



## Donor 4982

### 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: Italian, Irish, Scottish

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 149 mutations in the CFTR gene	1/496
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
Standard testing attached- 22 diseases by genotyping	Negative for mutations tested	

\*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:

Practice Code: [REDACTED]  
 Fairfax Cryobank - [REDACTED]  
 [REDACTED]  
 [REDACTED]  
 Physician: [REDACTED]  
 Report Generated: 2016-04-05

4982 [REDACTED]

DOB: [REDACTED]  
 Gender: Male  
 Ethnicity:  
 Procedure ID: 48548  
 Kit Barcode: [REDACTED]  
 Method: Genotyping  
 Specimen: Blood, #50825  
 Specimen Collection: 2016-03-25  
 Specimen Received: 2016-03-28  
 Specimen Analyzed: 2016-04-05

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Partner Not Tested

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SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED


4982 [REDACTED] 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](http://www.recombine.com/diseases). To speak with a Genetic Counselor, call [855.OUR.GENES](tel:855.OUR.GENES).

♂ Male

Panel: Fairfax Cryobank Panel V2 , Diseases Tested: 22, Mutations Tested: 452, Genes Tested: 22, 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.

**Spinal Muscular Atrophy:** Spinal Muscular Atrophy: Carrier status for SMA is assessed via genotyping and via copy number analysis by qPCR. Some individuals with a normal number of SMN1 copies (2 copies) may carry both copies of the gene on the same allele/chromosome; this analysis is not able to detect these individuals. Thus, a normal SMN1 result significantly reduces but does not eliminate the risk of being a carrier. Additionally, SMA may be caused by non-deletion mutations in the SMN1 gene; CarrierMap tests for some, but not all, of these mutations via genotyping. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

























**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 &amp; Mutations Assayed

● High Impact ● Treatment Benefits ● X-Linked ● Moderate Impact

H	T	X	M	Disease	#	Mutations
<span style="color: red;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	Alpha Thalassemia	9	♂ Genotyping   SEA 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
<span style="color: red;">●</span>	<span style="color: green;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	Beta Thalassemia	84	♂ Genotyping   c.124_127delTTCT (p.F42Lfs), c.17_18delCT, c.20delA (p.E7Gfs), c.217insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.230delC, c.25_26delAA, c.315+1G>A, c.315+2T>C, c.316-197C>T, c.316-146T>G, c.315+745C>G, c.316-1G>A, c.316-1G>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.93-21G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c.-50A>C, c.-78a>g, c.-79a>g, c.-81a>g, c.52A>T (p.K18X), c.-137c>g, c.-138c>t, c.-151c>t, c.118C>T (p.Q40X), c.169G>C (p.G57R), c.295G>A (p.V99M), c.34G>A (p.V12I), c.415G>C (p.A139P), c.47G>A (p.W16X), c.48G>A (p.W16X), c.-80t>a, c.2T>C (p.M1T), c.75T>A (p.G25G), c.444+111A>G, c.-29g>a, c.68_74delAAGTTGG, c.92G>C (p.R31T), c.27_28insG, c.92+1G>T, c.92+1G>C, c.93-15T>G, c.93-1G>C, c.112delT, c.113G>A (p.W38X), c.114G>A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.287_288insA (p.L97fs), c.271G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.84_85insC (p.L29fs), c.59A>G (p.N20S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.2T>G (p.M1R), c.1A>G (p.M1V), c.-137c>t, c.-136c>g, c.-142c>t, c.-140c>t
<span style="color: red;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	Bloom Syndrome	24	♂ Genotyping   c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2407insT, c.557_559delCAA (p.S186X), c.1284G>A (p.W428X), c.1701G>A (p.W567X), c.1933C>T (p.Q645X), c.2528C>T (p.T843I), c.2695C>T (p.R899X), c.3107G>T (p.C1036F), c.2923delC (p.Q975K), c.3558+1G>T, c.3875-2A>G, c.2074+2T>A, c.2343_2344dupGA (p.781EfsX), c.380delC (p.127Tfs), c.3564delC (p.1188Dfs), c.4008delG (p.1336Rfs), c.947C>G (p.S316X), c.2193+1_2193+9del9, c.1642C>T (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.C120Hfs), c.4076+1delG, c.3281C>A (p.S1094X)
<span style="color: red;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	<span style="color: gray;">●</span>	Canavan Disease	8	♂ Genotyping   c.433-2A>G, c.854A>C (p.E285A), c.693C>A (p.Y231X), c.914C>A (p.A305E), c.71A>G (p.E24G), c.654C>A (p.C218X), c.2T>C (p.M1T), c.79G>A (p.G27R)

