



Donor 4330

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: Danish, Croatian, French, 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 for 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 | | |
| Nephrotic Syndrome Type 2 | Negative for 27 mutations in the NPHS2 gene | Unknown residual risk |

*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.