Donor 4824

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: English, Irish, Norwegian, Swedish 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)	Negative for 28 mutations tested by genotyping in the HBB gene	1/290
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
Special Testing		
Alpha 1 Antitrypsin Deficiency	Negative for 4 mutations by genotyping in the SERPINA1 gene	1/700
3-Methylcrontonyl-CoA Carboxylase Deficiency (MCCC1-Related)	Negative by gene sequencing in the MCCC1 gene	1/592

3-Methylcrontonyl-CoA Carboxylase Deficiency (MCCC2-Related)	Negative by gene sequencing in the MCCC2 gene	1/272
Maple Syrup Urine Disease, Type 1 B	Negative by gene sequencing in the BCKDHB gene	1/983
Non-Syndromic Hearing Loss (GJB2-Related)	Negative by gene sequencing in the GJB2 gene	1/343
Bardet-Biedl Syndrome (BBS1-Related)	Negative by gene sequencing in the BBS1 gene	1/5587
Biotinidase Deficiency	Negative by gene sequencing in the BTD gene	1/43
Congenital Disorder of Glycosylation, Type 1 A	Negative by gene sequencing in the PMM2 gene	1/229
Factor XI Deficiency	Negative by gene sequencing in the F11 gene	1/119
Phenylalanine Hydroxylase Deficiency	Negative by gene sequencing in the PAH gene	1/197

*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]
[redacted]
Physician: [redacted]
Report Generated: 2015-09-08
Report Updated: 2015-09-08

4824 4824

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 29123
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #30564
Specimen Collection: 2015-08-28
Specimen Received: 2015-08-31
Specimen Analyzed: 2015-09-08

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

4824 4824 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-1-Antitrypsin Deficiency , Diseases Tested: 1, Mutations Tested: 4, 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.

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-1-Antitrypsin Deficiency	4	♂ Genotyping | c.226_228delTTC (p.76delF), c.A1131T (p.L377F), c.C187T (p.R63C), c.G1096A (p.E366K)

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CARRIER SCREENING REPORT

Patient	Sample	Referring Doctor
Patient Name: 4824 Donor **Date of Birth:** [redacted] **Reference #:** [redacted] **Indication:** Carrier Testing [redacted]	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 10/7/2016 **Date Received:** 1/30/2018 **Final Report:** 2/6/2018	[redacted] **Fairfax Cryobank, Inc.** [redacted] [redacted] [redacted] Fax: [redacted]

RESULT SUMMARY

Results: No clinically significant variant(s) detected

Gene(s) analyzed: *BBS1*

Interpretation: Screening for the presence of pathogenic variants in the *BBS1* gene which is associated with Bardet-Biedl syndrome (*BBS1*-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 Mount Sinai Genomics, Inc. 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.

Patient:	4824 Donor	DOB:	[redacted]	Lab #:	[redacted]

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.

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

This case has been reviewed and electronically signed by Guiqing Cai, Ph.D., DABMGG, Associate 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	Analytical Detection Rate
Bardet-Biedl Syndrome (*BBS1*-Related) (AR)	*BBS1*	Worldwide	1 in 392	93%	1 in 5587	>95%
NM_024649.4		Faroese	1 in 30	>95%	1 in 581	>95%

AR: Autosomal Recessive

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CARRIER SCREENING REPORT

Patient	Sample	Referring Doctor
Patient Name: Donor 4824 **Date of Birth:** [redacted] **Reference #:** FFAXCB-S41661158NG **Indication:** Carrier Testing **Test Type:** Unmask Additional Gene(s)	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 10/7/2016 **Date Received:** 5/18/2018 **Final Report:** 6/1/2018	[redacted] **Fairfax Cryobank, Inc.** [redacted]

RESULT SUMMARY

Results: No clinically significant variant(s) detected

Gene(s) analyzed: *MCCC1, MCCC2, BCKDHB,* and *GJB2*

Interpretation: Screening for the presence of pathogenic variants in the *MCCC1, MCCC2, BCKDHB,* and *GJB2* genes which are associated with 3-methylcrotonyl-CoA carboxylase deficiency (*MCCC1*-related), 3-methylcrotonyl-CoA carboxylase deficiency (*MCCC2*-related), maple syrup urine disease, type 1b, and non-syndromic hearing loss (*GJB2*-related), respectively, 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 genes 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 Mount Sinai Genomics, Inc. 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.

