Donor 4304

Genetic Testing Summary

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

Last Updated: 08/14/23

Donor Reported Ancestry: Dutch, Romanian, Russian, 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 (SMN1)	Negative for deletions in exon 7	1/700
Hb Beta Chain-Related Hemoglobinopathies	Negative for 28 mutations in the HBB gene	<1/500
Special Testing		
Genes: DLD, ACADVL, BTD, CYP21A2	Negative by genotyping	Completed 2014-2018- see attached.

*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
Fairfa obank - [redacted]	Name: DONOR 4304	Not tested
[redacted]	DOB: [redacted]	
	Ethnicity: Northern European	
	Sample Type: EDTA Blood	
	Date of Collection: 05/10/2012	
	Date Received: 05/14/2012	
	Barcode: [redacted]	
Report Date: 05/17/2012	Indication: Egg or Sperm Donor	

Counsyl Test Results (Egg or Sperm Donor)

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 4304

DONOR 4304'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.*
- Genetic counseling is recommended. To schedule a free appointment to speak with a genetic counselor about your results, please visit www.counsyl.com/appointment.

ENTERED DH 6.22.12

Lab Directors:

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.

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%.

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 4304 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

GENETICS & IVF
Institute

ENTERED DH 6.22.12

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted] **Fax #** [redacted] **Email** [redacted]

Patient name/Donor Alias Donor # 4304 **Patient DOB** N/A

Donor # 4304-[redacted] **Specimen type** Peripheral Blood

Collection Date 05/10/2012 **Accession #** [redacted]

Date Received 05/10/2012

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	50	**Type of banding**	GTG
Cells analyzed	5	**Band resolution**	550
Cells karyotyped	2		
Modal chromosome #	46		

