Donor 8463-PRS

Genetic Testing Summary

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

Last Updated: 08/27/18

Donor Reported Ancestry: Russian, German, Irish

Jewish Ancestry: Yes

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 97 mutations- in the CFTR gene	1/343
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/5400
Fragile X, PCR DNA Analysis	Normal Male	
Tay Sachs Disease enzyme analysis	Non-carrier by Hexosaminidase A activity	
Tay Sachs Disease	Negative for 5 mutations in the HEXA gene	1/363
Bloom Syndrome	Negative for 1 mutation in the BLM gene	1/3001
Canavan Disease	Negative for 4 mutations in the ASPA gene	1/2801
Fanconi Anemia Type C	Negative for 2 mutations in the FANCC gene	1/8001

Gaucher Disease	Negative for 8 mutations in the GBA gene	1/281
Mucolipidosis Type IV	Negative for 2 mutations in the MCOLN1 gene	1/3026
Niemann-Pick Type A and B	Negative for 4 mutations in the SMPD1 gene	1/1781
Glycogen Storage Disease Type 1A	Negative for 2 mutations in the G6P gene	1/7001
Maple Syrup Urine Disease	Negative for 4 mutations in th4 BCKAD gene	1/8001
Familial Dysautonomia	Negative for 2 mutations in the IKBKAP gene	1/5801

*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.