



## Donor 5007

### Genetic Testing Summary

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

Last Updated: 08/15/18

Donor Reported Ancestry: Irish, English, Spanish, Lebanese

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 (see attached)	Negative by gene sequencing in the CFTR gene	1/1250
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
<b>Special Testing</b>		
Autosomal Recessive Polycystic Kidney Disease (see attached)	Negative by gene sequencing in the PKHD1 gene	1/1428
Familial Dysautonomia (See attached)	Negative by gene sequencing in the IKBKAP gene	1/10,000
Gaucher Disease (see attached)	Negative for 6 mutations in the GBA gene	1/164

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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**Ordering Practice:**

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Practice Code: [REDACTED]  
Fairfax CryobankPhysician: [REDACTED]  
Report Generated: 2017-01-18

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**Donor 5007**

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DOB: [REDACTED]  
Gender: Male  
Ethnicity: European  
Procedure ID: 78414  
Kit Barcode: [REDACTED]  
Specimen: Sperm, #79231  
Specimen Collection: 2016-12-23  
Specimen Received: 2016-12-27  
Specimen Analyzed: 2017-01-18**TEST INFORMATION**Test: CarrierMap<sup>SEQ</sup> (Genotyping & Sequencing)  
Panel: Custom Panel  
Diseases Tested: 1  
Genes Tested: 1  
Genes Sequenced: 1

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**Partner Not Tested**

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**SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED**

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Donor 5007 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](http://recombine.com/diseases). To speak with a Genetic Counselor, call [855.OUR.GENES](tel: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.

**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

**Cystic Fibrosis (CFTR):** Mutations (150): ♂ Genotyping | c.1029delC, 1153\_1154insAT, c.1477delCA, c.1519\_1521delATC (p.507delI), c.1521\_1523delCTT (p.508delF), c.1545\_1546delTA (p.Y515Xfs), c.1585-1G>A, c.164+12T>C, c.1680-886A>G, c.1680-1G>A, c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.1818del84, c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATCAATCCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051\_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3039delC, c.3140-26A>G, c.325delTATinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C>T, c.3744delA, c.3773\_3774insT (p.L1258fs), c.442delA, c.489+1G>T, c.531delT, c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.803delA (p.N268fs), c.805\_806delAT (p.I269fs), c.933\_935delCTT (p.311delF), c.946delT, c.1645A>C (p.S549R), c.2128A>T (p.K710X), c.1000C>T (p.R334W), c.1013C>T (p.T338I), c.1364C>A (p.A455E), c.1477C>T (p.Q493X), c.1572C>A (p.C524X), c.1654C>T (p.Q552X), c.1657C>T (p.R553X), c.1721C>A (p.P574H), c.2125C>T (p.R709X), c.223C>T (p.R75X), c.2668C>T (p.Q890X), c.3196C>T (p.R1066C), c.3276C>G (p.Y1092X), c.3472C>T (p.R1158X), c.3484C>T (p.R1162X), c.349C>T (p.R117C), c.3587C>G (p.S1196X), c.3712C>T (p.Q1238X), c.3764C>A (p.S1255X), c.3909C>G (p.N1303K), c.1040G>A (p.R347H), c.1040G>C (p.R347P), c.1438G>T (p.G480C), c.1558G>T (p.V520F), c.1624G>T (p.G542X), c.1646G>A (p.S549N), c.1646G>T (p.S549I), c.1652G>A (p.G551D), c.1675G>A (p.A559T), c.1679G>C (p.R560T), c.178G>T (p.E60X), c.1865G>A (p.G622D), c.254G>A (p.G85E), c.271G>A (p.G91R), c.274G>T (p.E92X), c.3209G>A (p.R1070Q), c.3266G>A (p.W1089X), c.3454G>C (p.D1152H), c.350G>A (p.R117H), c.3611G>A (p.W1204X), c.3752G>A (p.S1251N), c.3846G>A (p.W1282X), c.3848G>T (p.R1283M), c.532G>A (p.G178R), c.988G>T (p.G330X), c.1090T>C (p.S364P), c.3302T>A (p.M1101K), c.617T>G (p.L206W), c.14C>T (p.P5L), c.19G>T (p.E7X), c.171G>A (p.W57X), c.313delA (p.I105fs), c.328G>C (p.D110H), c.580-1G>T, c.1055G>A (p.R352Q), c.1075C>A (p.Q359K), c.1079C>A (p.T360K), c.1647T>G (p.S549R), c.1976delA (p.N659fs), c.2290C>T (p.R764X), c.2737\_2738insG (p.Y913X), c.3067\_3072delATAGTG (p.I1023\_V1024delT), c.3536\_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940\_273+10250del21080bp (p.S18fs), c.4364C>G (p.S1455X), c.4003C>T (p.L1335F), c.2538G>A (p.W846X), c.200C>T (p.P67L), c.4426C>T (p.Q1476X), c.1116+1G>A, c.1986\_1989delAACT (p.T663R), c.2089\_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.263T>G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.658C>T (p.Q220X), c.868C>T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085-3367+260del17201, c.11C>A (p.S4X), c.3878\_3881delTATT (p.V1293fs), c.3700A>G (p.I1234V), c.416A>T (p.H139L), c.366T>A (p.Y122X), c.3767\_3768insC (p.A1256fs), c.613C>T (p.P205S), c.293A>G (p.Q98R), c.3731G>A (p.G1244E), c.535C>A (p.Q179K), c.3368-2A>G, c.455T>G (p.M152R), c.1610\_1611delAC (p.D537fs), c.3254A>G (p.H1085R), c.496A>G (p.K166E), c.1408\_1417delGTGATTATGG (p.V470fs), c.1585-8G>A, c.2909G>A (p.G970D), c.653T>A (p.L218X), c.1175T>G (p.V392G), c.3139\_3139+1delGG, c.3717+4A>G (IVS22+4A>G)

