



Donor 4200

Genetic Testing Summary

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

Last Updated: 08/23/18

Donor Reported Ancestry: Polish, Russian, Austrian, English, German

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 for 108 mutations in the CFTR gene	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/420
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 37 mutations tested by genotyping in the HBB gene	<1/500
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
ABCC8-Related Hyperinsulinism	Negative for 3 mutations in the ABCC8 gene	<1/500
Bloom Syndrome	Negative for 2 mutations in the BLM gene	<1/500
Canavan Disease	Negative for 4 mutations in the ASPA gene	<1/500
Familial Dysautonomia	Negative for 3 mutations in the IKBKAP gene	<1/500

Fanconi Anemia Type C	Negative for 4 mutations in the FANCC gene	<1/500
Gaucher Disease	Negative for 9 mutations in the GBA gene	1/330
Glycogen Storage Disease Type 1 A	Negative for 10 mutations in the G6PC gene	<1/500
Maple Syrup Urine Disease Type 3	Negative for 2 mutations in the DLD gene	<1/500
Maple Syrup Urine Disease Type 1B	Negative for 3 mutations in the BCKDHB gene	<1/500
Mucopolipidosis IV	Negative for 2 mutations in the MCOLN1 gene	<1/500
Niemann-Pick Disease Type A	Negative for 3 mutations in the SMPD1 gene	<1/500
Tay Sachs Disease	Negative for 4 mutations in the HEXA gene	1/280
Usher Syndrome Type 1F	Negative for 1 mutations in the PCDH15 gene	<1/500
Usher Syndrome Type 3	Negative for 1 mutations in the CLRN1 gene	<1/500

*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.