



## Donor 5612

### Genetic Testing Summary

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

Last Updated: 08/27/18

Donor Reported Ancestry: Mexican, Dutch, Norwegian, 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 gene sequencing in the CFTR gene        | 1/1250  |
| Spinal Muscular Atrophy (SMA) carrier screening     | Negative for deletions of exon 7 in the SMN1 gene   | 1/632   |
| Standard testing attached 22 diseases by sequencing | Negative for genes sequenced.                       |   |

\*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.

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**Ordering Practice:**

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Practice Code: [REDACTED]  
Fairfax Cryobank [REDACTED]  
[REDACTED]  
Physician: [REDACTED]  
Report Generated: 2017-09-19

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**Donor 5612**

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DOB: [REDACTED]  
Gender: Male  
Ethnicity: Latin American and European  
Procedure ID: 103294  
Kit Barcode: [REDACTED]  
Specimen: Blood, #104673  
Specimen Collection: 2017-09-07  
Specimen Received: 2017-09-08  
Specimen Analyzed: 2017-09-19

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**Partner Not Tested**

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**TEST INFORMATION**

Test: CarrierMap<sup>SEQ</sup> (Genotyping & Sequencing)  
Panel: Fairfax Cryobank Panel V2- Sequencing  
Diseases Tested: 22  
Genes Tested: 22  
Genes Sequenced: 18

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**SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED**

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Donor 5612 was not identified to carry any pathogenic mutations in the gene(s) tested.

No pathogenic mutations were identified in the genes tested, reducing but not eliminating the chance to be a carrier for the associated genetic diseases. CarrierMap assesses carrier status for genetic disease via molecular methods including targeted mutation analysis and/ or next-generation sequencing; other methodologies such as CBC and hemoglobin electrophoresis for hemoglobinopathies and enzyme analysis for Tay-Sachs disease may further refine risks for these conditions. Results should be interpreted in the context of clinical findings, family history, and/or other testing. A list of all the diseases and mutations screened for is included at the end of the report. This test does not screen for every possible genetic disease.

For additional disease information, please visit [recombine.com/diseases](http://recombine.com/diseases). To speak with a Genetic Counselor, call [855.OUR.GENES](tel:855.OUR.GENES).

Assay performed by   
Reprogenetics  
CLIA ID: 31D1054821  
3 Regent Street, Livingston, NJ 07039  
Lab Technician: Bo Chu

Recombine CLIA # 31D2100763  
Reviewed by Pere Colls, PhD, HCLD, Lab Director

## ADDITIONAL RESULTS: NO INCREASED REPRODUCTIVE RISK

The following results are not associated with an increased reproductive risk.

| Disease (Gene)                                | Donor 5612  | Partner Not Tested |
|---|---|--------------------|
| Spinal Muscular Atrophy: SMN1 Linked (SMN1) * | SMN1 Copy Number: 2 or more copies<br>Method: dPCR & Genotyping |                    |

### \* SMA Risk Information for Individuals with No Family History of SMA

|                  | Detection Rate | Pre-Test Carrier Risk | Post-Test Carrier Risk (2 SMN1 copies) | Post-Test Carrier Risk (3 SMN1 copies) |
|------------------|----------------|-----------------------|--|--|
| European         | 95%            | 1/35                  | 1/632                                  | 1/3,500                                |
| Ashkenazi Jewish | 90%            | 1/41                  | 1/350                                  | 1/4,000                                |
| Asian            | 93%            | 1/53                  | 1/628                                  | 1/5,000                                |
| African American | 71%            | 1/66                  | 1/121                                  | 1/3,000                                |
| Hispanic         | 91%            | 1/117                 | 1/1,061                                | 1/11,000                               |

For other unspecified ethnicities, post-test carrier risk is assumed to be <1%. For individuals with multiple ethnicities, it is recommended to use the most conservative risk estimate.

## Methods and Limitations

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

**Sequencing:** Sequencing is performed using a custom next-generation sequencing (NGS) platform. Only the described exons for each gene listed are sequenced. Variants outside of these regions may not be identified. Some splicing mutations may not be identified. Triplet repeat expansions, intronic mutations, and large insertions and deletions may not be detected. All identified variants are curated, and determination of the likelihood of their pathogenicity is made based on examining allele frequency, segregation studies, predicted effect, functional studies, case/control studies, and other analyses. All variants identified via sequencing that are reported to cause disease in the primary scientific literature will be reported. Variants considered to be benign and variants of unknown significance (VUS) are NOT reported. In the sequencing process, interval drop-out may occur, leading to intervals of insufficient coverage. Intervals of insufficient coverage will be reported if they occur.

**Spinal Muscular Atrophy:** Carrier status for SMA is assessed via copy number analysis by dPCR and via genotyping. Some individuals with a normal number of SMN1 copies (2 copies) may carry both copies of the gene on the same allele/chromosome; this analysis is not able to detect these individuals. Thus, a normal SMN1 result significantly reduces but does not eliminate the risk of being a carrier. Additionally, SMA may be caused by non-deletion mutations in the SMN1 gene; CarrierMap tests for some, but not all, of these mutations. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

**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 mix-up, 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.

This test was developed and its performance determined by Recombine, Inc., and it has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.





