Donor 4723

Genetic Testing Summary

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

Last Updated: 08/17/18

Donor Reported Ancestry: Italian, Irish, German 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 99 mutations in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 28 mutations tested by genotyping in the HBB gene	1/290
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
Special Testing		
Glycogen Storage Disease Type II (also known as Pompe Disease)	Negative for 12 mutations in the GAA gene	1/201
Persistent Mullerian Duct Syndrome: Type 1	Negative for 5 mutations in the AMH gene	Reduced risk

Rhizomelic Chondrodysplasia Puntata: Type 1	Negative for 7 mutations in the PEX7 gene	1/582

*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]
[redacted]
Physician: [redacted]
Report Generated: 2015-10-11
Report Updated: 2015-10-13

Donor 4723

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 31204
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #32751
Specimen Collection: 2015-09-24
Specimen Received: 2015-09-25
Specimen Analyzed: 2015-10-11

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 4723 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: Rhizomelic Chondrodysplasia Punctata: Type 1, Glycogen Storage Disease: Type 2 (Pompe Disease), Persistant Mullerian Duct Syndrome: Type1 , Diseases Tested: 3, Mutations Tested: 24, Genes Tested: 3, 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.

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
●	●	○	○	Glycogen Storage Disease: Type II	12	♂ Genotyping | c.C1935A (p.D645E), c.C2560T (p.R854X), c.-32-13T>G, c.525delT (p.E176Rfs), c.C710T (p.A237V), c.T896G (p.L299R), c.T953C (p.M318T), c.G1561A (p.E521K), c.C1634T (p.P545L), c.G1927A (p.G643R), c.C2173T (p.R725W), c.2707_2709delK (p.903delK)
●	●	○	○	Persistent Mullerian Duct Syndrome: Type 1	5	♂ Genotyping | c.G1144T (p.E382X), c.C571T (p.R191X), c. C1518G (p.H506Q), c. G1574A (p.C525Y), c.C283T (p.R95X)
●	○	○	○	Rhizomelic Chondrodysplasia Punctata: Type I	7	♂ Genotyping | c.903+1G>C, c.G649A (p.G217R), c.T875A (p.L292X), c.45_52insGGGACGCC (p.H18RfsX35), c.C120G (p.Y40X), c.T345G (p.Y115X), c.C653T (p.A218V)