeted test results for the specimen reported here indicate that NM_001267550.2:c.61876C>T (p.Arg20626*) in the TTN gene was **detected** (heterozygous) in this individual. Autosomal dominant mutations in TTN have been associated with autosomal dominant dilated cardiomyopathy-1G (CMD1G), familial hypertrophic cardiomyopathy-9 (CMH9), hereditary myopathy with early respiratory failure (Edstrom myopathy; HMERF), and tibial muscular dystrophy (Udd myopathy; TMD); biallelic mutations in TTN have been associated with limb-girdle muscular dystrophy type 2J (LGMD2J) and early-onset myopathy with fatal cardiomyopathy (Salih myopathy; EOMFC) (PubMed: 20301486, 24575448, 20301498, 20301582, 22238790; OMIM: 188840). Due to the high background frequency of TTN missense variants, the clinical significance of this type of variant is often not well established (PubMed: 26567375). Truncating variants in the TTN gene have been observed in individuals with dilated cardiomyopathy, especially end-stage dilated cardiomyopathy, and in the general population. However, truncating variants in the important domains (highly expressed exons) of the TTN gene are enriched in individuals with dilated cardiomyopathy as compared with the general population (PubMed: 25589632, 22335739, 24503780, 26777568). TTN truncating variants that affect highly expressed exons have 93% probability of pathogenicity (PubMed: 25589632). These truncating variants are located in the important domains (A band or I/A band junction) of the TTN gene (http://www.cardiodb.org/titin). This variant, p.Arg20626* (also reported as p.Arg18985*), has been reported in individuals with dilated cardiomyopathy (PubMed: 22335739, 24980681, 36264615). This variant has been observed at a frequency of less than 0.01% (20/280290 alleles) across the entire Broad dataset (individuals without severe childhood onset disease). The laboratory considers this variant to be pathogenic.

GENES TESTED:

Known Mutation Test

1 gene tested.

TTN

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). Methodology - Next Generation Sequencing (NGS), Sanger Sequencing, quantitative PCR (qPCR), repeat-primed PCR (rpPCR) or multiplex ligation-dependent probe amplification (MLPA) is selected by the laboratory to provide optimal results.

If NGS was performed:
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 / hg19), variants were detected in regions of at least 10x coverage. The known mutation genomic loci requested are evaluated for the presence or absence of variation compared to the human genome reference sequence. Bioinformatics: Fulgent Germline Pipeline v2019.1 or v2019.2 was used to generate variant calls for this test.

If Sanger Sequencing was performed:
DNA was amplified for the target region and sequenced bi-directionally using an ABI 3730XL instrument. The data was analyzed against the reference gene sequence and the known variant position as requested.

If qPCR was performed:
DNA was amplified for the target region and quantified using a QuantStudio 6 instrument. The data is compared to control genes and control individuals for the targets as requested.

If rpPCR was performed:
This analysis is performed by repeat-primed PCR (rpPCR) and amplicon length analysis. The scope of this assay is limited to repeat expansion analysis of the specified gene. Gene sequencing and deletion/duplication analysis are not included in this assay. This analysis does not include methylation studies.

If MLPA was performed:
DNA was amplified for the target regions and quantified using probesets using kits from MRC-Holland and an ABI 3730 instrument.The data is compared to control genes and control individuals for the targets as requested.

LIMITATIONS:

All laboratory tests have limitations. These results assume that the specimen received in the laboratory belongs to the named

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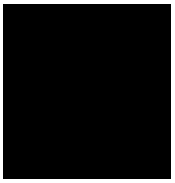
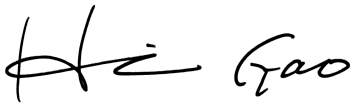


individual and that no mix-up or co-mingling of specimens has occurred. Positive results do not imply that there are no other pathogenic alterations in the patient's genome, and negative results do not rule out a genetic cause for the indication for testing. This assay assumes that any stated familial relationships are accurate. This assay is not designed or validated for the detection of somatic mosaicism or somatic mutations. This assay will only analyze the variant(s) requested. It is possible that the nomenclature for the variants tested may be different from the requested variants due to nomenclature differences in different isoforms of the gene. It is very important to provide us the isoform (NM number) of the gene for every variant to be tested. Result interpretation assumes that the human reference sequences are correct at the queried loci. 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 collected information available at the time of reporting; additional information may exist in the future which will not be represented. Rarely, due to systematic chemical or computational issues, or human error, DNA variants may be missed. If a positive familial control specimen is not provided or available, rare errors may occur.

