



Donor 4345

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: African American

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 for 99 mutations in the CFTR gene	1/290
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/4,300
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)	Negative for 28 mutations tested by genotyping in the HBB gene	1/43
Special Testing		
Alpha-1-Antitrypsin Deficiency	Negative for 4 mutation in the SERPINA1 gene	1/700
Amegakaryocytic Thrombocytopenia	Negative for 23 mutation in the MPL gene	Unknown residual risk
Werner Syndrome	Negative for 8 mutation in the WRN gene	1/326

*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.