Patient:	Donor 4824	DOB:	[redacted]	Lab #:	[redacted]

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.

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

This case has been reviewed and electronically signed by Ruth Kornreich, Ph.D., FACMG, 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	Analytical Detection Rate
Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	*BCKDHB*	Caucasian	1 in 433	56%	1 in 983	>95%
		Asian	1 in 163	57%	1 in 378	>95%
		Ashkenazi Jewish	1 in 97	>95%	1 in 1921	>95%
		Worldwide	1 in 327	72%	1 in 1165	>95%
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related) (AR) NM_020166.4	*MCCC1*	Caucasian	1 in 137	77%	1 in 592	88%
		Worldwide	1 in 147	75%	1 in 585	>95%
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related) (AR) NM_022132.4	*MCCC2*	Caucasian	1 in 112	59%	1 in 272	91%
		Worldwide	1 in 120	69%	1 in 385	>95%
Non-Syndromic Hearing Loss (*GJB2*-Related) (AR) NM_004004.5	*GJB2*	Caucasian	1 in 42	88%	1 in 343	>95%
		Asian	1 in 50	83%	1 in 289	>95%
		Ashkenazi Jewish	1 in 21	>95%	1 in 401	>95%
		Worldwide	1 in 43	82%	1 in 234	>95%

AR: Autosomal Recessive

Patient	Sample	Referring Doctor
Patient Name: 4824 Donor **Date of Birth:** [redacted] **Reference #:** P0269749 **Indication:** Carrier Testing **Test Type:** NGS single gene full sequencing test	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 10/7/2016 **Date Received:** 10/10/2016 **Final Report:** 10/24/2016	[redacted] **Fairfax Cryobank** [redacted] [redacted] [redacted] Fax: [redacted]

RESULTS

Results: No clinically significant variant(s) detected

Gene(s) analyzed: *BTD, PMM2, F11, PAH*

Interpretation: Screening for the presence of pathogenic variants in the *BTD, PMM2, F11,* and *PAH* genes which are associated with biotinidase deficiency, congenital disorder of glycosylation, type Ia, factor XI deficiency, and phenylalanine hydroxylase deficiency 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 Ozge Birsoy, Ph.D., FACMG, Assistant 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
Biotinidase Deficiency (AR)	*BTD*	Caucasian	1 in 12	74%	1 in 43
NM_000060.3		Hispanic	1 in 30	60%	1 in 74
Exons: 2-4; **Variants:** 12		Worldwide	1 in 25	60%	1 in 61
Congenital Disorder of Glycosylation, Type Ia (AR)	*PMM2*	Caucasian	1 in 42	82%	1 in 229
		Asian	1 in 449	20%	1 in 561
NM_000303.2		Ashkenazi Jewish	1 in 61	>95%	1 in 1201
Exons: **1-8;** Variants: **16**		Worldwide	1 in 124	76%	1 in 514
Factor XI Deficiency (AR)	*F11*	Caucasian	1 in 101	15%	1 in 119
NM_000128.3		Asian	1 in 163	35%	1 in 250
		Ashkenazi Jewish	1 in 11	>95%	1 in 201
Exons: 2-15; **Variants:** 11		Worldwide	1 in 92	31%	1 in 133
Phenylalanine Hydroxylase Deficiency (AR)	*PAH*	Caucasian	1 in 50	75%	1 in 197
		African	1 in 158	41%	1 in 267
NM_000277.1		Asian	1 in 78	41%	1 in 132
		Ashkenazi Jewish	1 in 225	17%	1 in 271
		Worldwide	1 in 65	66%	1 in 189
		Turkish	1 in 32	63%	1 in 85
		Irish	1 in 34	70%	1 in 111
		Sicilian	1 in 26	48%	1 in 49
		Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian and Moroccan	1 in 18	52%	1 in 36
Exons: 1-13; **Variants:** 29					

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