FISH

Probe(s)	N/A
Nuclei scored	N/A

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

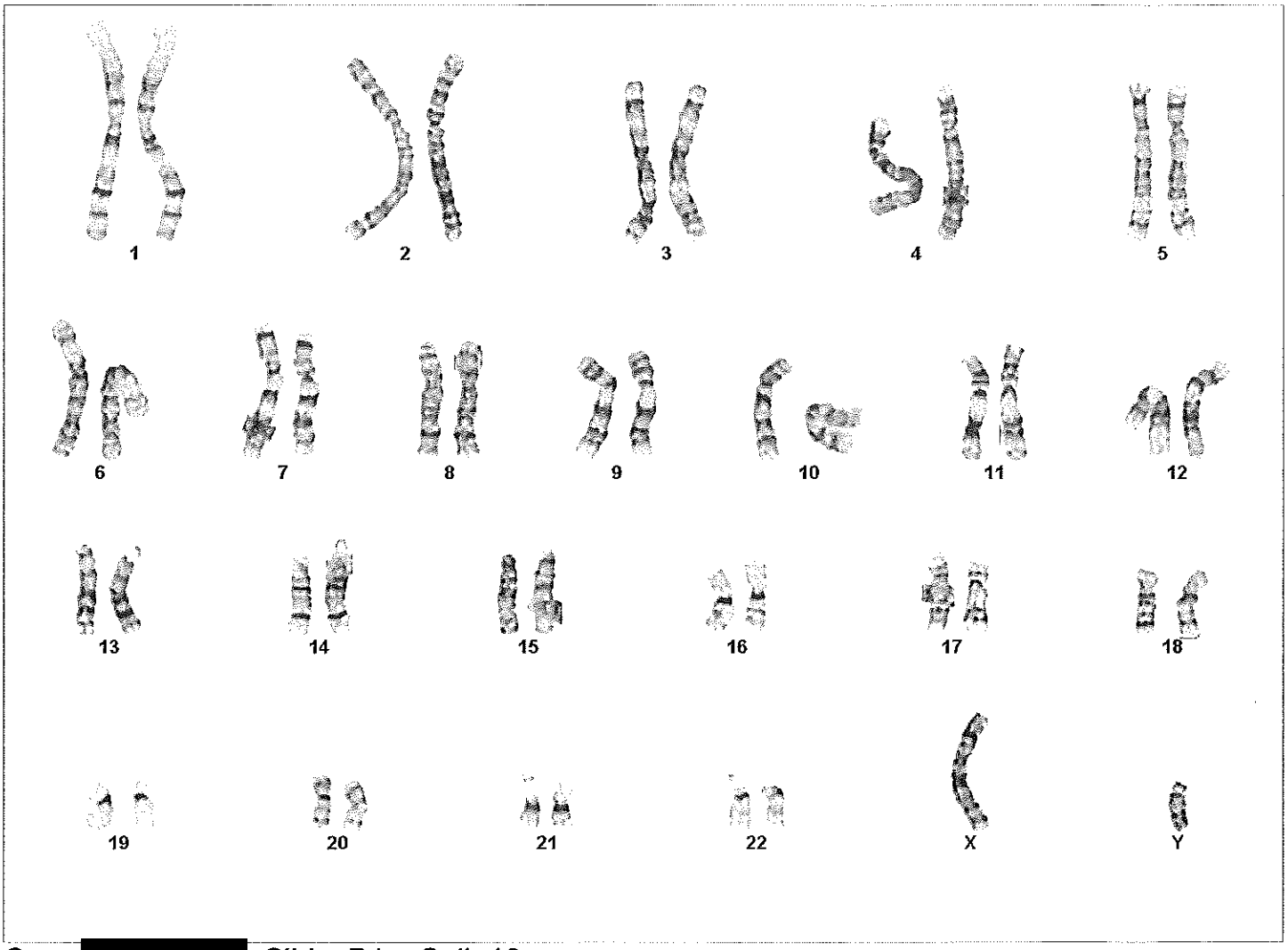
5/22/12
Date

Genetics and IVF Preimplantation Genetics Laboratory

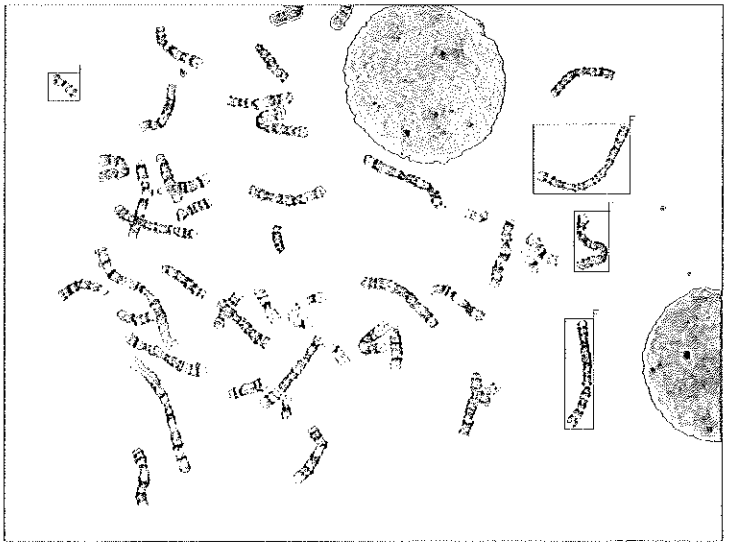
Patient name: DONOR # 4304

Case name: [redacted]

46,XY



Case: [redacted] Slide: B1 Cell: 10



PATIENT INFORMATION
4304,DONOR

REPORT STATUS **Final**

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 410.247.9100

DOB: [redacted] Age: [redacted]
GENDER: M

ORDERING PHYSICIAN
FAIRFAX CRYOBANK
CLIENT INFORMATION
507059
FAIRFAX CRYOBANK
[redacted]

SPECIMEN INFORMATION
SPECIMEN: [redacted]
REQUISITION:
LAB REF NO:

ID: 4304-[redacted]

