# Donor 2712

# Genetic Testing Summary

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

Last Updated: 04/23/24

Donor Reported Ancestry: German, Russian  Jewish Ancestry: Yes

| 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 97 mutations in the CFTR gene | 1/834 |
| Tay Sachs Enzyme | Non-carrier by Hexisaminidase A analysis | |
| Limited genetic testing of 9 genes | Negative by genotyping (2006) | |
| **Special Testing** | | |
| Gene: PAH | Non-carrier for 22 mutations by genotyping (2007) | |

*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.

genzyme

# Carrier Testing

**Patient Name:** Donor, 2712
**Referring Physician:** [redacted]
**Specimen #:** [redacted] **Client #:** [redacted]
**Patient ID:** [redacted] **Case #:** [redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 11/14/2006
Date Received: 11/15/2006
Lab ID: [redacted]
Hospital ID:
Specimen Type: **BLDPER**

Fairfax Cryobank
Genetics and IVF Institute
3015 Williams Drive
Suite 110
Fairfax VA 22031

**Ethnicity:** Ashkenazi Jewish

**Indication:** Gamete donor

| Disease | Result | Interpretation |
|---|---|---|
| Bloom Syndrome | Negative | Negative for mutation analyzed. This result reduces but does not eliminate the risk to be a carrier. |
| Canavan Disease | Negative | Carrier risk reduced from 1/57 (1.75%) to 1/2801 (0.04%). |
| Cystic Fibrosis | Negative | Carrier risk reduced from 1/26 (3.9%) to 1/834 (0.1%). |
| Familial Dysautonomia | Negative | Negative for mutations analyzed. This result reduces but does not eliminate the risk to be a carrier. |
| Fanconi Anemia - C | Negative | Negative for mutation analyzed. This result reduces but does not eliminate the risk to be a carrier. |
| Gaucher Disease | Negative | Carrier risk reduced from 1/15 (6.6%) to 1/281 (0.4%). |
| Mucolipidosis Type IV | Negative | Carrier risk reduced from 1/122 (0.8%) to 1/3026 (0.03%). |
| Niemann-Pick Type A | Negative | Carrier risk reduced from 1/90 (1.1%) to 1/1781 (0.06%). |
| Tay-Sachs - DNA | Negative | Carrier risk reduced from 1/30 (3.3%) to 1/363 (0.3%) based on mutation analysis. |

(REPORT CONTINUED ...)

Date: 11/27/2006

genzyme

# Carrier Testing

**Patient Name:** Donor, 2712 **Referring Physician:** [redacted] **Specimen #:** [redacted] **Patient ID:** [redacted]

...Continued From Page 1

| Disease | Result | Interpretation |
|---|---|---|
| Tay-Sachs - Enzyme | Hex. Activity:<br>1597 nmol/mg protein<br>Hex. Percent A: 61.1 | Non carrier<br>Plasma/Serum WBC<br>Non carrier range: Hex A >= 55% >= 55%<br>Carrier range : Hex A 20 - 48% 20 - 49% |

**COMMENTS:**

**DNA:**
The negative results from this analysis cannot eliminate the possibility that this individual carries a mutation not detected by this test. Unless otherwise noted, interpretations are based on a negative family history and the absence of symptoms.

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of these conditions. Although DNA-based testing is highly accurate, rare diagnostic errors may occur. Examples include misinterpretation because of genetic variants, blood transfusion, bone marrow transplantation, or erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

**Enzyme: [ White Blood Cells ]**
This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

**METHOD:**

DNA is isolated from the sample and amplified for disease specific regions using the polymerase chain reaction (PCR). Mutations are identified by hybridization to allele specific oligonucleotides or by solution-phase multiplex allele-specific primer extension with subsequent mutation-specific hybridization and detection.

This test was developed and its performance characteristics determined by Genzyme Genetics. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.

**MUTATIONS ANALYZED / DETECTION RATE**

| Bloom Syndrome |
|---|
| 2281del6ins7<br>Among Ashkenazi Jewish individuals, the mutation analyzed has a 1/100 carrier frequency and a 97% detection rate (Ellis, N. et al. Am. J. Hum. Genet. 63:1685, 1998; Shahrabani-Gargir, L. et al. Genetic Testing 2:293, 1998). In the non-Ashkenazi Jewish population, the mutation is very rare and the detection rate unknown. |

(REPORT CONTINUED ...)

