



Donor 5139

Genetic Testing Summary

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

Last Updated: 08/14/18

Donor Reported Ancestry: Swedish, Finnish

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
Standard testing attached- 22 diseases by genotyping	Negative for mutations tested	
Special testing		
Niemann-Pick Disease Type C1	Negative by gene sequencing for the NPC1 gene	1/1961
Niemann-Pick Disease Type C2	Negative by gene sequencing for the NPC2 gene	1/20000
Nonsyndromic Hearing Loss and Deafness-GJB2 related	Negative by gene sequencing for the GJB2 gene	1/1695
Nonsyndromic Hearing Loss and Deafness-MYO15A related	Negative by gene sequencing for the MYO15A gene	<1/500

*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.

Ordering Practice:

Practice Code: [REDACTED]
Fairfax Cryobank - [REDACTED]
[REDACTED]
Physician: [REDACTED]
Report Generated: 2016-04-15

5139

DOB: [REDACTED]
Gender: Male
Ethnicity: European
Procedure ID: 49672
Kit Barcode: [REDACTED]
Method: Genotyping
Specimen: Blood, #52375
Specimen Received: 2016-04-08
Specimen Analyzed: 2016-04-15

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED


5139 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: 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 & Mutations Assayed

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

H	T	X	M	Disease	#	Mutations
●	○	○	○	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
●	●	○	○	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
●	○	○	○	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)
●	○	○	○	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>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.1923delCTCAAACTinsA, c.1973delGAAATTCATCTinsAGAAA, 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_3539delICCAA (p.T1179fs), c.3659delC (p.T1220fs), c.54-5940_273+10250del21080bp (p.S18fs), c.4056G>C (p.Q1352H), 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+260del7201, c.11C>A (p.S4X), 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</p>
				Familial Dysautonomia	4	<p>♂ Genotyping c.2204+6T>C, c.2741C>T (p.P914L), c.2087G>C (p.R696P), c.2128C>T (p.Q710X)</p>
				Familial Hyperinsulinism: Type 1: ABCC8 Related	10	<p>♂ Genotyping c.3989-9G>A, c.4159_4161delITC (p.1387delF), c.4258C>T (p.R1420C), c.4477C>T (p.R1493W), c.2147G>T (p.G716V), c.4055G>C (p.R1352P), c.560T>A (p.V187D), c.4516G>A (p.E1506K), c.2506C>T (p.Q836X), c.579+2T>A</p>
				Fanconi Anemia: Type C	8	<p>♂ Genotyping c.456+4A>T, c.67delG, c.37C>T (p.Q13X), c.553C>T (p.R185X), c.1661T>C (p.L554P), c.1642C>T (p.R548X), c.66G>A (p.W22X), c.65G>A (p.W22X)</p>
				Gaucher Disease	6	<p>♂ 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)</p>
				Glycogen Storage Disease: Type IA	13	<p>♂ Genotyping c.376_377insTA, c.79delC, c.979_981delITC (p.327delF), c.1039C>T (p.Q347X), c.247C>T (p.R83C), c.724C>T (p.Q242X), c.248G>A (p.R83H), c.562G>C (p.G188R), c.648G>T, c.809G>T (p.G270V), c.113A>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.1141 delG (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_1063delTCT (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.1101 delT (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)

Ordering Practice:

Practice Code: [REDACTED]
Fairfax Cryobank [REDACTED]
[REDACTED]
Physician: [REDACTED]
Report Generated: 2017-07-25
Report Updated: 2017-07-25

Donor 5139

DOB: [REDACTED]
Gender: Male
Ethnicity: European
Procedure ID: 49672
Kit Barcode: [REDACTED]
Specimen: Blood, #99844
Specimen Collection: 2017-06-27
Specimen Received: 2017-07-06
Specimen Analyzed: 2017-07-25

Partner Not Tested

TEST INFORMATION


Test: CarrierMap^{SEQ} (Genotyping & Sequencing)
Panel: Custom Panel
Diseases Tested: 4
Genes Tested: 4
Genes Sequenced: 4

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 5139 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. 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.