Gene Specific Notes and Limitations

TTN
Due to the interference of tandem repeats within the TTN gene, the variants (sequencing and del/dup) in the genomic regions between exons 153-155 of transcript NM_133378.4 may not be detected by this test.

SIGNATURE:



Dr. Harry Gao, DABMG, FACMG on **Oct 16, 2023 10:01 PM**
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.

Counsyl

RESULTS RECIPIENT	MALE	FEMALE
FAIRFAX CRYOBANK - [redacted]	DONOR 4939	N/A
Attn: [redacted]	DOB: [redacted]	
NPI: 1417048786	Ethnicity: Mixed or Other Caucasian	
Report Date: 07/25/2014	Sample Type: EDTA Blood	
	Date of Collection: 07/21/2014	
	Date Received: 07/23/2014	
	Date Tested: 07/24/2014	
	Barcode: [redacted]	
	Indication: Egg or Sperm Donor	

Family Prep Screen

NEGATIVE

ABOUT THIS TEST

The Counsyl Family Prep Screen (version 1.0) tests known mutations to help you learn about your chance to have a child with a genetic disease.

PANEL DETAILS

Fairfax Cryobank Jewish Panel **(17 diseases tested)**

VERSION

DONOR 4939 **(Family Prep Screen 1.0)**

RESULTS SUMMARY

NEGATIVE

No known or potential disease-causing mutations were detected.

ENTERED OH 8·11·14

CLINICAL NOTES

- None

NEXT STEPS

- If necessary, patients can discuss residual risks with their physician or a genetic counselor.
- To schedule a complimentary appointment to speak with a clinical expert about these results, please visit counsyl.com/my/consults/.

Methods and Limitations

DONOR 4939 [Family Prep Screen 1.0]: targeted genotyping and copy number analysis.

Targeted genotyping: Targeted DNA mutation analysis is used to simultaneously determine the genotype of 179 variants associated with 16 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. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. 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*) and additional Tay-Sachs disease testing can be performed using a biochemical assay (*Gross et al. Genet. Med. 2008:10(1):54-56*).

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 DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Jelena Brezo

Jelena Brezo, PhD, FACMG

Diseases Tested

Autosomal Recessive Disorders

TARGETED GENOTYPING

ABCC8-related Hyperinsulinism - **Gene:** ABCC8. **Variants (3):** c.4160_4162delTCT, V187D, 3992-9G>A. **Detection rate:** Mixed or Other Caucasian <10%.

Bloom Syndrome - **Gene:** BLM. **Variant (1):** c.2207_2212del6ins7. **Detection rate:** Mixed or Other Caucasian <10%.

Canavan Disease - **Gene:** ASPA. **Variants (4):** E285A, Y231*, A305E, c.433-2A>G. **Detection rate:** Mixed or Other Caucasian 53%.