Date: 11/27/2006

D. Alexa Sirko-Osadsa, Ph.D.
Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

genzyme

# Carrier Testing

**Patient Name:** Donor, 2712 **Referring Physician:** [redacted] **Specimen #:** [redacted] **Patient ID:** [redacted]

...Continued From Page 2

## Canavan Disease

433-2A>G Y231X E285A A305E

Among Ashkenazi Jewish individuals with no family history, the prior carrier risk is 1/57 and the mutation detection rate is 98% (Feigenbaum et al. Am. J. Med. Genet. 124A(2):142, 2004; Kaul, R. et al., Am. J. Hum. Genet. 55:34, 1994). Among non-Jewish, European Caucasian individuals, the prior carrier risk is unknown and the mutations analyzed have an approximately 50% detection rate (Sistermans et al. Eur. J. Hum. Genet. 8:557, 2000). The detection rate may be lower in other non-Jewish ethnic groups.

## Cystic Fibrosis

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| ΔF311 | 2043delG | 3120+1G>A | 4016insT | 712-1G>T | G330X | Q359K/T360K | R347P | S549N |
| ΔF508 | 2055del9>A | 3120G>A | 405+1G>A | 935delA | G480C | Q493X | R352Q | S549R T>G |
| ΔI507 | 2105del13ins5 | 3171delC | 405+3A>C | 936delTA | G542X | Q552X | R553X | T338I |
| 1078delT | 2108delA | 3199del6 | 406-1G>A | A455E | G551D | Q890X | R560T | V520F |
| 1288insTA | 2143delT | 3659delC | 444delA | A559T | G85E | R1066C | R709X | W1089X |
| 1677delTA | 2183delAA>G | 3667del4 | 457TAT>G | C524X | K710X | R1158X | R75X | W1204X |
| 1717-1G>A | 2184delA | 3791delC | 574delA | CFTRdele2,3 | L206W | R1162X | R764X | W1282X |
| 1812-1G>A | 2184insA | 3849+10kbC>T | 621+1G>T | D1152H | M1101K | R117C | S1196X | Y1092X C>A |
| 1898+1G>A | 2307insA | 3876delA | 663delT | E60X | N1303K | R117H | S1251N | Y1092X C>G |
| 1898+5G>T | 2789+5G>A | 3905insT | 711+1G>T | E92X | P574H | R334W | S1255X | Y122X |
| 1949del84 | 2869insG | 394delTT | 711+5G>A | G178R | Q1238X | R347H | S364P | |

This 97 mutation assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V.

**Mutation Detection Rates among Ethnic Groups**

Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild presentation (e.g. congenital absence of the vas deferens, pancreatitis) detection rates may vary from those provided here.

| Ethnicity | Carrier risk reduction when no family history | Detection rate | References |
|---|---|---|---|
| African American | 1/65 to 1/338 | 81% | Genet in Med 3:168, 2001 |
| Ashkenazi Jewish | 1/26 to 1/834 | 97% | Am J Hum Genet 51:951, 1994 |
| Asian | | Not Provided | Insufficient data |
| Caucasian | 1/25 to 1/343 | 93% | Genet in Med 3:168, 2001; Genet in Med 4:90, 2002 |
| Hispanic | 1/46 to 1/205 | 78% | Genet in Med 3:168, 2001;www.dhs.ca.gov/pcfh/gdb/html/PDE/CFStudy.htm |
| Jewish, non-Ashkenazi | | Varies by country of origin | Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997 |
| Other or Mixed Ethnicity | | Not Provided | Detection rate not determined and varies with ethnicity |

## Familial Dysautonomia

2507+6T>C R696P

In the Ashkenazi Jewish population, the carrier frequency is approximately 1/30 and the mutations analyzed have a greater than 99.5% detection rate (Slaugenhaupt, SA. Am. J. Hum. Genet. 68:598, 2001; Anderson, SL. Am. J. Hum. Genet. 68:753, 2001). Familial dysautonomia is considered very rare among non-Ashkenazi Jewish individuals.

(REPORT CONTINUED ...)

Date: 11/27/2006

D. Alexa Sirko-Osadsa, Ph.D.
Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

**Patient Name:** Donor, 2712 **Referring Physician:** [redacted] **Specimen #:** [redacted] **Patient ID:** [redacted]

...Continued From Page 3

## Fanconi Anemia - C

IVS4+4 A>T

In the Ashkenazi Jewish population, the IVS4+4A>T mutation is the only FA-C mutation reported and has a carrier frequency of 1/89 (Auerbach, AD. Genetic Testing 1:27, 1997). This mutation has not been found in any affected individual of non-Jewish ancestry.

