Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 4282

Genetic Testing Summary

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

Last Updated: 06/16/23

Donor Reported Ancestry: Georgian 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 for 99 variants in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Genetic Disease panel- 101 disease performed in 2012- see attached.	Negative for other genes genotyped.	
Special Testing		
Genes: HEXA, DHCR7	Negative by genotyping	See attached
Genes: IDUA, PAH	Negative by gene sequencing	See attached

*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.

Counsyl

Results Re ent	Male	Female
Fairfax Cryobank [redacted]	Name: DONOR 4282 DOB: [redacted] Ethnicity: Mixed or Other Caucasian Sample Type: EDTA Blood Date of Collection: 09/27/2012 Date Received: 10/01/2012 Barcode: [redacted]	Not tested
Report Date: 10/09/2012	Indication: Egg or Sperm Donor	

Counsyl Test Results (Egg or Sperm Donor)

The Counsyl test (**Universal Panel**) uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual for **399 variants** associated with **101 diseases**. This report indicates which mutations, if any, were detected for each mutation panel. Because only select mutations are tested, the percentage of carriers detected varies by ethnicity. A full list of mutations tested is given on page 2. A negative test result does not eliminate the possibility that the individual is a carrier. Interpretation is given as an estimate of the risk of conceiving a child affected with a disease, which is based on reported ethnicity, the test results, and an assumption of no family history.*

DONOR 4282

DONOR 4282's DNA test shows that he is not a carrier of any disease-causing mutation tested.

Partner

The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Reproductive Risk Summary

No increased reproductive risks to highlight. Please refer to the following pages for detailed information about the results.

Clinical notes:

- The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional testing and genetic counseling.
- Individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and may also benefit from carrier testing by CBC and hemoglobin electrophoresis or HPLC. *ACOG Practice Bulletin No. 78. Obstet Gynecol 2007;109:229-37.*
- Additional Tay-Sachs disease testing can be performed using a biochemical assay, which has an excellent detection rate regardless of ancestry. *Gross et al. Genet Med 2008:10(1):54-56.*
- If necessary, patients can discuss residual risks with their physician or a genetic counselor. To schedule a complementary appointment to speak with a genetic counselor about these results, please visit counsyl.com/counseling/.

Lab Directors:

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*Limitations: In an unknown number of cases, nearby genetic variants may interfere with mutation detection. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately. Other possible sources of diagnostic error include sample mix-up, trace contamination, and technical errors. The reproductive risk summary is provided as an aid to genetic counseling. Inaccurate reporting of ethnicity may cause errors in risk calculation. For the purposes of risk calculations, it is assumed that mutations within the same gene are on different chromosomes.

This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's workup. CLIA Number: #05D1102604.

Counsyl

Male

Name: DONOR 4282
DOB: [redacted]

Female

Not tested

Mutations Tested

ABCC8-Related Hyperinsulinism - Gene: ABCC8. Variants (3): F1388del, V187D, 3992-9G>A. Detection rate: Mixed or Other Caucasian <10%.

Achromatopsia - Gene: CNGB3. Variants (3): R403Q, 819_826del8, T383fs. Detection rate: Mixed or Other Caucasian 62%.

Alkaptonuria - Gene: HGD. Variants (11): G161R, G270R, P230S, S47L, V300G, M368V, IVS1-1G>A, IVS5+1G>A, G152fs, R58fs, 1111_1112insC. Detection rate: Mixed or Other Caucasian 80%.

Alpha-1 Antitrypsin Deficiency - Gene: SERPINA1. Variants (1): Z allele. Detection rate: Mixed or Other Caucasian 95%.

Alpha-Mannosidosis - Gene: MAN2B1. Variants (1): R750W. Detection rate: Mixed or Other Caucasian 32%.

Andermann Syndrome - Gene: SLC12A6. Variants (2): Thr813fsX813, R1011X. Detection rate: Mixed or Other Caucasian <10%.

ARSACS - Gene: SACS. Variants (2): 6594delT, 5254C>T. Detection rate: Mixed or Other Caucasian <10%.

Aspartylglycosaminuria - Gene: AGA. Variants (1): C163S. Detection rate: Mixed or Other Caucasian <10%.

Ataxia With Vitamin E Deficiency - Gene: TTPA. Variants (1): 744delA. Detection rate: Mixed or Other Caucasian <10%.

Ataxia-Telangiectasia - Gene: ATM. Variants (8): R35X, Q1970X, 7517del4, 5762ins137, 2546_2548del, 3245ATC>TGAT, K1192K, E1978X. Detection rate: Mixed or Other Caucasian 65%.

Autosomal Recessive Polycystic Kidney Disease - Gene: PKHD1. Variants (4): Leu1965fs, T36M, R496X, V3471G. Detection rate: Mixed or Other Caucasian 18%.

Bardet-Biedl Syndrome, BBS1-Related - Gene: BBS1. Variants (1): M390R. Detection rate: Mixed or Other Caucasian 79%.

Bardet-Biedl Syndrome, BBS10-Related - Gene: BBS10. Variants (1): C91fs. Detection rate: Mixed or Other Caucasian 46%.

Biotinidase Deficiency - Gene: BTD. Variants (4): G98:d7i3, D252G, Q456H, R538C. Detection rate: Mixed or Other Caucasian 45%.

Bloom Syndrome - Gene: BLM. Variants (1): 2281del6ins7. Detection rate: Mixed or Other Caucasian <10%.

Canavan Disease - Gene: ASPA. Variants (4): E285A, Y231X, A305E, IVS2-2A>G. Detection rate: Mixed or Other Caucasian 53%.

Carnitine Palmitoyltransferase IA Deficiency - Gene: CPT1A. Variants (1): G710E. Detection rate: Mixed or Other Caucasian <10%.