H	T	X	M	Disease	#	Mutations
				Cystic Fibrosis	149	<p>♂ 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&gt;A, c.164+12T&gt;C, c.1680-886A&gt;G, c.1680-1G&gt;A, c.1766+1G&gt;A, c.1766+1G&gt;T, c.1766+5G&gt;T, c.1818del84, c.1911delG, c.1923delCTCAAACTinsA, c.1973delGAAATTCATCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G&gt;A, c.273+1G&gt;A, c.273+3A&gt;C, c.274-1G&gt;A, c.2988+1G&gt;A, c.3039delC, c.3140-26A&gt;G, c.325delTATinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C&gt;T, c.3744delA, c.3773_3774insT (p.L1258fs), c.442delA, c.489+1G&gt;T, c.531delT, c.579+1G&gt;T, c.579+5G&gt;A (IVS4+5G&gt;A), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.933_935delCTT (p.311delF), c.946delT, c.1645A&gt;C (p.S549R), c.2128A&gt;T (p.K710X), c.1000C&gt;T (p.R334W), c.1013C&gt;T (p.T338I), c.1364C&gt;A (p.A455E), c.1477C&gt;T (p.Q493X), c.1572C&gt;A (p.C524X), c.1654C&gt;T (p.Q552X), c.1657C&gt;T (p.R553X), c.1721C&gt;A (p.P574H), c.2125C&gt;T (p.R709X), c.223C&gt;T (p.R75X), c.2668C&gt;T (p.Q890X), c.3196C&gt;T (p.R1066C), c.3276C&gt;G (p.Y1092X), c.3472C&gt;T (p.R1158X), c.3484C&gt;T (p.R1162X), c.349C&gt;T (p.R117C), c.3587C&gt;G (p.S1196X), c.3712C&gt;T (p.Q1238X), c.3764C&gt;A (p.S1255X), c.3909C&gt;G (p.N1303K), c.1040G&gt;A (p.R347H), c.1040G&gt;C (p.R347P), c.1438G&gt;T (p.G480C), c.1558G&gt;T (p.V520F), c.1624G&gt;T (p.G542X), c.1646G&gt;A (p.S549N), c.1646G&gt;T (p.S549I), c.1652G&gt;A (p.G551D), c.1675G&gt;A (p.A559T), c.1679G&gt;C (p.R560T), c.178G&gt;T (p.E60X), c.1865G&gt;A (p.G622D), c.254G&gt;A (p.G85E), c.271G&gt;A (p.G91R), c.274G&gt;T (p.E92X), c.3209G&gt;A (p.R1070Q), c.3266G&gt;A (p.W1089X), c.3454G&gt;C (p.D1152H), c.350G&gt;A (p.R117H), c.3611G&gt;A (p.W1204X), c.3752G&gt;A (p.S1251N), c.3846G&gt;A (p.W1282X), c.3848G&gt;T (p.R1283M), c.532G&gt;A (p.G178R), c.988G&gt;T (p.G330X), c.1090T&gt;C (p.S364P), c.3302T&gt;A (p.M1101K), c.617T&gt;G (p.L206W), c.14C&gt;T (p.P5L), c.19G&gt;T (p.E7X), c.171G&gt;A (p.W57X), c.313delA (p.I105fs), c.328G&gt;C (p.D110H), c.580-1G&gt;T, c.1055G&gt;A (p.R352Q), c.1075C&gt;A (p.Q359K), c.1079C&gt;A (p.T360K), c.1647T&gt;G (p.S549R), c.1976delA (p.N659fs), c.2290C&gt;T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delICCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940_273+10250del21080bp (p.S18fs), c.4056G&gt;C (p.Q1352H), c.4364C&gt;G (p.S1455X), c.4003C&gt;T (p.L1335F), c.2538G&gt;A (p.W846X), c.200C&gt;T (p.P67L), c.4426C&gt;T (p.Q1476X), c.1116+1G&gt;A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.263T&gt;G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.658C&gt;T (p.Q220X), c.868C&gt;T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085-3367+260del7201, c.11C&gt;A (p.S4X), c.3700A&gt;G (p.I1234V), c.416A&gt;T (p.H139L), c.366T&gt;A (p.Y122X), c.3767_3768insC (p.A1256fs), c.613C&gt;T (p.P205S), c.293A&gt;G (p.Q98R), c.3731G&gt;A (p.G1244E), c.535C&gt;A (p.Q179K), c.3368-2A&gt;G, c.455T&gt;G (p.M152R), c.1610_1611delAC (p.D537fs), c.3254A&gt;G (p.H1085R), c.496A&gt;G (p.K166E), c.1408_1417delGTGATTATGG (p.V470fs), c.1585-8G&gt;A, c.2909G&gt;A (p.G970D), c.653T&gt;A (p.L218X), c.1175T&gt;G (p.V392G), c.3139_3139+1delGG</p>
				Familial Dysautonomia	4	<p>♂ Genotyping   c.2204+6T&gt;C, c.2741C&gt;T (p.P914L), c.2087G&gt;C (p.R696P), c.2128C&gt;T (p.Q710X)</p>
				Familial Hyperinsulinism: Type 1: ABCC8 Related	10	<p>♂ Genotyping   c.3989-9G&gt;A, c.4159_4161delITC (p.1387delF), c.4258C&gt;T (p.R1420C), c.4477C&gt;T (p.R1493W), c.2147G&gt;T (p.G716V), c.4055G&gt;C (p.R1352P), c.560T&gt;A (p.V187D), c.4516G&gt;A (p.E1506K), c.2506C&gt;T (p.Q836X), c.579+2T&gt;A</p>
				Fanconi Anemia: Type C	8	<p>♂ Genotyping   c.456+4A&gt;T, c.67delG, c.37C&gt;T (p.Q13X), c.553C&gt;T (p.R185X), c.1661T&gt;C (p.L554P), c.1642C&gt;T (p.R548X), c.66G&gt;A (p.W22X), c.65G&gt;A (p.W22X)</p>
				Gaucher Disease	6	<p>♂ Genotyping   c.84_85insG, c.1226A&gt;G (p.N409S), c.1343A&gt;T (p.D448V), c.1504C&gt;T (p.R502C), c.1297G&gt;T (p.V433L), c.1604G&gt;A (p.R535H)</p>
				Glycogen Storage Disease: Type IA	13	<p>♂ Genotyping   c.376_377insTA, c.79delC, c.979_981delITC (p.327delF), c.1039C&gt;T (p.Q347X), c.247C&gt;T (p.R83C), c.724C&gt;T (p.Q242X), c.248G&gt;A (p.R83H), c.562G&gt;C (p.G188R), c.648G&gt;T, c.809G&gt;T (p.G270V), c.113A&gt;T (p.D38V), c.975delG (p.L326fs), c.724delC</p>

