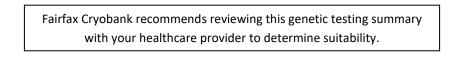


# Donor 4723

# **Genetic Testing Summary**



Last Updated: 08/17/18

Donor Reported Ancestry: Italian, Irish, German

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<br>cell anemia, beta thalassemia, alpha<br>thalassemia trait (aa/ and a-/a-) and<br>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<br>Hemoglobinopathy (including Beta<br>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   |  |  |  |
| Glycogen Storage Disease Type II (also<br>known as Pompe Disease)                                 | Negative for 12 mutations in the GAA gene                      | 1/201  |  |
| Persistent Mullerian Duct Syndrome:<br>Type 1   | Negative for 5 mutations in the AMH gene                       | Reduced risk   |  |

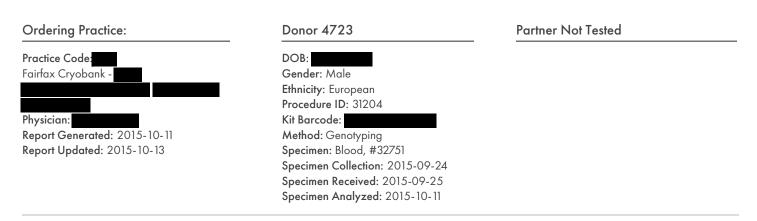
| Rhizomelic Chondrodysplasia Puntata:<br>Type 1 | Negative for 7 mutations in the PEX7 gene | 1/582 |
|--|---|-------|
|--|---|-------|

\*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.



# CarrierMap™



### SUMMARY OF RESULTS

#### NO MUTATIONS IDENTIFIED

## Donor 4723 was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

#### o'' Male

Panel: Rhizomelic Chondrodysplasia Punctata: Type 1, Glycogen Storage Disease: Type 2 (Pompe Disease), Persistant Mullerian Duct Syndrome: Type 1, Diseases Tested: 3, Mutations Tested: 24, Genes Tested: 3, Null Calls: 0

Assay performed by Reprogenetics CLIA ID: 31D1054821 Lab Technician Bo Chu

Reviewed by Pere Colls, PhD, HCLD, Lab Director



#### Methods and Limitations

**Genotyping:** Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

**Limitations:** In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mixup, bone marrow transplantation, blood transfusions, and technical errors.

The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.



# Carrier Map™

### Diseases & Mutations Assayed

🛑 High Impact 🌒 Treatment Benefits 🔵 X-Linked 💛 Moderate Impact

| нтхм |   |    | Mutations  |
|------|---|----|--|
|      | Glycogen Storage Disease: Type II               | 12 | ♂ Genotyping   c.C1935A (p.D645E), c.C2560T (p.R854X), c32-13T>G, c.525delT<br>(p.E176Rfs), c.C710T (p.A237V), c.T896G (p.L299R), c.T953C (p.M318T), c.G1561A<br>(p.E521K), c.C1634T (p.P545L), c.G1927A (p.G643R), c.C2173T (p.R725W),<br>c.2707_2709delK (p.903delK) |
|      | Persistent Mullerian Duct Syndrome: Type<br>1   | 5  | σ <sup>a</sup> Genotyping   c.G1144T (p.E382X), c.C571T (p.R191X), c. C1518G (p.H506Q), c.<br>G1574A (p.C525Y), c.C283T (p.R95X)   |
|      | Rhizomelic Chondrodysplasia Punctata:<br>Type I | 7  | σ³ Genotyping   c.903+1G>C, c.G649A (p.G217R), c.T875A (p.L292X),<br>c.45_52insGGGACGCC (p.H18RfsX35), c.C120G (p.Y40X), c.T345G (p.Y115X),<br>c.C653T (p.A218V)   |