Carnitine Palmitoyltransferase II Deficiency - Gene: CPT2. Variants (3): Q413fs, S113L, R124X. Detection rate: Mixed or Other Caucasian 80%.

Cartilage-Hair Hypoplasia - Gene: RMRP. Variants (1): g.70A>G. Detection rate: Mixed or Other Caucasian 48%.

Choroideremia - Gene: CHM. Variants (1): IVS13+2dupT. Detection rate: Mixed or Other Caucasian <10%.

Citrullinemia Type 1 - Gene: ASS1. Variants (2): IVS6-2A>G, G390R. Detection rate: Mixed or Other Caucasian 20%.

CLN3-Related Neuronal Ceroid Lipofuscinosis - Gene: CLN3. Variants (1): 461_677del. Detection rate: Mixed or Other Caucasian 96%.

CLN5-Related Neuronal Ceroid Lipofuscinosis - Gene: CLN5. Variants (1): 2467AT. Detection rate: Mixed or Other Caucasian <10%.

Cohen Syndrome - Gene: VPS13B. Variants (1): 3348_3349delCT. Detection rate: Mixed or Other Caucasian <10%.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Variants (4): V231M, F119L, R141H, P113L. Detection rate: Mixed or Other Caucasian 72%.

Congenital Disorder of Glycosylation Type Ib - Gene: MPI. Variants (1): R295H. Detection rate: Mixed or Other Caucasian <10%.

Congenital Finnish Nephrosis - Gene: NPHS1. Variants (2): 121_122del, R1109X. Detection rate: Mixed or Other Caucasian <10%.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Variants (1): 143-1G>C. Detection rate: Mixed or Other Caucasian <10%.

Cystic Fibrosis - Gene: CFTR. Variants (99): G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, E92X, Y122X, G178R, R347H, Q493X, V520F, S549N, P574H, M1101K, D1152H, 2143delT, 394delTT, 444delA, 1078delT, 3876delA, 3905insT, 1812-1G>A, 3272-26A>G, 2183AA>G, S549R(A>C), R117C, L206W, G330X, T338I, R352Q, S364P, G480C, C524X, S549R(T>G), Q552X, A559T, G622D, R709X, K710X, R764X, Q890X, R1066C, W1089X, Y1092X, R1158X, S1196X, W1204X(c.3611G>A), Q1238X, S1251N, S1255X, 3199del6, 574delA, 663delT, 935delA, 936delTA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2108delA, 3171delC, 3667del4, 3791delC, 1288insTA, 2184insA, 2307insA, 2869insG, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1898+1G>T, 1898+5G>T, 3120G>A, 457TAT>G, 3849+4A>G, Q359K/T360K. Detection rate: Mixed or Other Caucasian 91%.

Cystinosis - Gene: CTNS. Variants (4): 57 kb deletion, 537del21, W138X, L158P. Detection rate: Mixed or Other Caucasian 67%.

D-Bifunctional Protein Deficiency - Gene: HSD17B4. Variants (2): G16S, N457Y. Detection rate: Mixed or Other Caucasian 35%.

Factor XI Deficiency - Gene: F11. Variants (4): E117X, F283L, IVS14+1G>A, IVS14del14. Detection rate: Mixed or Other Caucasian <10%.

Familial Dysautonomia - Gene: IKBKAP. Variants (2): IVS20+6T>C, R696P. Detection rate: Mixed or Other Caucasian <10%.

Familial Mediterranean Fever - Gene: MEFV. Variants (4): M694V, V726A, M680I, M694I. Detection rate: Mixed or Other Caucasian <10%.

Fanconi Anemia Type C - Gene: FANCC. Variants (3): IVS4+4A>T, 322delG, R548X. Detection rate: Mixed or Other Caucasian 54%.

Galactosemia - Gene: GALT. Variants (8): S135L, Q188R, F171S, L195P, K285N, IVS2-2A>G, T138M, Y209C. Detection rate: Mixed or Other Caucasian 80%.

Gaucher Disease - Gene: GBA. Variants (10): N370S, L444P, 84GG, IVS2+1G>A, V394L, R496H, D409H, D409V, R463C, R463H. Detection rate: Mixed or Other Caucasian 60%.

GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness - Gene: GJB2. Variants (7): 35delG, 167delT, 235delC, E120del, W24X, W77R, L90P. Detection rate: Mixed or Other Caucasian 79%.

Glutaric Acidemia Type 1 - Gene: GCDH. Variants (1): R402W. Detection rate: Mixed or Other Caucasian 40%.

Glycogen Storage Disease Type Ia - Gene: G6PC. Variants (7): R83C, Q347X, Q27fsdelC, 459insTA, R83H, G188R, Q242X. Detection rate: Mixed or Other Caucasian 61%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Variants (2): 1211delCT, G339C. Detection rate: Mixed or Other Caucasian 46%.

Glycogen Storage Disease Type III - Gene: AGL. Variants (3): 1484delT, Q6X, 17delAG. Detection rate: Mixed or Other Caucasian 45%.

Glycogen Storage Disease Type V - Gene: PYGM. Variants (4): R49X, G204S, 708/709del, W797R. Detection rate: Mixed or Other Caucasian 80%.

GRACILE Syndrome - Gene: BCS1L. Variants (1): S78G. Detection rate: Mixed or Other Caucasian <10%.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Variants (28): Hb S, K17X, Q39X, Phe41fs, Ser9fs, IVS-II-654, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-I-1(G>A), -88C>T, -28A>G, -29A>G, Lys8fs, Phe71fs, IVS-II-849(A>C), IVS-II-849(A>G), Gly24 T>A, -87C>G, Hb C, W15X, Gly16fs, Glu6fs, Hb E, Hb D-Punjab, Hb O-Arab. Detection rate: Mixed or Other Caucasian 83%.

Hereditary Fructose Intolerance - Gene: ALDOB. Variants (3): A149P, N334K, A174D. Detection rate: Mixed or Other Caucasian 75%.