H	T	X	M	Disease	#	Mutations
●	○	○	○	Joubert Syndrome	2	♂ Genotyping   c.218G>T (p.R73L), c.218G>A (p.R73H)
●	●	○	○	Maple Syrup Urine Disease: Type 1B	6	♂ Genotyping   c.1114G>T (p.E372X), c.548G>C (p.R183P), c.832G>A (p.G278S), c.970C>T (p.R324X), c.487G>T (p.E163X), c.853C>T (p.R285X)
●	●	○	○	Maple Syrup Urine Disease: Type 3	8	♂ Genotyping   c.104_105insA, c.685G>T (p.G229C), c.214A>G (p.K72E), c.1081A>G (p.M361V), c.1123G>A (p.E375K), c.1178T>C (p.I393T), c.1463C>T (p.P488L), c.1483A>G (p.R495G)
●	○	○	○	Mucopolipidosis: Type IV	5	♂ Genotyping   c.-1015_788del6433, c.406-2A>G, c.1084G>T (p.D362Y), c.304C>T (p.R102X), c.244delC (p.L82fsX)
●	○	○	○	Nemaline Myopathy: NEB Related	1	♂ Genotyping   c.7434_7536del2502bp
●	○	○	○	Niemann-Pick Disease: Type A	6	♂ Genotyping   c.996delC, c.1493G>T (p.R498L), c.911T>C (p.L304P), c.1267C>T (p.H423Y), c.1734G>C (p.K578N), c.1493G>A (p.R498H)
●	●	○	○	Sickle-Cell Anemia	1	♂ Genotyping   c.20A>T (p.E7V)
●	○	○	○	Spinal Muscular Atrophy: SMN1 Linked	19	♂ Genotyping   DEL EXON 7, c.22_23insA, c.43C>T (p.Q15X), c.91_92insT, c.305G>A (p.W102X), c.400G>A (p.E134K), c.439_443delGAAGT, c.558delA, c.585_586insT, c.683T>A (p.L228X), c.734C>T (p.P245L), c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S), c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T qPCR   DEL EXON 7
●	○	○	○	Tay-Sachs Disease	76	♂ Genotyping   c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.532C>T (p.R178C), c.533G>A (p.R178H), c.805G>A (p.G269S), c.1510C>T (p.R504C), c.1496G>A (p.R499H), c.509G>A (p.R170Q), c.1003A>T (p.I335F), c.910_912delITC (p.305delF), c.749G>A (p.G250D), c.632T>C (p.F211S), c.629C>T (p.S210F), c.613delC, c.611A>G (p.H204R), c.598G>A (p.V200M), c.590A>C (p.K197T), c.571-1G>T, c.540C>G (p.Y180X), c.538T>C (p.Y180H), c.533G>T (p.R178L), c.508C>T (p.R170W), c.409C>T (p.R137X), c.380T>G (p.L127R), c.346+1G>C, c.116T>G (p.L39R), c.78G>A (p.W26X), c.1A>G (p.M1V), c.1495C>T (p.R499C), c.459+5G>A (IVS4+5G>A), c.1422-2A>G, c.535C>T (p.H179Y), c.1141delG (p.V381fs), c.796T>G (p.W266G), c.155C>A (p.S52X), c.426delT (p.F142fs), c.413-2A>G, c.570+3A>G, c.536A>G (p.H179R), c.1146+1G>A, c.736G>A (p.A246T), c.1302C>G (p.F434L), c.778C>T (p.P260S), c.1008G>T (p.Q336H), c.1385A>T (p.E462V), c.964G>A (p.D322N), c.340G>A (p.E114K), c.1432G>A (p.G478R), c.1178G>C (p.R393P), c.805+1G>C, c.1426A>T (p.R476X), c.623A>T (p.D208V), c.1537C>T (p.Q513X), c.1511G>T (p.R504L), c.1307_1308delTA (p.I436fs), c.571-8A>G, c.624_627delTCCT (p.D208fs), c.1211_1212delITG (p.L404fs), c.621T>G (p.D207E), c.1511G>A (p.R504H), c.1177C>T (p.R393X), c.2T>C (p.M1T), c.1292G>A (p.W431X), c.947_948insA (p.Y316fs), c.607T>G (p.W203G), c.1061_1063delITCT (p.F354_Y355delinsX), c.615delG (p.L205fs), c.805+2T>C, c.1123delG (p.E375fs), c.1121A>G (p.Q374R), c.1043_1046delTCAA (p.F348fs), c.1510delC (p.R504fs), c.1451T>C (p.L484P), c.964G>T (p.D322Y)
●	○	○	○	Usher Syndrome: Type 1F	7	♂ Genotyping   c.733C>T (p.R245X), c.2067C>A (p.Y684X), c.7C>T (p.R3X), c.1942C>T (p.R648X), c.1101delT (p.A367fsX), c.2800C>T (p.R934X), c.4272delA (p.L1425fs)
●	○	○	○	Usher Syndrome: Type 3	5	♂ Genotyping   c.144T>G (p.N48K), c.359T>A (p.M120K), c.300T>G (p.Y176X), c.634C>T (p.Q212X), c.221T>C (p.L74P)
●	○	○	○	Walker-Warburg Syndrome	1	♂ Genotyping   c.1167insA (p.F390fs)