



Donor 4549

Genetic Testing Summary

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

Last Updated: 08/20/18

Donor Reported Ancestry: Norwegian, Swedish, German, English

Jewish Ancestry: No

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	1/310
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/700
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/500 for Beta-Thalassemia <1/500 for Sickle Cell
Special Testing		
Hereditary Thymine Uraciluria	Negative for 1 mutation in the DPYD gene	1/210
Autosomal Recessive Polycystic Kidney Disease	Negative for 40 mutation in the PKHD1 gene	1/113
Smith-Lemli-Opitz Syndrome	Negative for 50 mutation in the DHCR7 gene	1/465

Bardet Biedl Syndrome BBS1 Related	Negative for 1 mutation in the BBS1 gene	1/750
Medium Chain Acyl-CoA Dehydrogenase Deficiency	Negative for 8 mutation in the ACADM gene	1/550

*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.