Hereditary Thymine-Uraciluria - Gene: DPYD. Variants (1): IVS14+1G>A. Detection rate: Mixed or Other Caucasian 52%.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related - Gene: LAMA3. Variants (1): R650X. Detection rate: Mixed or Other Caucasian <10%.

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related - Gene: LAMB3. Variants (3): R42X, Q243X, R635X. Detection rate: Mixed or Other Caucasian 48%.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related - Gene: LAMC2. Variants (1): R95X. Detection rate: Mixed or Other Caucasian <10%.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Variants (9): 1278insTATC, IVS12+1G>C, G269S, IVS9+1G>A, R178H, IVS7+1G>A, 7.6kb del, G250D, R170W. Detection rate: Mixed or Other Caucasian 23%.

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency - Gene: CBS. Variants (1): G307S. Detection rate: Mixed or Other Caucasian 14%.

Hurler Syndrome - Gene: IDUA. Variants (2): W402X, Q70X. Detection rate: Mixed or Other Caucasian 67%.

Hypophosphatasia, Autosomal Recessive - Gene: ALPL. Variants (4): 1559delT, F310L, D361V, E174K. Detection rate: Mixed or Other Caucasian 30%.

Inclusion Body Myopathy 2 - Gene: GNE. **Variants (2):** M712T, V572L. **Detection rate:** Mixed or Other Caucasian <10%.
Isovaleric Acidemia - Gene: IVD. **Variants (1):** A311V. **Detection rate:** Mixed or Other Caucasian 47%.
Joubert Syndrome 2 - Gene: TMEM216. **Variants (1):** 35G>T. **Detection rate:** Mixed or Other Caucasian <10%.
Krabbe Disease - Gene: GALC. **Variants (2):** Ex11-17del, T513M. **Detection rate:** Mixed or Other Caucasian 58%.
Limb-Girdle Muscular Dystrophy Type 2D - Gene: SGCA. **Variants (1):** R77C. **Detection rate:** Mixed or Other Caucasian 32%.
Limb-Girdle Muscular Dystrophy Type 2E - Gene: SGCB. **Variants (1):** S114F. **Detection rate:** Mixed or Other Caucasian 12%.
Lipoamide Dehydrogenase Deficiency - Gene: DLD. **Variants (2):** 105insA, G229C. **Detection rate:** Mixed or Other Caucasian <10%.
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency - Gene: HADHA. **Variants (1):** E474Q. **Detection rate:** Mixed or Other Caucasian 87%.
Maple Syrup Urine Disease Type 1B - Gene: BCKDHB. **Variants (3):** R183P, G278S, E372X. **Detection rate:** Mixed or Other Caucasian <10%.
Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. **Variants (2):** K304E, Y42H. **Detection rate:** Mixed or Other Caucasian 78%.
Megalencephalic Leukoencephalopathy With Subcortical Cysts - Gene: MLC1. **Variants (4):** 135InsC, c.176G>A, c.278C>T, IVS2-10T>A. **Detection rate:** Mixed or Other Caucasian 13%.
Metachromatic Leukodystrophy - Gene: ARSA. **Variants (5):** P426L, IVS2+1G>A, c.1204+1G>A, I179S, p.Thr409Ile. **Detection rate:** Mixed or Other Caucasian 53%.
Mucolipidosis IV - Gene: MCOLN1. **Variants (2):** 511_6944del, IVS3-2A>G. **Detection rate:** Mixed or Other Caucasian <10%.
Muscle-Eye-Brain Disease - Gene: POMGNT1. **Variants (1):** IVS17+1G>A. **Detection rate:** Mixed or Other Caucasian 75%.
NEB-Related Nemaline Myopathy - Gene: NEB. **Variants (1):** R2478_D2512del. **Detection rate:** Mixed or Other Caucasian <10%.
Niemann-Pick Disease Type C - Gene: NPC1. **Variants (1):** I1061T. **Detection rate:** Mixed or Other Caucasian 17%.
Niemann-Pick Disease, SMPD1-Associated - Gene: SMPD1. **Variants (4):** fsP330, L302P, R496L, p.R608del. **Detection rate:** Mixed or Other Caucasian 38%.
Nijmegen Breakage Syndrome - Gene: NBN. **Variants (1):** 657del5. **Detection rate:** Mixed or Other Caucasian 78%.
Northern Epilepsy - Gene: CLN8. **Variants (1):** R24G. **Detection rate:** Mixed or Other Caucasian <10%.
Pendred Syndrome - Gene: SLC26A4. **Variants (5):** IVS8+1G>A, L236P, E384G, T416P, H723R. **Detection rate:** Mixed or Other Caucasian 69%.
PEX1-Related Zellweger Syndrome Spectrum - Gene: PEX1. **Variants (2):** 2097_2098insT, G843D. **Detection rate:** Mixed or Other Caucasian 68%.
Phenylalanine Hydroxylase Deficiency - Gene: PAH. **Variants (13):** IVS-10int-546, I65T, R261Q, R408W, IVS12+1G>A, R408Q, Y414C, L48S, R158Q, G272X, P281L, E280K, S349P. **Detection rate:** Mixed or Other Caucasian 43%.
Polyglandular Autoimmune Syndrome Type 1 - Gene: AIRE. **Variants (2):** Y85C, R257X. **Detection rate:** Mixed or Other Caucasian 65%.
Pompe Disease - Gene: GAA. **Variants (4):** D645E, R854X, IVS1-13T>G, 525delT. **Detection rate:** Mixed or Other Caucasian 67%.
PPT1-Related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. **Variants (3):** T75P, R122W, R151X. **Detection rate:** Mixed or Other Caucasian 53%.
Primary Carnitine Deficiency - Gene: SLC22A5. **Variants (1):** 760C>T. **Detection rate:** Mixed or Other Caucasian <10%.
Primary Hyperoxaluria Type 1 - Gene: AGXT. **Variants (2):** G170R, I244T. **Detection rate:** Mixed or Other Caucasian 42%.
Primary Hyperoxaluria Type 2 - Gene: GRHPR. **Variants (2):** 103delG, c.403_405+2delAAGT. **Detection rate:** Mixed or Other Caucasian 37%.
PROP1-Related Combined Pituitary Hormone Deficiency - Gene: PROP1. **Variants (1):** Ser101fs. **Detection rate:** Mixed or Other Caucasian 55%.
Pseudocholinesterase Deficiency - Gene: BCHE. **Variants (1):** D70G. **Detection rate:** Mixed or Other Caucasian 83%.
Pycnodysostosis - Gene: CTSK. **Variants (1):** X330W. **Detection rate:** Mixed or Other Caucasian <10%.
Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. **Variants (4):** G217R, A218V, L292X, IVS9+1G>C. **Detection rate:** Mixed or Other Caucasian 70%.
Salla Disease - Gene: SLC17A5. **Variants (1):** R39C. **Detection rate:** Mixed or Other Caucasian <10%.
Segawa Syndrome - Gene: TH. **Variants (1):** R233H. **Detection rate:** Mixed or Other Caucasian <10%.
Short Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. **Variants (1):** R107C. **Detection rate:** Mixed or Other Caucasian <10%.
Sjogren-Larsson Syndrome - Gene: ALDH3A2. **Variants (1):** P315S. **Detection rate:** Mixed or Other Caucasian 24%.
Smith-Lemli-Opitz Syndrome - Gene: DHCR7. **Variants (13):** IVS8-1G>C, T93M, W151X(c.452G>A), V326L, R352Q, R352W, R404C, S169L, R242C, R242H, F302L, G410S, E448L. **Detection rate:** Mixed or Other Caucasian 69%.
Spinal Muscular Atrophy - Gene: SMN1. **Variants (1):** SMN1 copy number. **Detection rate:** Mixed or Other Caucasian 95%.
Steroid-Resistant Nephrotic Syndrome - Gene: NPHS2. **Variants (2):** R138Q, R138X. **Detection rate:** Mixed or Other Caucasian 33%.
Sulfate Transporter-Related Osteochondrodysplasia - Gene: SLC26A2. **Variants (4):** C653S, R178X, R279W, IVS1+2T>C. **Detection rate:** Mixed or Other Caucasian 75%.
TPP1-Related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. **Variants (3):** G284V, R208X, IVS5-1G>C. **Detection rate:** Mixed or Other Caucasian 60%.
Tyrosinemia Type I - Gene: FAH. **Variants (6):** IVS12+5G>A, Q64H, P261L, W262X, E357X, IVS6-1G>T. **Detection rate:** Mixed or Other Caucasian 50%.
Usher Syndrome Type 1F - Gene: PCDH15. **Variants (1):** R245X. **Detection rate:** Mixed or Other Caucasian <10%.
Usher Syndrome Type 3 - Gene: CLRN1. **Variants (1):** N48K. **Detection rate:** Mixed or Other Caucasian <10%.
Very Long Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. **Variants (1):** V283A. **Detection rate:** Mixed or Other Caucasian 20%.
Wilson Disease - Gene: ATP7B. **Variants (2):** H1069Q, R778L. **Detection rate:** Mixed or Other Caucasian 40%.
X-Linked Juvenile Retinoschisis - Gene: RS1. **Variants (3):** E72K, G74V, G109R. **Detection rate:** Mixed or Other Caucasian 20%.