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. In the sequencing process, interval drop-out may occur, leading to intervals of insufficient coverage. Intervals of insufficient coverage will be reported if they occur.

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

Niemann-Pick Disease: Type C1 (NPC1): Mutations (14): ♂ Genotyping | c.2783A>C (p.Q928P), c.3263A>G (p.Y1088C), c.3467A>G (p.N1156S), c.3107C>T (p.T1036M), c.3182T>C (p.I1061T), c.2974G>C (p.G992R), c.2932C>T (p.R978C), c.2848G>A (p.V950M), c.2665G>A (p.V889M), c.2324A>C (p.Q775P), c.1133T>C (p.V378A), c.530G>A (p.C177Y), c.337T>C (p.C113R), c.2974G>T (p.G992W) Sequencing | NM_000271:1-25

Niemann-Pick Disease: Type C2 (NPC2): Mutations (11): ♂ Genotyping | c.58G>T (p.E20X), c.436C>T (p.Q146X), c.358C>T (p.P120S), c.352G>T (p.E118X), c.332delA (p.N1111fs), c.295T>C (p.C99R), c.199T>C (p.S67P), c.190+5G>A, c.141C>A (p.C47X), c.133C>T (p.Q45X), c.115G>A (p.V39M) Sequencing | NM_006432:1-5

Nonsyndromic Hearing Loss and Deafness: GJB2 Related (GJB2): Mutations (29): ♂ Genotyping | c.167delT, c.235delC, c.312_325delGAAGTTCATCAAGG, c.358delGAG (p.120delE), c.35delG, c.370C>T (p.Q124X), c.427C>T (p.R143W), c.109G>A (p.V37I), c.231G>A (p.W77X), c.551G>C (p.R184P), c.71G>A (p.W24X), c.229T>C (p.W77R), c.269T>C (p.L90P), c.617A>G (p.N206S), c.299_300delAT (p.H100Rfs), c.283G>A (p.V95M), c.134G>A (p.G45E), c.139G>T (p.E47X), c.35G>T, c.487A>G (p.M163V), c.250G>C (p.V84L), c.44A>C (p.K15T), c.334_335delAA (p.K112fs), c.516G>A (p.W172X), c.290_291insA (p.Y97fs), c.439G>A (p.E147K), c.-23+1G>A, c.550C>T (p.R184W), c.-259C>T Sequencing | NM_004004:1-2

Nonsyndromic Hearing Loss and Deafness: MYO15A Related (MYO15A): Mutations (10): ♂ Genotyping | c.453_455delCGAinsTGGACGCCTGGTCGGGCAGTGG (p.E152GfsX81), c.7801A>T (p.K2601X), c.6337A>T (p.I2113F), c.3866+1G>T, c.3313G>T (p.E1105X), c.3334delG (p.G1112fs), c.8148G>T (p.Q2716H), c.6331A>T (p.N2111Y), c.3685C>T (p.Q1229X), c.3866+1G>A Sequencing | NM_016239:2-65

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
Niemann-Pick Disease: Type C1	♂ Acadian: Unknown	>99%	Unknown
	♂ General: 1/194	15.60%	1/230
	♂ Japanese: Unknown	18.18%	Unknown
	♂ Portuguese: 1/194	25.00%	1/259
Niemann-Pick Disease: Type C2	♂ General: 1/194	75.00%	1/776
Nonsyndromic Hearing Loss and Deafness: GJB2 Related	♂ Ashkenazi Jewish: 1/20	95.83%	1/480
	♂ Chinese: 1/100	82.26%	1/564
	♂ European: 1/53	82.47%	1/302
	♂ Ghanaian: Unknown	90.91%	Unknown
	♂ Indian: Unknown	66.98%	Unknown
	♂ Israeli: 1/16	93.10%	1/232
	♂ Japanese: 1/75	75.00%	1/300
	♂ Roma: Unknown	>99%	Unknown
	♂ United States: 1/34	45.22%	1/62
Nonsyndromic Hearing Loss and Deafness: MYO15A Related	♂ Balinese: 1/6	>99%	<1/600
	♂ Pakistani: 1/77	24.00%	1/101