## Gaucher Disease

| | | | | |
|---|---|---|---|---|
| 1226G | 84GG | 1448C | IVS2+1G>A | 1604A |

*Alternative Nomenclature:*

| | | | | |
|---|---|---|---|---|
| N370S | G insertion | L444P | Intron 2 | R496H |

Among Ashkenazi Jewish individuals, the prior carrier risk is 1/15 and the mutations analyzed have a detection rate of 95% (Beutler, E. et al. Am. J. Hum. Genet. 52:85, 1993). The prior carrier risk is lower and the mutations analyzed have a 70% detection rate among non-Ashkenazi Jewish individuals (Beutler, E. et al. AJDC 147:1175, 1993).

## Mucolipidosis Type IV

IVS3-2A>G EX1-EX7del

The mutations analyzed account for 96% of the MLIV mutations in affected individuals of Ashkenazi Jewish descent (Slaugenhaupt et al. Am. J. Hum. Genet. 65:773, 1999). The frequency of these two mutations is 1/127 among healthy Ashkenazi Jewish individuals (Edelmann et al. Am. J. Hum. Genet. 70:1023, 2002). The calculated carrier frequency is 1/122 in the Ashkenazi Jewish population. Mucolipidosis Type IV is considered extremely rare among non-Ashkenazi Jewish individuals.

## Niemann-Pick Type A

R496L fsP330 L302P

Among Ashkenazi Jewish individuals, the three Niemann-Pick type A disease mutations analyzed have a detection rate of 95% and a carrier frequency of 1/90 (Schuchman, E.H. & Miranda, S.R.P. Genetic Testing 1(1):13, 1997). The detection rate and prior carrier frequency in the non-Ashkenazi Jewish population have not been determined but are expected to be low.

## Tay-Sachs - DNA

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| +TATC1278 | 1421+1 G>C | Δ7.6kb | G269S | IVS7+1 G>A | IVS9+1 G>A | R247W | R249W |

Four mutations and two pseudodeficiency alleles detect approximately 94% of Ashkenazi Jewish and 60% of non-Jewish individuals identified as carriers by enzyme analysis (Kaback, M. et al. JAMA 270:2307,1993). Two additional mutations, Δ7.6kb and IVS7+1G>A, are frequent among French Canadians. The prior carrier risk is 1/30 among Ashkenazi Jews and may vary by ethnic and geographic origin for non-Jewish individuals. The benign pseudodeficiency alleles (R247W, R249W) do not increase the risk for Tay-Sachs disease and account for 36% of non-Jewish and 2% of Ashkenazi Jewish individuals with positive enzyme results. Given the variability in prior carrier risk and detection rate and the high proportion of pseudodeficiency alleles, individual risk reductions are not provided for individuals of mixed or non-Ashkenazi Jewish ethnicity.

Under the direction of:

D. Alexa Sirko-Osadsa, Ph.D.

**Additional approvals by:**

*Enzyme:Stanford Marenberg, Ph.D.*

*Cystic Fibrosis: Lynne Rosenblum-Vos, Ph. D.*



Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

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# Chromosome Analysis

**Patient Name:** 2712, .
**Referring Physician:** [redacted]
**Specimen #:** [redacted] **Client #:** [redacted]
**Patient ID:** [redacted]

DOB: Not Given
SSN:

Date Collected: 12/19/2006
Date Received: 12/20/2006
Lab ID: [redacted]
Hospital ID:
Specimen Type: **Peripheral Blood**

Fairfax Cryobank
Genetics and IVF Institute
3015 Williams Drive
Suite 110
Fairfax VA 22031

**Indication:** Gamete donor

| | | | | | |
|---|---|---|---|---|---|
| **Metaphases Counted:** | 20 | | | **Banding Technique:** | GTW |
| **Metaphases Analyzed:** | 5 | **Number of Cultures:** | 2 | **Banding Resolution:** | 525 |
| **Metaphases Karyotyped:** | 2 | | | **Dept. Section:** | B1 |

**RESULTS: 46,XY**
**Male karyotype**

**INTERPRETATION:**

This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect small rearrangements and low level mosaicism, and cannot detect microdeletions.