COLLECTED: 05/10/2012 00:00
RECEIVED: 05/10/2012 22:57
REPORTED: 05/11/2012 15:44

Test Name	In Range	Out of Range	Reference Range	Lab
[redacted]				
CBC (INCLUDES DIFF-PLT)				QBA
WHITE BLOOD CELL COUNT	4.9		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	4.79		4.20-5.80 Million/uL	
HEMOGLOBIN	15.6		13.2-17.1 g/dL	
HEMATOCRIT	46.4		38.5-50.0 %	
MCV	97		80-100 fL	
MCH	32.6		27-33 pg	
MCHC	33.7		32-36 g/dL	
PLATELET COUNT	228		140-400 Thousand/uL	
RDW	14.1		11.0-15.0 %	
MPV	8.8		7.5-11.5 fL	
ABSOLUTE NEUTROPHILS	2298		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1838		850-3900 cells/uL	
ABSOLUTE MONOCYTES	642		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	123		15-500 cells/uL	
ABSOLUTE BASOPHILS	0		0-200 cells/uL	
NEUTROPHILS	46.9		%	
LYMPHOCYTES	37.5		%	
REACTIVE LYMPHOCYTES	0.0		%	
MONOCYTES	13.1		%	
EOSINOPHILS	2.5		%	
BASOPHILS	0.0		%	
COMMENT				
HEMOGLOBINOPATHY EVALUATION				QBA
RED BLOOD CELL COUNT	4.79		4.20-5.80 Million/uL	
HEMOGLOBIN	15.6		13.2-17.1 g/dL	
HEMATOCRIT	46.4		38.5-50.0 %	
MCV	97		80-100 fL	
MCH	32.6		27-33 pg	
RDW	14.1		11.0-15.0 %	
HEMOGLOBIN A	97.8		>96.0 %	
HEMOGLOBIN F	NONE DETECTED		0.0-1.9	
HEMOGLOBIN A2	2.2		1.8-3.5 %	
HGB SCREEN INTERPRETATION				
	THE HEMOGLOBINOPATHY SCREEN IS NORMAL.			

Recombine

CarrierMap™

A COMPREHENSIVE GENETIC CARRIER SCREEN

Assay performed by Reprogenetics
CLIA ID: 31D1054821

Patient Information

Patient: DONOR 4304
DOB: [redacted]
Gender: Male
Ethnicity: EUROPEAN
Procedure ID: 12362
Report Date: 11/05/2014
Report Updated: 11/05/2014

Sample Information

Specimen Type: Blood
Specimen Number: [redacted]
Barcode: [redacted]
Date Specimen Collected: 10/24/2014
Date Specimen Received: 10/27/2014
Date Specimen Analyzed: 11/05/2014

Ordering Practice

[redacted]

Summary of Genetic Testing Results

You did not test positive for any of the mutations assayed.

Disease Groups

High Impact	Treatment Benefits	X-Linked	Moderate Impact
These diseases have a significant impact on life expectancy and quality of life.	Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.	These diseases are passed down by female carriers. Carriers may have symptoms.	These diseases typically do not affect life expectancy but can affect quality of life.

Summary of Testing Performed

Diseases Tested: 1	Mutations Tested: 8	Genes Tested: 1

All other mutations analyzed by Recombine were not detected. This reduces but does not eliminate your chance to be a carrier for the associated genetic diseases. Recombine does not screen for every possible genetic disease.

Learn More

A list of all the genetic diseases and mutations you were screened for is included in the latter pages of this report. For disease information, please visit **www.recombine.com/our-test**. To speak with a Genetic Counselor, call **855.OUR.GENES**

Lab Technician: Sana Khurshid

Reviewed by: Pere Colls, PhD, HCLD

**Methods and Limitations: Recombine developed this genetic assay using the Illumina Infinium Custom HD Genotyping Assay. The test is intended for clinical preconception and/or prenatal screening purposes and is not validated for detection of homozygous mutations. False positive or negative results may occur for reasons that include: genetic variants, assay limitations, sample mix-up, sample contamination, and molecular and technical errors. Recombine tests for Spinal Muscular Atrophy via an Identity-by-State shared haplotype comparison algorithm. Detection is limited to haplotypes within our library of known carriers of the most common mutation (deletion of Exon 7).*

Diseases & Mutations Assayed

Groups	Disease	#	Mutations
●●○○	Maple Syrup Urine Disease: Type 3 (DLD)	8	c.A1483G (p.R495G), c.C1463T (p.P488L), c.T1178C (p.I393T), c.G1123A (p.E375K), c.A1081G (p.M361V), c.A214G (p.K72E), c.104_105insA, c.G685T (p.G229C)

Recombine

CarrierMap™

A COMPREHENSIVE GENETIC CARRIER SCREEN

Assay performed by Reprogenetics
CLIA ID: 31D1054821

Patient Information

Patient: DONOR DONOR 4304
DOB: [redacted]
Gender: Male
Ethnicity: EUROPEAN
Procedure ID: 17466
Report Date: 03/13/2015
Report Updated: 03/13/2015

Sample Information

Specimen Type: Blood
Specimen Number: [redacted]
Barcode: [redacted]
Date Specimen Collected: 03/06/2015
Date Specimen Received: 03/09/2015
Date Specimen Analyzed: 03/13/2015

Ordering Practice

[redacted]

Summary of Genetic Testing Results

You did not test positive for any of the mutations assayed.

