Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4870

Genetic Testing Summary

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

Last Updated: 08/17/18

Donor Reported Ancestry: German, Scottish, English, French, Irish 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	
Special Testing		
Gaucher Disease	Negative for 6 mutations by genotyping in the GBA gene	1/164
Stargardt Disease	Negative by gene sequencing in the ABCA4 gene	1/2564

Glycogen Storage Disease Type II	Negative by gene sequencing in the GAA gene	1/1389
Familial Dysautonomia	Negative by gene sequencing in the IKBKAP gene	1/10000
Pendred Syndrome	Negative by gene sequencing in the SLC26A4 gene	1/203
Zellweger Spectrum Disorder PEX6	Negative by gene sequencing in the PEX6 gene	1/424

*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: 2016-01-29

4870 Donor

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 30219
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #31657
Specimen Collection: 2015-09-11
Specimen Received: 2015-09-14
Specimen Analyzed: 2016-01-29

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

4870 Donor 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: 6, 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.

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
●	●	○	○	Gaucher Disease	6	♂ Genotyping | c.84_85insG, c.A1226G (p.N409S), c.A1343T (p.D448V), c.C1504T (p.R502C), c.G1297T (p.V433L), c.G1604A (p.R535H)

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2015-10-06
Report Updated: 2015-10-06

4870 Donor

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 30219
Kit Barcode: [redacted]
Method: Genotyping & Sequencing
Specimen: Blood, #31657
Specimen Collection: 2015-09-11
Specimen Received: 2015-09-14
Specimen Analyzed: 2015-10-06

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

4870 Donor 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: Glycogen Storage Disease: Type II Sequencing , Diseases Tested: 1, Mutations Tested: 14, Genes Tested: 1, 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.

Sequencing: The Illumina TruSight One platform is used to perform sequencing. 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.

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
●	●	○	○	Glycogen Storage Disease: Type II	14	♂ Genotyping | c.C1935A (p.D645E), c.C2560T (p.R854X), c.-32-13T>G, c.525delT (p.E176Rfs), c.C710T (p.A237V), c.T896G (p.L299R), c.T953C (p.M318T), c.G1561A (p.E521K), c.C1634T (p.P545L), c.G1927A (p.G643R), c.C2173T (p.R725W), c.2707_2709delK (p.903delK) Sequencing | NM_001079804:2-20

CooperGenomics
a CooperSurgical company
Reprogenetics℠ Recombine℠ Genesis Genetics℠

CarrierMap℠

Ordering Practice

Practice Code: [redacted]
Fairfax Cryobank
[redacted]
Physician: [redacted]
Report Generated: 2018-06-18

4870 Donor

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 30,219
Kit Barcode: [redacted]
Specimen: Blood, #31,657
Specimen Collection: 2015-09-11
Specimen Received: 2015-09-14
Specimen Analyzed: 2018-06-18

TEST INFORMATION

Test: Carriermap GEN (Genotyping)
Panel: Custom Panel
Diseases Tested: 2
Genes Tested: 2
Mutations Tested: 14

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

4870 Donor 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 .

Assay performed by Reprogenetics
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

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 existing mutations 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 does not currently regulate laboratory developed tests (LDTs).

Diseases & Mutations Assayed

Pendred Syndrome (SLC26A4): Mutation(s) (7): ♂ Genotyping | c.1001+1G>A, c.1151A>G (p.E384G), c.1246A>C (p.T416P), c.2168A>G (p.H723R), c.707T>C (p.L236P), c.716T>A (p.V239D), c.919-2A>G

Zellweger Spectrum Disorders: PEX6 Related (PEX6): Mutation(s) (7): ♂ Genotyping | c.1130+1G>A (IVS3+1G>A), c.1301delC (p.S434Ffs), c.1601T>C (p.L534P), c.1688+1G>A (IVS7+1G>A), c.1962-1G>A (p.L655fsX3), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT (p.D268Cfs)