## Residual Risk Information

Detection rates are calculated from the primary literature and may not be available for all ethnic populations. The values listed below are for genotyping. Sequencing provides higher detection rates and lower residual risks for each disease. More precise values for sequencing may become available in the future.

| Disease   | Carrier Rate               | Detection Rate | Residual Risk |
|---|----------------------------|----------------|---------------|
| Alpha Thalassemia                               | ♂ General: 1/48            | 50.67%         | 1/97          |
| Beta Thalassemia                                | ♂ African American: 1/75   | 84.21%         | 1/475         |
|   | ♂ Indian: 1/24             | 74.12%         | 1/93          |
|   | ♂ Sardinians: 1/23         | 97.14%         | 1/804         |
|   | ♂ Spaniard: 1/51           | 93.10%         | 1/739         |
| Bloom Syndrome                                  | ♂ Ashkenazi Jewish: 1/134  | 96.67%         | 1/4,020       |
|   | ♂ European: Unknown        | 66.22%         | Unknown       |
|   | ♂ Japanese: Unknown        | 50.00%         | Unknown       |
| Canavan Disease                                 | ♂ Ashkenazi Jewish: 1/55   | 98.86%         | 1/4,840       |
|   | ♂ European: Unknown        | 53.23%         | Unknown       |
| Cystic Fibrosis                                 | ♂ African American: 1/62   | 69.99%         | 1/207         |
|   | ♂ Ashkenazi Jewish: 1/23   | 96.81%         | 1/721         |
|   | ♂ Asian: 1/94              | 65.81%         | 1/275         |
|   | ♂ European: 1/25           | 94.96%         | 1/496         |
|   | ♂ Hispanic American: 1/48  | 77.32%         | 1/212         |
|   | ♂ Native American: 1/53    | 84.34%         | 1/338         |
| Familial Dysautonomia                           | ♂ Ashkenazi Jewish: 1/31   | >99%           | <1/3,100      |
| Familial Hyperinsulinism: Type 1: ABCC8 Related | ♂ Ashkenazi Jewish: 1/52   | 98.75%         | 1/4,160       |
|   | ♂ Finnish: 1/101           | 45.16%         | 1/184         |
| Fanconi Anemia: Type C                          | ♂ Ashkenazi Jewish: 1/101  | >99%           | <1/10,100     |
|   | ♂ General: Unknown         | 30.00%         | Unknown       |
| Gaucher Disease                                 | ♂ Ashkenazi Jewish: 1/15   | 87.16%         | 1/117         |
|   | ♂ General: 1/112           | 31.60%         | 1/164         |
|   | ♂ Spaniard: Unknown        | 44.29%         | Unknown       |
|   | ♂ Turkish: 1/236           | 59.38%         | 1/581         |
| Glycogen Storage Disease: Type IA               | ♂ Ashkenazi Jewish: 1/71   | >99%           | <1/7,100      |
|   | ♂ Chinese: 1/159           | 80.00%         | 1/795         |
|   | ♂ European: 1/177          | 76.88%         | 1/765         |
|   | ♂ Hispanic American: 1/177 | 27.78%         | 1/245         |
|   | ♂ Japanese: 1/177          | 89.22%         | 1/1,641       |
| Joubert Syndrome                                | ♂ Ashkenazi Jewish: 1/92   | >99%           | <1/9,200      |
| Maple Syrup Urine Disease: Type 1B              | ♂ Ashkenazi Jewish: 1/97   | >99%           | <1/9,700      |
| Maple Syrup Urine Disease: Type 3               | ♂ Ashkenazi Jewish: 1/94   | >99%           | <1/9,400      |
|   | ♂ General: Unknown         | 68.75%         | Unknown       |
| Mucopolipidosis: Type IV                        | ♂ Ashkenazi Jewish: 1/97   | 96.15%         | 1/2,522       |
| Nemaline Myopathy: NEB Related                  | ♂ Ashkenazi Jewish: 1/108  | >99%           | <1/10,800     |

| Disease                      | Carrier Rate              | Detection Rate | Residual Risk |
|------------------------------|---------------------------|----------------|---------------|
| Niemann-Pick Disease: Type A | ♂ Ashkenazi Jewish: 1/101 | 95.00%         | 1/2,020       |
| Sickle-Cell Anemia           | ♂ African American: 1/10  | >99%           | <1/1,000      |
|                              | ♂ Hispanic American: 1/95 | >99%           | <1/9,500      |
| Tay-Sachs Disease            | ♂ Argentinian: 1/280      | 82.35%         | 1/1,587       |
|                              | ♂ Ashkenazi Jewish: 1/29  | 99.53%         | 1/6,177       |
|                              | ♂ Cajun: 1/30             | >99%           | <1/3,000      |
|                              | ♂ European: 1/280         | 25.35%         | 1/375         |
|                              | ♂ General: 1/280          | 32.09%         | 1/412         |
|                              | ♂ Indian: Unknown         | 85.71%         | Unknown       |
|                              | ♂ Iraqi Jewish: 1/140     | 56.25%         | 1/320         |
|                              | ♂ Japanese: 1/127         | 82.81%         | 1/739         |
|                              | ♂ Moroccan Jewish: 1/110  | 22.22%         | 1/141         |
|                              | ♂ Portuguese: 1/280       | 92.31%         | 1/3,640       |
|                              | ♂ Spaniard: 1/280         | 67.65%         | 1/865         |
|                              | ♂ United Kingdom: 1/161   | 71.43%         | 1/564         |
| Usher Syndrome: Type 1F      | ♂ Ashkenazi Jewish: 1/126 | 93.75%         | 1/2,016       |
| Usher Syndrome: Type 3       | ♂ Ashkenazi Jewish: 1/120 | >99%           | <1/12,000     |
|                              | ♂ Finnish: 1/134          | >99%           | <1/13,400     |
| Walker-Warburg Syndrome      | ♂ Ashkenazi Jewish: 1/150 | >99%           | <1/15,000     |