Cystic Fibrosis - **Gene:** CFTR. **Variants (99):** G85E, R117H, R334W, R347P, A455E, G542*, G551D, R553*, R560T, R1162*, W1282*, N1303K, c.1521_1523delCTT, c.1519_1521delATC, c.2052delA, c.3528delC, c.489+1G>T, c.579+1G>T, c.1585-1G>A, c.1766+1G>A, 2789+5G>A, c.2988+1G>A, 3849+10kbC>T, E60*, R75*, E92*, Y122*, G178R, R347H, Q493*, V520F, S549N, P574H, M1101K, D1152H, c.2012delT, c.262_263delTT, c.313delA, c.948delT, c.3744delA, c.3773dupT, c.1680-1G>A, 3272-26A>G, c.2051_2052delAAinsG, S549R, R117C, L206W, G330*, T338I, R352Q, S364P, G480C, C524*, S549R, Q552*, A559T, G622D, R709*, K710*, R764*, Q890*, R1066C, W1089*, Y1092X, R1158*, S1196*, W1204*, Q1238*, S1251N, S1255*, c.3067_3072del6, c.442delA, c.531delT, c.803delA, c.805_806delAT, c.1545_1546delTA, M607_Q643del, c.1911delG, c.1923_1931del9ins1, c.1976delA, c.3039delC, c.3536_3539delCCAA, c.3659delC, c.1155_1156dupTA, c.2052dupA, c.2175dupA, c.2738insG, 296+12T>C, c.273+1G>A, 405+3A>C, c.274-1G>A, 711+5G>A, c.580-1G>T, c.1766+1G>T, 1898+5G>T, Q996, c.325_327delTATinsG, 3849+4A>G, c.1075_1079del5ins5. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection rate:** Mixed or Other Caucasian 91%.

Familial Dysautonomia - **Gene:** IKBKAP. **Variants (2):** IVS20+6T>C, R696P. **Detection rate:** Mixed or Other Caucasian <10%.

Fanconi Anemia Type C - **Gene:** FANCC. **Variants (3):** IVS4+4A>T, c.67delG, R548*. **Detection rate:** Mixed or Other Caucasian 54%.

Gaucher Disease - **Gene:** GBA. **Variants (10):** N409S, L483P, c.84dupG, c.115+1G>A, V433L, R535H, D448H, D448V, R502C, R502H. **Detection rate:** Mixed or Other Caucasian 60%.

Glycogen Storage Disease Type Ia - **Gene:** G6PC. **Variants (7):** R83C, Q347*, c.79delC, c.379_380dupTA, R83H, G188R, Q242*. **Detection rate:** Mixed or Other Caucasian 61%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - **Gene:** HBB. **Variants (28):** E7V, K18*, Q40*, c.126_129delCTTT, c.27dupG, IVS-II-654, IVS-II-745, c.315+1G>A, IVS-I-6, IVS-I-110, IVS-I-5, c.92+1G>A, -88C>T, -28A>G, -29A>G, c.25_26delAA, c.217dupA, c.316-2A>C, c.316-2A>G, G25, -87C>G, E7K, W16*, c.51delC, c.20delA, E27K, E122Q, E122K. **Detection rate:** Mixed or Other Caucasian 83%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. **Variants (9):** c.1274_1277dupTATC, c.1421+1G>C, G269S, c.1073+1G>A, R178H, c.805+1G>A, 7.6kb del, G250D, R170W. **Detection rate:** Mixed or Other Caucasian 23%.

Lipoamide Dehydrogenase Deficiency - **Gene:** DLD. **Variants (2):** c.104dupA, G229C. **Detection rate:** Mixed or Other Caucasian <10%.

Maple Syrup Urine Disease Type 1B - **Gene:** BCKDHB. **Variants (3):** R183P, G278S, E372*. **Detection rate:** Mixed or Other Caucasian <10%.

Mucolipidosis IV - **Gene:** MCOLN1. **Variants (2):** 511_6944del, c.406-2A>G. **Detection rate:** Mixed or Other Caucasian <10%.

Niemann-Pick Disease, SMPD1-associated - **Gene:** SMPD1. **Variants (4):** c.996delC, L304P, R498L, c.1829_1831delGCC. **Detection rate:** Mixed or Other Caucasian 38%.

Usher Syndrome Type 1F - **Gene:** PCDH15. **Variant (1):** R245*. **Detection rate:** Mixed or Other Caucasian <10%.

Usher Syndrome Type 3 - **Gene:** CLRN1. **Variant (1):** N48K. **Detection rate:** Mixed or Other Caucasian <10%.