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
Pendred Syndrome	♂ European: 1/58	42.11%	1/100
	♂ Japanese: Unknown	45.83%	Unknown
	♂ Pakistani: Unknown	29.82%	Unknown
Zellweger Spectrum Disorders: PEX6 Related	♂ General: 1/288	30.00%	1/411

Patient	Sample	Referring Doctor
Patient Name: 4870 Donor **Date of Birth:** [redacted] **Reference #:** N/A **Indication:** Carrier Testing **Test Type:** NGS single gene full sequencing test	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 8/24/2016 **Date Received:** 8/25/2016 **Final Report:** 9/8/2016	[redacted] Fax: [redacted]

RESULTS

Results: No clinically significant variant(s) detected

Gene(s) analyzed: *SLC26A4, PEX6*

Interpretation: Screening for the presence of pathogenic variants in the *SLC26A4* and *PEX6* genes which are associated with Pendred syndrome and Zellweger syndrome spectrum (PEX6-Related) was performed by next generation sequencing and possibly genotyping for select variants on DNA extracted from this patient's sample. No clinically significant variants were detected during this analysis. This negative result does not rule out the possibility that a pathogenic variant in the gene examined is present.

Genetic counseling is recommended.

This technology may not detect all small insertion/deletions and is not diagnostic for large duplications/deletions and structural genomic variation. The coding DNA sequence of the gene plus at least five base pairs flanking splice sites were sequenced and analyzed relative to the hg19 assembly. A mutation(s) deep in intronic sequences or in untranslated regions of the gene except a portion described above or a pathogenic variant(s) in other genes not included in this test could be present in this patient. The analytical sensitivity of this test is estimated at 99% for single base substitutions and 97% overall. All potentially pathogenic variants were subjected to Sanger sequencing or genotyping by allele specific primer extension analysis for confirmation of the result. Any benign variants identified during this analysis were not reported.

Please note that this carrier screening test masks likely benign variants and variants of uncertain significance (VUS) if there are any. Only known pathogenic variants or likely pathogenic variants which are expected to result in deleterious effects on protein function are reported. If reporting of likely benign variants and VUS is desired in this patient, please contact the laboratory (tel. 212-241-2537) to request an amended report.

Comments: This test was developed and its performance characteristics were determined by The Genetic Testing Laboratory at the Mount Sinai School of Medicine. It is considered acceptable for patient testing. It has not been cleared or approved by the FDA. The FDA has determined that such clearance or approval is not necessary.

This type of mutation analysis generally provides highly accurate genotype information for point mutations and single nucleotide polymorphisms. Despite this level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, mosaicism or other rare genetic variants that interfere with analysis. In addition, families should understand the limitations of the testing and that rare diagnostic errors may occur for the reasons described.

[redacted]

For Disease Specific Standards and Guidelines:
https://www.acmg.net/
Additional disease-specific references available upon request.

[redacted]

This case has been reviewed and electronically signed by Ruth Kornreich, Ph.D., Co-Laboratory Director

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.

Table of Residual Risks by Ethnicity

Please note: This table displays residual risks after a negative result for each of the genes and corresponding disorders. **If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.**

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Pendred Syndrome (AR)	*SLC26A4*	Caucasian	1 in 88	57%	1 in 203
NM_000441.1		African	1 in 76	>95%	1 in 1501
		Asian	1 in 74	75%	1 in 293
Exons: 1-19, 21; **Variants:** 13		Worldwide	1 in 80	71%	1 in 273
Zellweger Syndrome Spectrum	*PEX6*	Worldwide	1 in 280	34%	1 in 424
(*PEX6*-Related) (AR)		French Canadian	1 in 55	>95%	1 in 1081
NM_000287.3		Sephardic Jewish - Yemenite	1 in 18	>95%	1 in 341
Exons: 1, 3, 5, 7-15; **Variants:** 7					

Note: A list of specific variants tested will be provided upon request.
AR: Autosomal Recessive