Risk Calculations

Below are the full test results for all diseases on the panel. Listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that his future children could inherit each disease. A negative result does not rule out the possibility of being a carrier of untested mutations. Estimates of post-test carrier risk assume a negative family history.

Disease	DONOR 4282 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
ABCC8-Related Hyperinsulinism	1 in 110	1 in 50,000	1 in 50,000
Achromatopsia	1 in 230	1 in 79,000	1 in 30,000
Alkaptonuria	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Alpha-1 Antitrypsin Deficiency	1 in 680	1 in 93,000	1 in 4,700
Alpha-Mannosidosis	1 in 520	1 in 730,000	1 in 500,000
Andermann Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
ARSACS	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Aspartylglycosaminuria	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Ataxia With Vitamin E Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Ataxia-Telangiectasia	1 in 450	1 in 290,000	1 in 100,000
Autosomal Recessive Polycystic Kidney Disease	1 in 75	1 in 18,000	1 in 15,000
Bardet-Biedl Syndrome, BBS1-Related	1 in 750	1 in 480,000	1 in 100,000
Bardet-Biedl Syndrome, BBS10-Related	1 in 290	1 in 180,000	1 in 100,000
Biotinidase Deficiency	1 in 220	1 in 110,000	1 in 61,000
Bloom Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Canavan Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Cartilage-Hair Hypoplasia	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Choroideremia	< 1 in 500	1 in 100,000	1 in 100,000
Citrullinemia Type 1	1 in 150	1 in 70,000	1 in 56,000
CLN3-Related Neuronal Ceroid Lipofuscinosis	1 in 5,600	< 1 in 1,000,000	1 in 200,000
CLN5-Related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	1 in 560	1 in 360,000	1 in 100,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 300	1 in 33,000	1 in 3,000
Cystinosis	1 in 670	1 in 600,000	1 in 200,000
D-Bifunctional Protein Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Factor XI Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 340	1 in 220,000	1 in 100,000
Galactosemia	1 in 430	1 in 150,000	1 in 30,000
Gaucher Disease	1 in 280	1 in 120,000	1 in 50,000
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	1 in 200	1 in 34,000	1 in 7,000
Glutaric Acidemia Type 1	1 in 170	1 in 67,000	1 in 40,000
Glycogen Storage Disease Type Ia	1 in 450	1 in 320,000	1 in 130,000
Glycogen Storage Disease Type Ib	1 in 660	1 in 930,000	1 in 500,000
Glycogen Storage Disease Type III	1 in 290	1 in 180,000	1 in 100,000
Glycogen Storage Disease Type V	1 in 790	1 in 500,000	1 in 100,000
GRACILE Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000	1 in 10,000
Hereditary Fructose Intolerance	1 in 320	1 in 100,000	1 in 26,000
Hereditary Thymine-Uraciluria	1 in 210	1 in 83,000	1 in 40,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 390	1 in 470,000	1 in 360,000
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	1 in 290	1 in 290,000	1 in 250,000

