Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4604

Genetic Testing Summary

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

Last Updated: 11/07/23

Donor Reported Ancestry: German, Russian, Hungarian, Italian 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 130 mutations in the CFTR gene	1/476
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Standard testing attached- 21 diseases by genotyping	Negative for mutations tested	
Special Testing		
D-Bifunctional Protein Deficiency	Negative for 6 mutations in the HSD17B4 gene	1/259
ARSA- related disorder	Carrier: ARSA-related disorder (ARSA)	Partner testing is recommended before using this donor. Only if the egg source is also a carrier is a child at increased risk. Chance of being a carrier is 1/100 in general population.

*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: 2015-09-08
Report Updated: 2015-09-08

Donor 4604

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 29138
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Sperm, #30380
Specimen Collection: 2015-08-25
Specimen Received: 2015-08-28
Specimen Analyzed: 2015-09-08

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4604 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 , Diseases Tested: 21, Mutations Tested: 382, Genes Tested: 22, Null Calls: 0

Panel: D-Bifunctional Protein Deficiency , Diseases Tested: 1, Mutations Tested: 6, 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.

Spinal Muscular Atrophy: Spinal Muscular Atrophy is tested for via an Identity-by-State shared haplotype comparison algorithm. Detection is limited to haplotypes within our library of known carriers of the most common mutation (deletion of Exon 7).

