



## Donor 5200

### Genetic Testing Summary

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

Last Updated: 08/04/21

Donor Reported Ancestry: English, 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	1/4800
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) by genotyping	Negative for 28 mutations tested in the HBB gene	1/290
Tay Sachs Enzyme Analysis	Non-carrier by Hexosaminidase A analysis	

\*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.

RESULTS RECIPIENT  
FAIRFAX CRYOBANK [REDACTED]

MALE

FEMALE

DONOR 5200

N/A

DOB: [REDACTED]

Ethnicity: Northern European

Sample Type: EDTA Blood

Date of Collection: 02/24/2015

Date Received: 02/26/2015

Date Tested: 03/03/2015

Barcode: [REDACTED]

Indication: Egg or sperm donor

Report Date: 03/03/2015

## Family Prep Screen

**NEGATIVE**

### ABOUT THIS TEST

The Counsyl Family Prep Screen (version 1.0) tests known mutations to help you learn about your chance to have a child with a genetic disease.

### PANEL DETAILS

Fairfax Cryobank Fundamental Panel (3 diseases tested)

### VERSION

DONOR 5200 (Family Prep Screen 1.0)

### RESULTS SUMMARY

**NEGATIVE**

No known or potential disease-causing mutations were detected.

### CLINICAL NOTES

- None

### NEXT STEPS

- If necessary, patients can discuss residual risks with their physician or a genetic counselor.
- To schedule a complimentary appointment to speak with a clinical expert about these results, please visit [counsyl.com/my/consults/](http://counsyl.com/my/consults/).



RESULTS RECIPIENT  
FAIRFAX CRYOBANK [REDACTED]  
[REDACTED]  
Report Date: 03/03/2015

MALE  
DONOR 5200  
DOB: [REDACTED]  
Ethnicity: Northern European  
Barcode: [REDACTED]  
FEMALE  
N/A

## Methods and Limitations

**DONOR 5200 (Family Prep Screen 1.0): targeted genotyping and copy number analysis.**

**Targeted genotyping:** Targeted DNA mutation analysis is used to simultaneously determine the genotype of 127 variants associated with 2 diseases. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

**Copy number analysis:** Targeted copy number analysis is used to determine the copy number of exon 7 of the SMN1 gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of SMN1 are carriers with two SMN1 genes on one chromosome and a SMN1 deletion on the other chromosome. In addition, a small percentage of SMA cases are caused by nondeletion mutations in the SMN1 gene. Thus, a test result of two SMN1 copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more SMN1 gene copies. Some SMA cases arise as the result of de novo mutation events which will not be detected by carrier testing.

**Limitations:** In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. The Counsyl test does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (ACOG Practice Bulletin No. 78, Obstet. Gynecol. 2007;109:229-37).

This test was developed and its performance characteristics determined by Counsyl, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's workup. CLIA Number: #05D1102604.

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### LAB DIRECTORS

*H. Peter Kang*

H. Peter Kang, MD, MS, FACP

*Rebecca Mar-Heyming*

Rebecca Mar-Heyming, PhD, DABMG



Counsyl

RESULTS RECIPIENT

FAIRFAX CRYOBANK

Report Date: 03/03/2015

MALE

DONOR 5200

DOB:

Ethnicity: Northern European

Barcode:

FEMALE

N/A

## Diseases Tested

### Autosomal Recessive Disorders

#### TARGETED GENOTYPING

**Cystic Fibrosis** - Gene: CFTR. Variants (99): G85E, R117H, R334W, R347P, A455E, G542\*, G551D, R553\*, R560T, R1162\*, W1282\*, N1303K, c.1521\_1523delCTT, c.1519\_1521delATC, c.2052delA, c.3528delC, c.489+1G>T, c.579+1G>T, c.1585-1G>A, c.1766+1G>A, 2789+5G>A, c.2988+1G>A, 3849+10kbC>T, E60\*, R75\*, E92\*, Y122\*, G178R, R347H, Q493\*, V520F, S549N, P574H, M1101K, D1152H, c.2012delT, c.262\_263delTT, c.313delA, c.948delT, c.3744delA, c.3773dupT, c.1680-1G>A, 3272-26A>G, c.2051\_2052delAAInsG, S549R(c.1645A>C), R117C, L206W, G330\*, T338I, R352Q, S364P, G480C, C524\*, S549R(c.1647T>G), Q552\*, A559T, G622D, R709\*, K710\*, R764\*, Q890\*, R1066C, W1089\*, Y1092X, R1158\*, S1196\*, W1204\*, Q1238\*, S1251N, S1255\*, c.3067\_3072delG, c.442delA, c.531delT, c.803delA, c.805\_806delAT, c.1545\_1546delTA, M607\_Q643del, c.1911delG,

c.1923\_1931del9InsI, c.1976delA, c.3039delC, c.3536\_3539delCAA, c.3659delC, c.1155\_1156dupTA, c.2052dupA, c.2175dupA, c.2738nsG, 296+12T>C, c.273+1G>A, 405+3A>C, c.274-1G>A, 711+5G>A, c.580-1G>T, c.1766+1G>T, 1898+5G>T, Q996, c.325\_327delTATInsG, 3849+4A>G, c.1075\_1079delSins5. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. Detection rate: Northern European 91%.

**Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)** - Gene: HBB. Variants (28): E7V, K18\*, Q40\*, c.126\_129delCTTT, c.27dupG, IVS-II-654, IVS-II-745, c.315+1G>A, NS-I-6, IVS-I-110, IVS-I-5, c.92+1G>A, -88C>T, -28A>G, -29A>G, c.25\_26delAA, c.217dupA, c.316-2A>C, c.316-2A>G, G25, -87C>G, E7K, W16\*, c.51delC, c.20delA, E27K, E122Q, E122K. Detection rate: Northern European 83%.

#### COPY NUMBER ANALYSIS

**Spinal Muscular Atrophy** - Gene: SMN1. Variant (1): SMN1 copy number. Detection rate: Northern European 95%.


 RESULTS RECIPIENT  
 FAIRFAX CRYOBANK - [REDACTED]

Report Date: 03/03/2015

 MALE  
 DONOR 5200  
 DOB: [REDACTED]  
 Ethnicity: Northern European  
 Barcode: [REDACTED]

 FEMALE  
 N/A

## Risk Calculations

Below are the risk calculations for all diseases tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's future children could inherit each disease. These risks are inherent to all carrier screening tests, may vary by ethnicity, are predicated on a negative family history and are present even after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation.

Disease	DONOR 5200 Residual Risk	Reproductive Risk
Cystic Fibrosis	1 in 300	1 in 33,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 290	1 in 58,000
Spinal Muscular Atrophy	SMN1: 3+ copies 1 in 4,800	1 in 670,000