



Donor 4760

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, Polish, German, Russian

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 99 mutations in the CFTR gene	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/370
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/290
ABCC8-related Hyperinsulinism	Negative for 3 mutations tested in the ABCC8 gene	
Bloom Syndrome	Negative for 1 mutation tested in the BLM gene	
Canavan Disease	Non-carrier for 4 mutations tested in the ASPA gene	

Familial Dysautonomia	Negative for 2 mutations tested in the IKBKAP gene	
Fanconi Anemia Type C	Negative for 3 mutations tested in the FANCC gene	
Gaucher Disease	Negative for 10 mutations tested in the GBA gene	
Glycogen Storage Disease 1 A	Negative for 7 mutations tested in the G6PC gene	
Hexosaminidase A Deficiency (including Tay Sachs Disease)	Negative for 9 mutations tested in the HEXA gene	
Lipoamide Dehydrogenase Deficiency	Negative for 2 mutations tested in the DLD gene	
Maple Syrup Urine Disease Type 1 B	Negative for 3 mutations tested in the BCKDHB gene	
Mucopolipidosis IV	Negative for 2 mutations tested in the MCOLN1 gene	
Neimann-Pick Disease, SMPD1-associated	Negative for 4 mutations tested in the SMPD1 gene	
Usher Syndrome Type 1 F	Negative for 1 mutation tested in the PCDH15 gene	
Usher Syndrome Type 3	Negative for 1 mutation tested in the CLRN1 gene	
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	

*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.