Counsyl

Male

Name: DONOR 4282
DOB [redacted]

Female

Not tested

Disease	DONOR 4282 Residual Risk	Post-test Reproductive Risk	Pre-test Reproductive Risk
Hurler Syndrome	1 in 480	1 in 300,000	1 in 100,000
Hypophosphatasia, Autosomal Recessive	1 in 230	1 in 140,000	1 in 100,000
Inclusion Body Myopathy 2	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 470	1 in 470,000	1 in 250,000
Joubert Syndrome 2	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Krabbe Disease	1 in 360	1 in 210,000	1 in 89,000
Limb-Girdle Muscular Dystrophy Type 2D	1 in 660	< 1 in 1,000,000	1 in 800,000
Limb-Girdle Muscular Dystrophy Type 2E	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	1 in 1,200	1 in 690,000	1 in 90,000
Maple Syrup Urine Disease Type 1B	1 in 250	1 in 250,000	1 in 250,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 270	1 in 63,000	1 in 14,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 430	1 in 340,000	1 in 160,000
Mucolipidosis IV	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Muscle-Eye-Brain Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
NEB-Related Nemaline Myopathy	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Niemann-Pick Disease Type C	1 in 230	1 in 180,000	1 in 150,000
Niemann-Pick Disease, SMPD1-Associated	1 in 400	1 in 400,000	1 in 250,000
Nijmegen Breakage Syndrome	1 in 720	1 in 450,000	1 in 100,000
Northern Epilepsy	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Pendred Syndrome	1 in 220	1 in 63,000	1 in 20,000
PEX1-Related Zellweger Syndrome Spectrum	1 in 350	1 in 160,000	1 in 50,000
Phenylalanine Hydroxylase Deficiency	1 in 88	1 in 17,000	1 in 10,000
Polyglandular Autoimmune Syndrome Type 1	1 in 400	1 in 230,000	1 in 80,000
Pompe Disease	1 in 480	1 in 300,000	1 in 100,000
PPT1-Related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 600	1 in 850,000	1 in 500,000
Primary Hyperoxaluria Type 2	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
PROP1-Related Combined Pituitary Hormone Deficiency	1 in 250	1 in 110,000	1 in 50,000
Pseudocholinesterase Deficiency	1 in 160	1 in 18,000	1 in 3,000
Pycnodysostosis	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 530	1 in 330,000	1 in 100,000
Salla Disease	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Segawa Syndrome	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	1 in 160	1 in 100,000	1 in 100,000
Sjogren-Larsson Syndrome	1 in 330	1 in 330,000	1 in 250,000
Smith-Lemli-Opitz Syndrome	1 in 320	1 in 130,000	1 in 40,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000	1 in 4,800
Steroid-Resistant Nephrotic Syndrome	1 in 600	1 in 950,000	1 in 640,000
Sulfate Transporter-Related Osteochondrodysplasia	1 in 420	1 in 180,000	1 in 45,000
TPP1-Related Neuronal Ceroid Lipofuscinosis	1 in 740	1 in 870,000	1 in 350,000
Tyrosinemia Type I	1 in 350	1 in 240,000	1 in 120,000
Usher Syndrome Type 1F	1 in 190	1 in 150,000	1 in 150,000
Usher Syndrome Type 3	< 1 in 500	< 1 in 1,000,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 110	1 in 39,000	1 in 31,000
Wilson Disease	1 in 140	1 in 50,000	1 in 30,000
X-Linked Juvenile Retinoschisis	< 1 in 500	1 in 50,000	1 in 50,000

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 1172
Fairfax Cryobank -
[redacted]
Report Generated: 2015-09-22
Report Updated: 2015-09-22

Donor 4282

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 30107
Kit Barcode: [redacted]
Method: Genotyping & Sequencing
Specimen: Blood, #31520
Specimen Collection: 2015-09-10
Specimen Received: 2015-09-11
Specimen Analyzed: 2015-09-22

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4282 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: Sequencing: Smith-Lemli-Opitz Syndrome , Diseases Tested: 1, Mutations Tested: 51, 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.

Sequencing: The Illumina TruSight One platform is used to perform sequencing. 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.

Diseases & Mutations Assayed

High Impact Treatment Benefits X-Linked Moderate Impact

H	T	X	M	Disease	#	Mutations
●	○	○	○	Smith-Lemli-Opitz Syndrome	51	♂ Genotyping | c.964-1G>C, c.A356T (p.H119L), c.C1054T (p.R352W), c.C1210T (p.R404C), c.C278T (p.T93M), c.G1055A (p.R352Q), c.G1139A (p.C380Y), c.G1337A (p.R446Q), c.G452A (p.W151X), c.G453A (p.W151X), c.G744T (p.W248C), c.G976T (p.V326L), c.T326C (p.L109P), c.T470C (p.L157P), c.1342G>A (p.E448K), c.1228G>A (p.G410S), c.906C>G (p.F302L), c.725G>A (p.R242H), c.724C>T (p.R242C), c.506C>T (p.S169L), c.A1G (p.M1V) Sequencing | NM_001360:3-9

Recombine

CarrierMap™

Ordering Practice:

Practice Code: 1172
Fairfax Cryobank - [redacted]
[redacted]
Report Generated: 2017-02-21

Donor 4282

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 30107
Kit Barcode: [redacted]
Specimen: Saliva, #83405
Specimen Collection: 2017-02-02
Specimen Received: 2017-02-06
Specimen Analyzed: 2017-02-21

TEST INFORMATION

Test: CarrierMap^{SEQ} (Genotyping & Sequencing)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Genes Sequenced: 1

Partner Not Tested

SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

Donor 4282 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.