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	10	♂ Genotyping | SEA deletion, 11.1kb 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	83	♂ Genotyping | 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.a-78g, c.a-79g, c.a-81g, c.A52T (p.K18X), c.c-137g, c.c-138t, c.c-151t, c.C118T (p.Q40X), c.G169C (p.G57R), c.G295A (p.V99M), c.G34A (p.V12I), c.G415C (p.A139P), c.G47A (p.W16X), c.G48A (p.W16X), c.t-80a, c.T2C (p.M1T), c.T75A (p.G25G), c.444+111A>G, c.g-29a, c.68_74delAAGTTGG, c.G92C (p.R31T), c.27_28insG, c.92+1G>T, c.92+1G>C, c.93-15T>G, c.93-1G>C, c.112delT, c.G113A (p.W38X), c.G114A (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.316-106C>T, 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.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.c-137t, c.c-136g, c.c-142t, c.c-140t
●	○	○	○	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.C2528T (p.T843I), c.C2695T (p.R899X), c.G3107T (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.C947G (p.S316X), c.2193+1_2193+9del9, c.C1642T (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.Cys120Hisfs), c.4076+1delG, c.C3281A (p.S1094X)
●	○	○	○	Canavan Disease	8	♂ Genotyping | c.433-2A>G, c.A854C (p.E285A), c.C693A (p.Y231X), c.C914A (p.A305E), c.A71G (p.E24G), c.C654A (p.C218X), c.T2C (p.M1T), c.G79A (p.G27R)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Cystic Fibrosis	130	♂ Genotyping | c.1029delC, 1153_1154insAT, 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.1973delGAAATTCAATCCTinsAGAAA, 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.A1645C (p.S549R), c.A2128T (p.K710X), c.C1000T (p.R334W), c.C1013T (p.T338I), c.C1364A (p.A455E), c.C1477T (p.Q493X), c.C1572A (p.C524X), c.C1654T (p.Q552X), c.C1657T (p.R553X), c.C1721A (p.P574H), c.C2125T (p.R709X), c.C223T (p.R75X), c.C2668T (p.Q890X), c.C3196T (p.R1066C), c.C3276G (p.Y1092X), c.C3472T (p.R1158X), c.C3484T (p.R1162X), c.C349T (p.R117C), c.C3587G (p.S1196X), c.C3712T (p.Q1238X), c.C3764A (p.S1255X), c.C3909G (p.N1303K), c.G1040A (p.R347H), c.G1040C (p.R347P), c.G1438T (p.G480C), c.G1624T (p.G542X), c.G1646A (p.S549N), c.G1646T (p.S549I), c.G1652A (p.G551D), c.G1675A (p.A559T), c.G1679C (p.R560T), c.G178T (p.E60X), c.G1865A (p.G622D), c.G254A (p.G85E), c.G271A (p.G91R), c.G274T (p.E92X), c.G3209A (p.R1070Q), c.G3266A (p.W1089X), c.G3454C (p.D1152H), c.G350A (p.R117H), c.G3611A (p.W1204X), c.G3752A (p.S1251N), c.G3846A (p.W1282X), c.G3848T (p.R1283M), c.G532A (p.G178R), c.G988T (p.G330X), c.T1090C (p.S364P), c.T3302A (p.M1101K), c.T617G (p.L206W), c.C14T (p.P5L), c.G19T (p.E7X), c.G171A (p.W57X), c.313delA (p.I105fs), c.G328C (p.D110H), c.580-1G>T, c.G1055A (p.R352Q), c.C1075A (p.Q359K), c.C1079A (p.T360K), c.T1647G (p.S549R), c.1976delA (p.N659fs), c.C2290T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.G3808A (p.D1270N), c.G4056C (p.Q1352H), c.C4364G (p.S1455X), c.C4003T (p.L1335F), c.G2538A (p.W846X), c.C200T (p.P67L), c.C4426T (p.Q1476X), c.1116+1G>A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.T263G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.C658T (p.Q220X), c.C868T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085-3367+260del7201, c.C11A (p.S4X), c.A3700G (p.I1234V), c.A416T (p.H139L), c.T366A (p.Y122X)
●	○	○	○	D-Bifunctional Protein Deficiency	6	♂ Genotyping | c.G46A (p.G16S), c.63G>T (p.L21F), c.422_423delAG, c.652G>T (p.V218L), c.1369A>T (p.N457Y), c.1369A>G (p.N457D)
●	○	○	○	Familial Dysautonomia	4	♂ Genotyping | c.2204+6T>C, c.C2741T (p.P914L), c.G2087C (p.R696P), c.C2128T (p.Q710X)
●	○	○	○	Familial Hyperinsulinism: Type 1: ABCC8 Related	10	♂ Genotyping | c.3989-9G>A, c.4159_4161delTTC (p.1387delF), c.C4258T (p.R1420C), c.C4477T (p.R1493W), c.G2147T (p.G716V), c.G4055C (p.R1352P), c.T560A (p.V187D), c.4516G>A (p.E1506K), c.C2506T (p.Q836X), c.579+2T>A
●	●	○	○	Fanconi Anemia: Type C	8	♂ Genotyping | c.456+4A>T, c.67delG, c.C37T (p.Q13X), c.C553T (p.R185X), c.T1661C (p.L554P), c.C1642T (p.R548X), c.G66A (p.W22X), c.G65A (p.W22X)
●	●	○	○	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)
●	●	○	○	Glycogen Storage Disease: Type IA	13	♂ Genotyping | c.376_377insTA, c.79delC, c.979_981delTTC (p.327delF), c.C1039T (p.Q347X), c.C247T (p.R83C), c.C724T (p.Q242X), c.G248A (p.R83H), c.G562C (p.G188R), c.G648T, c.G809T (p.G270V), c.A113T (p.D38V), c.975delG (p.L326fs), c.724delC
●	○	○	○	Joubert Syndrome	1	♂ Genotyping | c.G35T (p.R12L)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Maple Syrup Urine Disease: Type 1B	6	♂ Genotyping | c.G1114T (p.E372X), c.G548C (p.R183P), c.G832A (p.G278S), c.C970T (p.R324X), c.G487T (p.E163X), c.C853T (p.R285X)
●	●	○	○	Maple Syrup Urine Disease: Type 3	8	♂ Genotyping | c.104_105insA, c.G685T (p.G229C), c.A214G (p.K72E), c.A1081G (p.M361V), c.G1123A (p.E375K), c.T1178C (p.I393T), c.C1463T (p.P488L), c.A1483G (p.R495G)
●	○	○	○	Mucolipidosis: Type IV	4	♂ Genotyping | c.406-2A>G, c.G1084T (p.D362Y), c.C304T (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.G1493T (p.R498L), c.T911C (p.L304P), c.C1267T (p.H423Y), c.G1734C (p.K578N), c.1493G>A (p.R498H)
●	○	○	○	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
●	○	○	○	Tay-Sachs Disease	30	♂ Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.C532T (p.R178C), c.G533A (p.R178H), c.G805A (p.G269S), c.C1510T (p.R504C), c.G1496A (p.R499H), c.G509A (p.R170Q), c.A1003T (p.I335F), c.910_912delTTC (p.305delF), c.G749A (p.G250D), c.T632C (p.F211S), c.C629T (p.S210F), c.613delC, c.A611G (p.H204R), c.G598A (p.V200M), c.A590C (p.K197T), c.571-1G>T, c.C540G (p.Y180X), c.T538C (p.Y180H), c.G533T (p.R178L), c.C508T (p.R170W), c.C409T (p.R137X), c.T380G (p.L127R), c.346+1G>C, c.T116G (p.L39R), c.G78A (p.W26X), c.A1G (p.M1V)
●	○	○	○	Usher Syndrome: Type 1F	6	♂ Genotyping | c.C733T (p.R245X), c.2067C>A (p.Y684X), c.C7T (p.R3X), c.C1942T (p.R648X), c.2800C>T (p.R934X), c.4272delA (p.L1425fs)
●	○	○	○	Usher Syndrome: Type 3	4	♂ Genotyping | c.T144G (p.N48K), c.T359A (p.M120K), c.300T>G (p.Y176X), c.C634T (p.Q212X)
●	○	○	○	Walker-Warburg Syndrome	1	♂ Genotyping | c.1167insA (p.F390fs)

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fulgent

[redacted]

Patient Information
4604, Donor
DOB: [redacted]
Sex: M
MR#: 4604
FD Patient#: [redacted]

Accession:
[redacted]
FD Test#: [redacted]
Order#: [redacted]
Ext Test#: [redacted]
Ext Order#: [redacted]
Specimen Type: DNA
Collected: Sep 21, 2023
Received Date: Sep 28, 2023
Authorized Date: Oct 03, 2023

Physician:
Seitz, Suzanne
ATTN: Seitz, Suzanne
Fairfax Cryobank
3015 Williams Drive
Fairfax, VA 22031 US

Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Oct 31, 2023**

FINAL Report

TEST PERFORMED

Known Mutation / Site-Specific Testing
(1 Variant)

RESULTS:

1 VARIANT DETECTED

Gene Info		Variant Info			
GENE	INHERITANCE	VARIANT	ZYGOSITY	CLASSIFICATION	RESULTS
ARSA NM_000487.6	Autosomal Recessive	c.109_116del p.Asp37Leufs*36	Heterozygous	**Pathogenic**	**DETECTED**

INTERPRETATION:

Notes and Recommendations:

- Genetic counseling is recommended.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.

