



Donor 4590

Genetic Testing Summary

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

Last Updated: 10/09/18

Donor Reported Ancestry: Norwegian, Swedish, Finnish, Danish, Irish

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	1/300
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/610
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 28 mutations tested by genotyping in the HBB gene	1/290
Tay Sachs enzyme analysis	Non-carrier by Hexosaminidase A activity	
Special Testing		
Herlitz Junctional Epidermolysis Bullosa LAMB3 Panel	Negative for 3 mutations in the LAMB3 gene	<1/500
Smith-Lemli-Opitz Syndrome	Negative by gene sequencing in the DHCR7 gene	1/1754
Wilson Disease	Negative by gene sequencing in the ATP7B gene	1/3125

*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.