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

Tay-Sachs Disease (HEXA): Mutations (78): ♂ 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_912delTTC (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_1212delTG (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), c.1351C>G (p.L451V), c.571-2A>G (IVS5-2A>G) Sequencing | NM_000520:1-14

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
Tay-Sachs Disease	♂ Argentinian: 1/280	82.35%	1/1,587
	♂ Ashkenazi Jewish: 1/29	99.53%	1/6,177
	♂ Cajun: 1/30	>99%	<1/3,000
	♂ European: 1/280	25.35%	1/375
	♂ General: 1/280	32.09%	1/412
	♂ Indian: Unknown	85.71%	Unknown
	♂ Iraqi Jewish: 1/140	56.25%	1/320
	♂ Japanese: 1/127	82.81%	1/739
	♂ Moroccan Jewish: 1/110	22.22%	1/141
	♂ Portuguese: 1/280	92.31%	1/3,640
	♂ Spaniard: 1/280	67.65%	1/865
	♂ United Kingdom: 1/161	71.43%	1/564

Patient Information
Name: Donor 4282
Date of Birth: [redacted]
Sema4 ID: [redacted]
Client ID: [redacted]
Indication: Carrier Screening

Specimen Information
Specimen Type: Purified DNA
Date Collected: 07/15/2021
Date Received: 07/20/2021
Final Report: 08/03/2021

Referring Provider
[redacted]
Fairfax Cryobank, Inc.
[redacted]
[redacted]
[redacted]

Custom Carrier Screen (ECS)

Number of genes tested: 2

SUMMARY OF RESULTS AND RECOMMENDATIONS

Negative

Negative for all genes tested: ***IDUA, and PAH***
To view a full list of genes and diseases tested
please see Table 1 in this report

AR=Autosomal recessive; XL=X-linked

Recommendations

- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder.

Test description

This patient was tested for the genes listed above using one or more of the following methodologies: target capture and short-read sequencing, long-range PCR followed by short-read sequencing, targeted genotyping, and/or copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please view the Table of Residual Risks Based on Ethnicity at the end of this report or at **go.sema4.com/residualrisk** for gene transcripts, sequencing exceptions, specific detection rates, and residual risk estimates after a negative screening result. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. Only known pathogenic or likely pathogenic variants are reported. This carrier screening test does not report likely benign variants and variants of uncertain significance (VUS). If reporting of likely benign variants and VUS are desired in this patient, please contact the laboratory at 800-298-6470, option 2 to request an amended report.

Alice K Tanner

Alice Tanner, Ph.D., M.S., CGC, FACMG, Laboratory Director
Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D

Genes and diseases tested

For specific detection rates and residual risk by ethnicity, please visit **go.sema4.com/residualrisk**

Table 1: List of genes and diseases tested with detailed results

Disease	Gene	Inheritance Pattern	Status	Detailed Summary
⊖ **Negative**				
Mucopolysaccharidosis Type I	*IDUA*	AR	Reduced Risk (see table below)	
Phenylalanine Hydroxylase Deficiency	*PAH*	AR	Reduced Risk (see table below)	

AR=Autosomal recessive; XL=X-linked

Table 2: Residual Risk by ethnicity for negative results

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	Analytical Detection Rate
Mucopolysaccharidosis Type I (AR) NM_000203.4	*IDUA*	African	1 in 376	90%	1 in 3,900	99%
		Ashkenazi Jewish	1 in 1088	99%	1 in 109,000	
		East Asian	1 in 236	63%	1 in 630	
		Finnish	1 in 184	99%	1 in 18,300	
		European (Non-Finnish)	1 in 115	97%	1 in 3,300	
		Native American	1 in 416	92%	1 in 5,000	
		South Asian	1 in 114	97%	1 in 4,100	
		Worldwide	1 in 144	95%	1 in 2,700	
Phenylalanine Hydroxylase Deficiency (AR) NM_000277.1	*PAH*	African	1 in 143	86%	1 in 1,000	99%
		Ashkenazi Jewish	1 in 17	99%	1 in 1,200	
		East Asian	1 in 68	54%	1 in 150	
		Finnish	1 in 158	76%	1 in 650	
		European (Non-Finnish)	1 in 37	89%	1 in 340	
		Native American	1 in 70	87%	1 in 550	
		South Asian	1 in 121	81%	1 in 640	
		Worldwide	1 in 50	88%	1 in 400	
		Turkish	1 in 32	63%	1 in 85	
		Irish	1 in 34	91%	1 in 370	
		Sicilian	1 in 26	48%	1 in 49	
		Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian, Moroccan	1 in 18	88%	1 in 140	

* Carrier detection by HEXA enzyme analysis has a detection rate of approximately 98% (Applies to *HEXA* gene testing only).

ǂ Carrier frequencies include milder and reduced penetrance forms of the disease. Therefore, carrier frequencies may appear higher than reported in the literature (Applies to *BTD, F9, GJB2, GJB1, GLA,* and *MEFV* gene testing only).

‡ Please note that *GJB2* testing includes testing for the two upstream deletions, del(GJB6-D13S1830) and del(GJB6-D13S1854) (PMID:11807148 and 15994881) (Applies to *GJB2* gene testing only).

AR: Autosomal recessive; N/A: Not available; XL: X-linked

Test methods and comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)

NGS was performed on a panel of genes for the purpose of identifying pathogenic or likelypathogenic variants.

