



## Donor 6882-PRS

### Genetic Testing Summary

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

Last Updated: 08/30/18

Donor Reported Ancestry: 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 97 mutations- in the CFTR gene	1/834
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/350
Fragile X, PCR DNA Analysis	Normal Male	
Familial Dysautonomia	Negative for 2 mutations in the IKBKAP gene	1/6001
Canavan Disease	Negative for 4 mutations in the ASPA gene	1/3637
Gaucher Disease	Negative for 8 mutations in the GBA gene	1/316
Mucopolidosis Type IV	Negative for 2 mutations in the MCOLN1 gene	1/2521

Fanconi Anemia Type C	Negative for 2 mutations in the FAC gene	1/8801
Niemann-Pick Type A and B	Negative for 4 mutations in the ASM gene	1/2968
Tay Sachs Disease	Negative for 7 mutations in the HEXA gene	1/501
Maple Syrup Urine Disease	Negative for 4 mutations in the BCKDHA and BCKDHB genes	1/8001
Glycogen Storage Disease Type 1 A	Negative for 2 mutations in the G6P gene	1/7001
Bloom Syndrome	Negative for 1 mutation in the BLM gene	1/10601

\*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.