Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4334

Genetic Testing Summary

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

Last Updated: 07/20/18

Donor Reported Ancestry: African American, Native American 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 for 99 mutations in the CFTR gene	1/290
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 of the SMN1 gene	1/120
Hb Beta Chain Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 28 mutations tested by genotyping in the HBB gene	1/43
Tay Sachs Enzyme Analysis	Non Carrier by Hexosaminidase A activity	
Special testing Request:		
Alpha Thalassemia	Negative for 10 mutations in the HBA1 and HBA2 genes	1/97
Polyglandular Autoimmune Syndrome Type 1	Negative for 5 mutations in the AIRE gene	Unknown residual risk
Alpha-1-Antitrypsin Deficiency	Negative for 4 mutation in the SERPINA1 gene	1/700

Amegakaryocytic Thrombocytopenia	Negative for 3 mutation in the MPL gene	Unknown residual risk
Werner Syndrome	Negative for 6 mutation in the WRN gene	1/326

*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.

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2015-11-10
Report Updated: 2015-11-10

Donor 4334

DOB:
Gender: Male
Ethnicity: African
Procedure ID: 34498
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #35983
Specimen Collection: 2015-11-02
Specimen Received: 2015-11-03
Specimen Analyzed: 2015-11-10

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4334 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: Alpha Thalassemia , Diseases Tested: 1, Mutations Tested: 10, Genes Tested: 2, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
Lab Technician Bo Chu

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 >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.

Diseases & Mutations Assayed

High Impact Treatment Benefits X-Linked Moderate Impact

H	T	X	M	Disease	#	Mutations
●	○	○	○	Alpha Thalassemia	10	♂ Genotyping | SEA deletion, 11.1kb deletion, c.207C>A (p.N69K), c.223G>C (p.D75G), c.2T>C (p.M1T), c.207C>G (p.N69K), c.340_351delCTCCCCGCCGAG (p.L114_ E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2016-11-01

Donor 4334

DOB:
Gender: Male
Ethnicity: African
Procedure ID: 34498
Kit Barcode: [redacted]
Specimen: Blood, #35983
Specimen Collection: 2015-11-02
Specimen Received: 2015-11-03
Specimen Analyzed: 2016-11-01

TEST INFORMATION

Test: CarrierMap^{GEN} (Genotyping)
Panel: Custom Panel
Diseases Tested: 3
Genes Tested: 3
Mutations Tested: 13

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4334 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

Alpha-1-Antitrypsin Deficiency (SERPINA1): Mutations (4): ♂ Genotyping | c.226_228delTTC (p.76delF), c.1131A>T (p.L377F), c.187C>T (p.R63C), c.1096G>A (p.E366K)

Amegakaryocytic Thrombocytopenia (MPL): Mutations (3): ♂ Genotyping | c.79+2T>A (IVS1+2T>A), c.127C>T (p.R43X), c.305G>C (p.R102P)

Werner Syndrome (WRN): Mutations (6): ♂ Genotyping | c.3139-1G>C (IVS25-1G>C), c.3913C>T (p.R1305X), c.3493C>T (p.Q1165X), c.1730A>T (p.K577M), c.1336C>T (p.R368X), c.2089-3024A>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
Alpha-1-Antitrypsin Deficiency	♂ European: 1/35	95.00%	1/700
	♂ General: Unknown	95.00%	Unknown
Amegakaryocytic Thrombocytopenia	♂ Ashkenazi Jewish: 1/76	>99%	<1/7,600
	♂ General: Unknown	64.81%	Unknown
Werner Syndrome	♂ General: 1/224	31.25%	1/326
	♂ Japanese: 1/87	65.62%	1/253

CooperGenomics℠
a CooperSurgical company
Reprogenetics℠ Recombine℠ Genesis Genetics℠

CarrierMap℠

Ordering Practice

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2018-05-22

Donor 4334

DOB:
Gender: Male
Ethnicity: African
Procedure ID: 34,498
Kit Barcode: [redacted]
Specimen: Blood, #35,983
Specimen Collection: 2015-11-02
Specimen Received: 2015-11-03
Specimen Analyzed: 2018-05-22

TEST INFORMATION

Test: Carriermap GEN (Genotyping)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Mutations Tested: 5

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4334 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 www.coopergenomics.com/diseases . To speak with a genetic counselor, call 855.687.4363 .

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

Recombine CLIA ID: 31D2100763
Reviewed by: Pere Colls, PhD, HCLD

Methods and Limitations

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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 does not currently regulate laboratory developed tests (LDTs).

Diseases & Mutations Assayed

Polyglandular Autoimmune Syndrome: Type I (AIRE): Mutation(s) (5): ♂ Genotyping | c.1163_1164insA (p.M388IfsX36), c.254A>G (p.Y85C), c.415C>T (p.R139X), c.769C>T (p.R257X), c.967_979delCTGTCCCCTCCGC (p.L323SfsX51)

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
Polyglandular Autoimmune Syndrome: Type I	♂ Finnish: 1/80	90.48%	1/840
	♂ Iranian Jewish: 1/48	>99%	<1/4800
	♂ Italian: Unknown	27.78%	Unknown
	♂ Norwegian: 1/142	47.92%	1/273
	♂ Sardinians: 1/61	81.82%	1/336
	♂ United Kingdom: Unknown	70.00%	Unknown
	♂ United States: Unknown	65.62%	Unknown