Agilent SureSelect^{TM}QXT technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a numberof UTR, intronic or promoter regions that contain previously reported mutations. Samples were pooled and sequenced on the Illumina HiSeq 2500 platform in the Rapid Run mode or theIllumina NovaSeq platform in the Xp workflow, using 100 bp paired-end reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. The exons contained within these regions are noted within Table 1 (as "Exceptions") and will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas, or regions that fall into the Exceptions mentioned above. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition,a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants(Richards et al, 2015). All potentially pathogenic variants may be confirmed by either aspecific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likelybenign variants or variants of uncertain significance identified during this analysis will not be reported.

Copy Number Variant Analysis (Analytical Detection Rate >95%)

Large duplications and deletions were called from the relative read depths on anexon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either acustom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenicsingle-exon CNVs will be confirmed and reported, if detected.

Exon Array (Confirmation method) (Accuracy >99%)

The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targetedexon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each arraymatrix has approximately 180,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGHprobes are enriched to target the exonic regions of the genes in this panel.

Quantitative PCR (Confirmation method) (Accuracy >99%)

The relative quantification PCR is utilized on a Roche Universal Library Probe (UPL)system, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probesets that specific to the target region and a control region with known genomic copynumber. Relative genomic copy numbers are calculated based on the standard ΔΔCt formula.

Long-Range PCR (Analytical Detection Rate >99%)

Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2* , *HBA1* and *HBA2* and *GBA* . The PCR products were then prepared for short-read NGS sequencing and sequenced.Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with The sequencing output to analyze the results. For *CYP21A2* , a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where two copies of a gene are locatedon the same chromosome in tandem, only the second copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However,because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in thetandem allele and this patient is therefore less likely to be a carrier. When anindividual carries both a duplication allele and a pathogenic variant, or multiplepathogenic variants, the current analysis may not be able to determine the phase(cisrans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing isrequired to determine the carrier status.

Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >28,000 variants and genomic frequency data from>138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining

the *a priori* risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate.They are provided only as a guide for assessing approximate risk given a negative result,and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor,medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

Sanger Sequencing (Confirmation method) (Accuracy >99%)

Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with theABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. Falsenegative results may occur if rare variants interfere with amplification or annealing.

SELECTED REFERENCES

Carrier Screening

Grody W et al. ACMG position statement on prenatal/preconception expanded carrier screening. *Genet Med*.2013 15:482-3.

Variant Classification:

Richards S et al. Standards and guidelines for the interpretation of sequence variants: ajoint consensus recommendation of the American College of Medical Genetics and Genomicsand the Association for Molecular Pathology. *Genet Med*.2015 May;17(5):405-24

Additional disease-specific references available upon request.

GENETICS & IVF
Institute

Cytogenetic Report

Client Fairfax Cryobank - [redacted]

Address [redacted]

Reporting Phone # [redacted]

Patient name/Donor Alias Donor # 4282

Donor # [redacted]

Collection Date 09/27/2012

Date Received 09/28/2012

Patient DOB N/A

Specimen type Peripheral Blood

Accession # [redacted]

RESULTS

CYTOGENETIC ANALYSIS

Cells counted	20	Type of banding	GTG
Cells analyzed	5	Band resolution	550
Cells karyotyped	2		
Modal chromosome #	46		

KARYOTYPE 46,XY

FISH

Probe(s) N/A

Nuclei scored N/A

INTERPRETATION

Normal male karyotype
No clonal numerical or structural abnormalities were identified. This normal cytogenetic result does not exclude the possibility of the presence of subtle rearrangements beyond the technical limits of detection with this test.

Comments

Wayne S. Stanley, Ph.D., FACMG
Clinical Cytogeneticist

10/9/12
Date

REVIEWED
10/16/12

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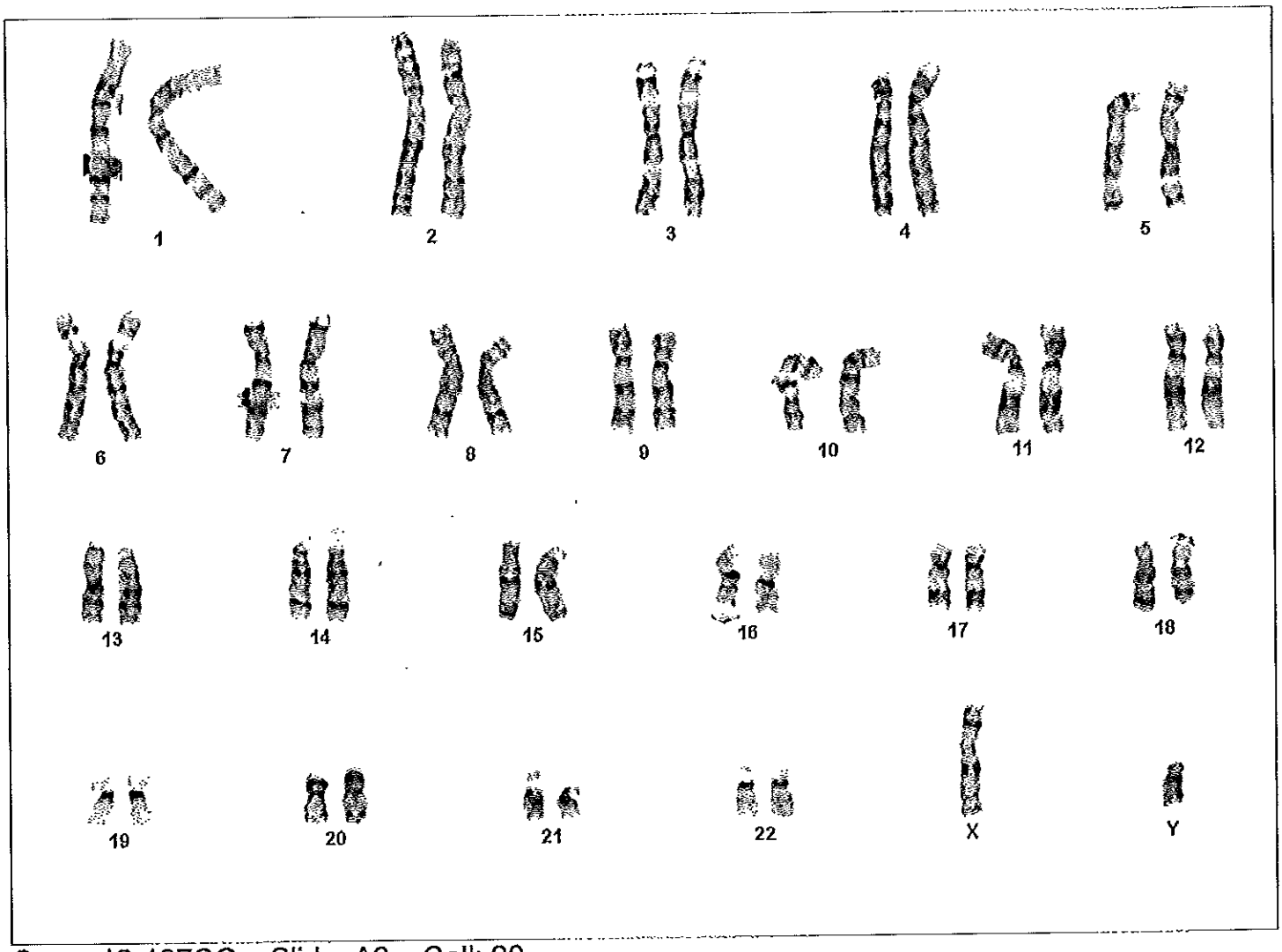
Genetics and IVF Preimplantation Genetics Laboratory

