# Donor 9420-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: New Zealand, English, Scottish, Estonian 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 | 1/611 |
| Fragile X, PCR DNA Analysis | Normal Male | |
| Tay Sachs Disease enzyme analysis | Non-carrier by Hexosaminidase A activity | |
| Bloom Syndrome | Negative for 1 mutation in the BLM gene | 1/3300 |
| Canavan Disease | Negative for 4 mutations in the ASPA gene | 1/2801 |
| Fanconi Anemia Type C | Negative for 2 mutations in the FANCC gene | 1/8801 |

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|---|---|---|
| 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 the 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.