Sequencing | NM\_000492:1-27

## 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
Cystic Fibrosis	♂ African American: 1/62	69.99%	1/207
	♂ Ashkenazi Jewish: 1/23	96.81%	1/721
	♂ Asian: 1/94	65.81%	1/275
	♂ European: 1/25	94.96%	1/496
	♂ Hispanic American: 1/48	77.32%	1/212
	♂ Native American: 1/53	84.34%	1/338

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**Ordering Practice:**

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Practice Code: [REDACTED]  
Fairfax CryobankPhysician: [REDACTED]  
Report Generated: 2017-02-27

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DOB: [REDACTED]  
Gender: Male  
Ethnicity: European  
Procedure ID: 78414  
Kit Barcode: [REDACTED]  
Specimen: Sperm, #79231  
Specimen Collection: 2016-12-23  
Specimen Received: 2016-12-27  
Specimen Analyzed: 2017-02-27**TEST INFORMATION**Test: CarrierMap<sup>SEQ</sup> (Genotyping & Sequencing)  
Panel: Custom Panel  
Diseases Tested: 3  
Genes Tested: 3  
Genes Sequenced: 2

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**Partner Not Tested**

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## Diseases & Mutations Assayed

**Autosomal Recessive Polycystic Kidney Disease (PKHD1):** Mutations (40): ♂ Genotyping | c.5895insA (p.L1966fsX1969), c.9689delA (p.D3230fs), c.107C>T (p.T36M), c.1486C>T (p.R496X), c.10412T>G (p.V3471G), c.10658T>C (p.I3553T), c.10174C>T (p.Q3392X), c.9530T>C (p.I3177T), c.9053C>T (p.S3018F), c.8870T>C (p.I2957T), c.8011C>T (p.R2671X), c.6992T>A (p.I2331K), c.5221G>A (p.V1741M), c.4991C>T (p.S1664F), c.3761\_3762delCCinsG (p.A1254fs), c.2414C>T (p.P805L), c.664A>G (p.I222V), c.10036T>C (p.C3346R), c.383delC, c.4220T>G (p.L1407R), c.11612G>A (p.W3871X), c.5984A>G (p.E1995G), c.10637delT (p.V3546fs), c.3747T>G (p.C1249W), c.5750A>G (p.Q1917R), c.10865G>A (p.C3622Y), c.50C>T (p.A17V), c.8063G>T (p.C2688F), c.10402A>G (p.I3468V), c.1529delG (p.G510fs), c.657C>T (p.G219G), c.5513A>G (p.Y1838C), c.10856delA (p.K3619fs), c.5381-9T>G (IVS33-9T>G), c.3229-2A>C (IVS28-2A>C), c.10505A>T (p.E3502V), c.2269A>C (p.I757L), c.4165C>A (p.P1389T), c.10364delC (p.S3455fs), c.7350+653A>G (IVS46+653A>G) Sequencing | NM\_138694:2-67

**Familial Dysautonomia (IKBKAP):** Mutations (4): ♂ Genotyping | c.2204+6T>C, c.2741C>T (p.P914L), c.2087G>C (p.R696P), c.2128C>T (p.Q710X) Sequencing | NM\_003640:2-37

**Gaucher Disease (GBA):** Mutations (6): ♂ Genotyping | c.84\_85insG, c.1226A>G (p.N409S), c.1343A>T (p.D448V), c.1504C>T (p.R502C), c.1297G>T (p.V433L), c.1604G>A (p.R535H)



## 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
Autosomal Recessive Polycystic Kidney Disease	♂ Finnish: 1/45	84.21%	1/285
	♂ French: 1/71	62.50%	1/189
	♂ General: 1/71	37.11%	1/113
Familial Dysautonomia	♂ Ashkenazi Jewish: 1/31	>99%	<1/3,100
Gaucher Disease	♂ Ashkenazi Jewish: 1/15	87.16%	1/117
	♂ General: 1/112	31.60%	1/164
	♂ Spaniard: Unknown	44.29%	Unknown
	♂ Turkish: 1/236	59.38%	1/581