COPY NUMBER ANALYSIS

Spinal Muscular Atrophy - **Gene:** SMN1. **Variant (1):** SMN1 copy number. **Detection rate:** Mixed or Other Caucasian 95%.

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 4939 Residual Risk	Reproductive Risk
ABCC8-related Hyperinsulinism	1 in 110	1 in 50,000
Bloom Syndrome	< 1 in 500	< 1 in 1,000,000
Canavan Disease	< 1 in 500	< 1 in 1,000,000
Cystic Fibrosis	1 in 300	1 in 33,000
Familial Dysautonomia	< 1 in 500	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 340	1 in 220,000
Gaucher Disease	1 in 280	1 in 120,000
Glycogen Storage Disease Type Ia	1 in 450	1 in 320,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 390	1 in 470,000
Lipoamide Dehydrogenase Deficiency	< 1 in 500	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 250	1 in 250,000
Mucolipidosis IV	< 1 in 500	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 400	1 in 400,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000
Usher Syndrome Type 1F	1 in 190	1 in 150,000
Usher Syndrome Type 3	< 1 in 500	< 1 in 1,000,000

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors.

The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

Recombine

CarrierMap™

Ordering Practice:

Practice Code [redacted]
Fairfax Cryobank
[redacted]
Physician [redacted]
Report Generated: 2016-03-08

Donor 4939

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 43177
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Sperm, #44928
Specimen Collection: 2016-02-03
Specimen Received: 2016-02-05
Specimen Analyzed: 2016-03-08

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4939 was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♂ Male

Panel: Custom Panel , Diseases Tested: 1, Mutations Tested: 21, Genes Tested: 1, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician Bo Chu

Recombine CLIA # 31D2100763
Reviewed by Pere Colls, PhD, HCLD, Lab Director

This test was developed and its performance determined by Recombine Inc. and it has not been cleared or approved by the U.S. Food and Drug Administration.

Diseases & Mutations Assayed

High Impact Treatment Benefits X-Linked Moderate Impact

H	T	X	M	Disease	#	Mutations
●	●	○	○	Carnitine Palmitoyltransferase II Deficiency	21	♂ Genotyping | c.109_110insGC, c.1238_1239delAG, c.1737delC, c.1923_1935delGAAGGCCTTAGAA, c.534_558delGAACCCTGCAAAAAGTGACACTATCinsT, c.1649A>G (p.Q550R), c.1883A>C (p.Y628S), c.359A>G (p.Y120C), c.983A>G (p.D328G), c.149C>A (p.P50H), c.1507C>T (p.R503C), c.1810C>T (p.P604S), c.1891C>T (p.R631C), c.338C>T (p.S113L), c.370C>T (p.R124X), c.680C>T (p.P227L), c.1646G>A (p.G549D), c.452G>A (p.R151Q), c.520G>A (p.E174K), c.1148T>A (p.F383Y), c.1342T>C (p.F448L)

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2016-05-11

Donor 4939

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 43177
Kit Barcode: [redacted]
Specimen: Sperm, #44928
Specimen Collection: 2016-02-03
Specimen Received: 2016-02-05
Specimen Analyzed: 2016-05-11

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Mutations Tested: 1

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4939 was not identified to carry any of the mutation(s) tested.

No pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

For additional disease information, please visit recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

Assay performed by Reprogenetics
CLIA ID: 31D1054821
3 Regent Street, Livingston, NJ 07039
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763
Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in the genes tested. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors. The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

This test was developed and its performance determined by Recombine, Inc., and it has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

Diseases & Mutations Assayed

Retinitis Pigmentosa: DHDDS Related : Mutations (1): ♂ Genotyping | c.124A>G (p.K42E)

Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

Disease	Carrier Rate	Detection Rate	Residual Risk
Retinitis Pigmentosa: DHDDS Related	♂ Ashkenazi Jewish: 1/91	>99%	<1/9,100