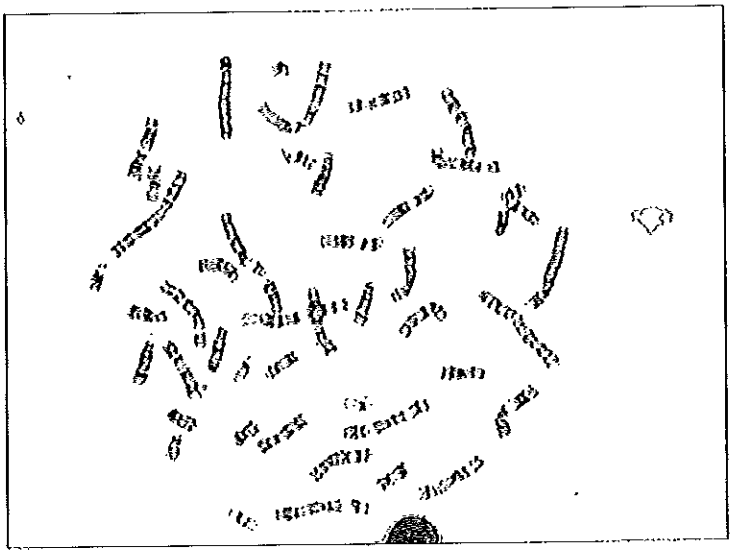
Patient name: DONOR # 4282

Case name: [redacted]

46,XY



Case: 12-137CG Slide: A2 Cell: 20



Quest Diagnostics

Report Status: Final

ID4282, DONOR

Patient Information	Specimen Information	Client Information
ID4282, DONOR **DOB:** [redacted] Gender: M Phone: NG Patient ID: [redacted]	Specimen: [redacted] Requisition: [redacted] Collected: 09/27/2012 Received: 09/28/2012 / 03:48 CDT Reported: 10/03/2012 / 06:17 CDT	Client #: [redacted] AUS0000 [redacted] FAIRFAX CRYOBANK [redacted]

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT		6.21 H	4.20-5.80 Million/uL	IG
HEMOGLOBIN	16.8		13.2-17.1 g/dL	
HEMATOCRIT		51.7 H	38.5-50.0 %	
MCV	83.2		80.0-100.0 fL	
MCH	27.1		27.0-33.0 pg	
RDW	14.2		11.0-15.0 %	
HEMOGLOBIN A	98.1		>96.0 %	IG
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	1.9		1.8-3.5 %	
INTERPRETATION				
Normal phenotype.				
CHOLESTEROL, TOTAL	181		125-200 mg/dL	IG
AST	15		10-40 U/L	IG
ALT	16		9-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	6.7		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT		6.21 H	4.20-5.80 Million/uL	
HEMOGLOBIN	16.8		13.2-17.1 g/dL	
HEMATOCRIT		51.7 H	38.5-50.0 %	
MCV	83.2		80.0-100.0 fL	
MCH	27.1		27.0-33.0 pg	
MCHC	32.5		32.0-36.0 g/dL	
RDW	14.2		11.0-15.0 %	
PLATELET COUNT	207		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	4114		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	2070		850-3900 cells/uL	
ABSOLUTE MONOCYTES	369		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	127		15-500 cells/uL	
ABSOLUTE BASOPHILS	20		0-200 cells/uL	
NEUTROPHILS	61.4		%	
LYMPHOCYTES	30.9		%	
MONOCYTES	5.5		%	
EOSINOPHILS	1.9		%	
BASOPHILS	0.3		%	
ABO GROUP AND RH TYPE				IG
ABO GROUP	O			
RH TYPE	RH(D) POSITIVE			

OK dehydration [redacted] 10/10/12

PERFORMING SITE:

IG QUEST DIAGNOSTICS-IRVING, 4770 REGENT BLVD., IRVING, TX 75063 Laboratory Director: ELISABETH S BROCKIE, DO, CLIA: 45D0697943

ENTERED 10/16/12

CLIENT SERVICES: 866.697.8378

SPECIMEN: [redacted]