Disease Groups

High Impact	Treatment Benefits	X-Linked	Moderate Impact
These diseases have a significant impact on life expectancy and quality of life.	Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.	These diseases are passed down by female carriers. Carriers may have symptoms.	These diseases typically do not affect life expectancy but can affect quality of life.

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Diseases Tested: 1	Mutations Tested: 8	Genes Tested: 1

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Learn More

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Lab Technician: Bo Chu

Reviewed by: Pere Colls, PhD, HCLD

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Diseases & Mutations Assayed

Groups	Disease	#	Mutations
●●○○	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	8	c.1837C>T (p.R613W), c.1405C>T (p.R469W), c.1372T>C (p.F458L), c.1322G>A (p.G441D), c.1226C>T (p.T409M), c.1144A>C (p.K382Q), c.848T>C (p.V283A), c.779C>T (p.T260M)

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
[redacted]
Report Generated: 2017-07-13

Donor 4304

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 12362
Kit Barcode: [redacted]
Specimen: Sperm, #98833
Specimen Collection: 2017-06-23
Specimen Received: 2017-06-24
Specimen Analyzed: 2017-07-13

TEST INFORMATION

Test: CarrierMap^{SEQ} (Genotyping & Sequencing)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Genes Sequenced: 1

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4304 was not identified to carry any pathogenic mutations in the gene(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.

Sequencing: Sequencing is performed using a custom next-generation sequencing (NGS) platform. Only the described exons for each gene listed are sequenced. Variants outside of these regions may not be identified. Some splicing mutations may not be identified. Triplet repeat expansions, intronic mutations, and large insertions and deletions may not be detected. All identified variants are curated, and determination of the likelihood of their pathogenicity is made based on examining allele frequency, segregation studies, predicted effect, functional studies, case/control studies, and other analyses. All variants identified via sequencing that are reported to cause disease in the primary scientific literature will be reported. Variants considered to be benign and variants of unknown significance (VUS) are NOT reported. In the sequencing process, interval drop-out may occur, leading to intervals of insufficient coverage. Intervals of insufficient coverage will be reported if they occur.

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

Biotinidase Deficiency (BTD): Mutations (21): ♂ Genotyping | c.98_104delGCGGCTGinsTCC (p.C33FfsX68), c.1368A>C (p.Q456H), c.755A>G (p.D252G), c.1612C>T (p.R538C), c.235C>T (p.R79C), c.100G>A (p.G34S), c.1330G>C (p.D444H), c.511G>A (p.A171T), c.1207T>G (p.F403V), c.470G>A (p.R157H), c.1595C>T (p.T532M), c.1489C>T (p.P497S), c.341G>T (p.G114V), c.1052delC (p.T351fs), c.393delC (p.F131Lfs), c.1049delC (p.A350fs), c.1239delC (p.Y414Ifs), c.1240_1251delTATCTCCACGTC (p.Y414_V417del), c.278A>G (p.Y93C), c.595G>A (p.V199M), c.933delT (p.S311Rfs) Sequencing | NM_000060:1-4

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
Biotinidase Deficiency	♂ General: 1/123	78.32%	1/567

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
[redacted]
Report Generated: 2018-01-17

Donor 4304

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 12362
Kit Barcode: [redacted]
Specimen: Sperm, #98833
Specimen Collection: 2017-06-23
Specimen Received: 2017-06-24
Specimen Analyzed: 2018-01-17

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 4304 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

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Diseases & Mutations Assayed

21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia (CYP21A2):
Mutations (1): ♂ Genotyping | c.293-13C>G

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
21-Hydroxylase-Deficient Classical Congenital Adrenal Hyperplasia	♂ European: 1/62	27.65%	1/86
	♂ General: 1/62	29.34%	1/88