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The targeted test results for the specimen reported here indicate that NM_000487.6:c.109_116del (p.Asp37Leufs*36) in the ARSA gene was **detected** (heterozygous) in this individual. Biallelic mutations in ARSA have been associated with metachromatic leukodystrophy (MLD), which is also known as arylsulfatase A deficiency (PubMed: 20301309; OMIM: 250100). Metachromatic leukodystrophy is a progressive and severe demyelinating disease which is inherited in an autosomal recessive fashion and caused by arylsulfatase A (ARSA) deficiency (OMIM: 250100). Current ARSA enzymatic activity assays cannot distinguish between MLD and ARSA pseudodeficiency, which is also associated with reduced ARSA enzyme activity but does not cause MLD (PubMed: 20301309, 9668161). ARSA pseudodeficiency alleles have been reported with high carrier frequencies in certain populations (PubMed: 21695197, 20301309, 2565866). This frameshift variant is the result of the deletion of 8 base pairs, which leads to an out-of-frame transcript and the introduction of a premature stop codon. The introduced stop codon is predicted to be located at least 50 nucleotides upstream of the canonical donor splice site of the penultimate exon and is consistent with the resulting transcript being targeted for nonsense mediated decay (PubMed: 25741868, 27618451, 11532962, 18066079). There is sufficient evidence that loss of function in this gene is a known disease mechanism for metachromatic leukodystrophy (PubMed: 9090526, 20301309). This variant was previously observed as compound heterozygous with a second disease mutation in three patients with metachromatic leukodystrophy (PubMed: 9090526, 25965562). This variant has been observed at a frequency of less than 0.01% (2/165594 alleles) across the entire Broad dataset (individuals without severe childhood onset disease). The laboratory considers this variant to be pathogenic.

GENES TESTED:

Known Mutation Test

1 gene tested.

ARSA

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). Methodology - Next Generation Sequencing (NGS), Sanger Sequencing, quantitative PCR (qPCR), repeat-primed PCR (rpPCR) or multiplex ligation-dependent probe amplification (MLPA) is selected by the laboratory to provide optimal results.

If NGS was performed:
DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37 / hg19), variants were detected in regions of at least 10x coverage. The known mutation genomic loci requested are evaluated for the presence or absence of variation compared to the human genome reference sequence. Bioinformatics: Fulgent Germline Pipeline v2019.1 or v2019.2 was used to generate variant calls for this test.

If Sanger Sequencing was performed:
DNA was amplified for the target region and sequenced bi-directionally using an ABI 3730XL instrument. The data was analyzed against the reference gene sequence and the known variant position as requested.

If qPCR was performed:
DNA was amplified for the target region and quantified using a QuantStudio 6 instrument. The data is compared to control genes and control individuals for the targets as requested.

If rpPCR was performed:
This analysis is performed by repeat-primed PCR (rpPCR) and amplicon length analysis. The scope of this assay is limited to repeat expansion analysis of the specified gene. Gene sequencing and deletion/duplication analysis are not included in this assay. This analysis does not include methylation studies.

If MLPA was performed:
DNA was amplified for the target regions and quantified using probesets using kits from MRC-Holland and an ABI 3730 instrument.The data is compared to control genes and control individuals for the targets as requested.

LIMITATIONS:

All laboratory tests have limitations. These results assume that the specimen received in the laboratory belongs to the named

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individual and that no mix-up or co-mingling of specimens has occurred. Positive results do not imply that there are no other pathogenic alterations in the patient's genome, and negative results do not rule out a genetic cause for the indication for testing. This assay assumes that any stated familial relationships are accurate. This assay is not designed or validated for the detection of somatic mosaicism or somatic mutations. This assay will only analyze the variant(s) requested. It is possible that the nomenclature for the variants tested may be different from the requested variants due to nomenclature differences in different isoforms of the gene. It is very important to provide us the isoform (NM number) of the gene for every variant to be tested. Result interpretation assumes that the human reference sequences are correct at the queried loci. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (https://www.genenames.org) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the collected information available at the time of reporting; additional information may exist in the future which will not be represented. Rarely, due to systematic chemical or computational issues, or human error, DNA variants may be missed. If a positive familial control specimen is not provided or available, rare errors may occur.

SIGNATURE:

Geetu Mendiratta-Vij, PhD, FACMG on **Oct 31, 2023 12:20 PM**
Electronically signed

DISCLAIMER:

This test was developed and its performance characteristics determined by**Fulgent Genetics**. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or **info@fulgentgenetics.com**. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.