Signed: David M. De Bauche

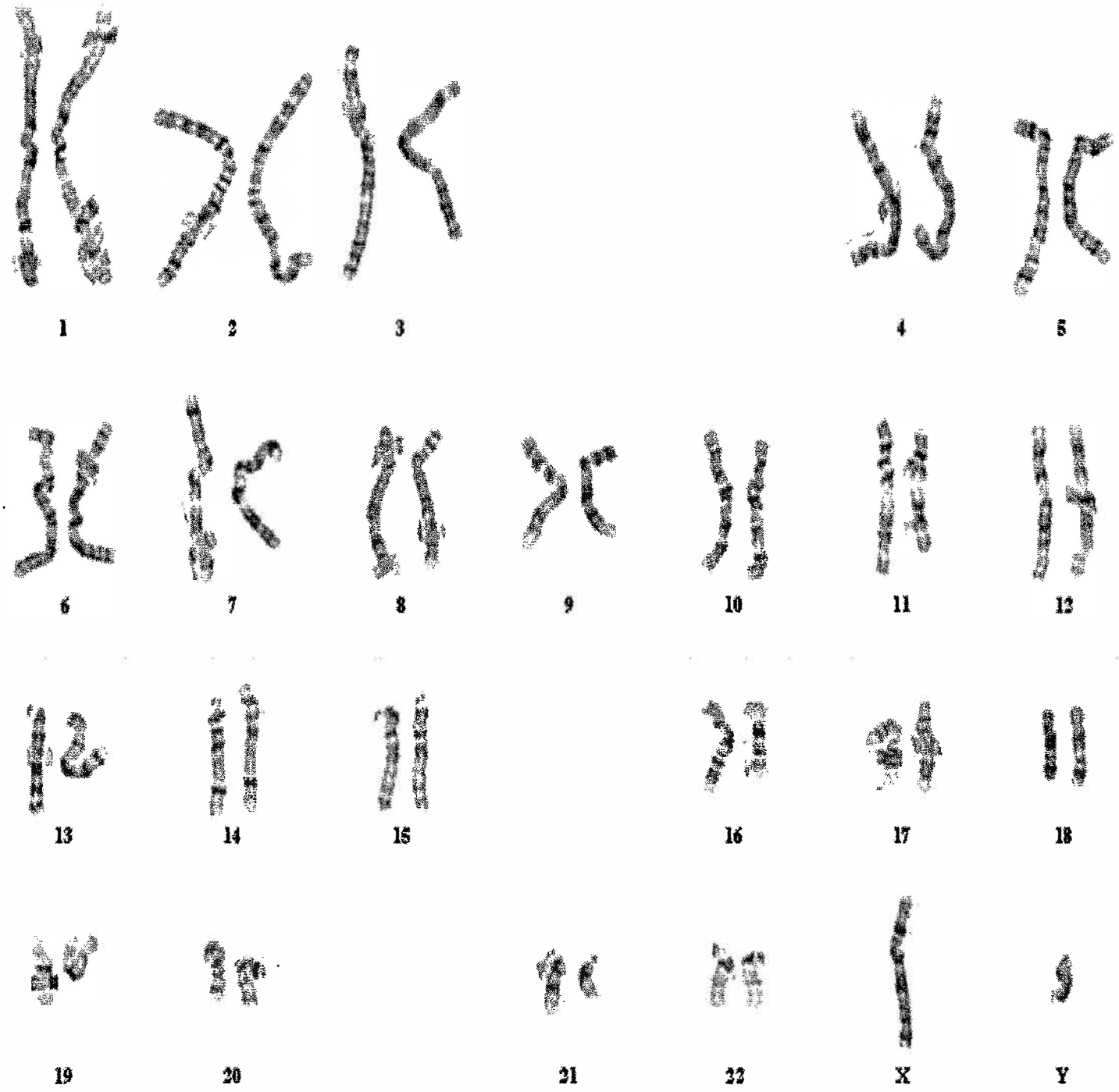
Date: 12/27/2006

David M. De Bauche, Ph.D.



Testing Performed At Genzyme Genetics 10421 University Center Drive Suite 100 Tampa, FL 33612 1-800-966-4440

genzyme




Specimen #: [redacted]
Specimen Type: BLDPER
Patient Name: 2712, .
Image ID: AKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 12/20/2006
Date Reviewed: 12/27/2006
Reviewed By: DMD

genzyme
GENERAL
genetics

QUEST DIAGNOSTICS INCORPORATED
CLIENT SERVICE 410.247.9100

PATIENT INFORMATION
**2712**

DOB: Age: ?
GENDER: M

REPORT STATUS **Final**

ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
507059
FAIRFAX CRYOBANK
3015 WILLIAMS DR STE 110
FAIRFAX, VA 22031

SPECIMEN INFORMATION
SPECIMEN:
REQUISITION: [redacted]
LAB REF NO:

COLLECTED: 11/09/2006 00:00
RECEIVED: 11/10/2006 22:19
REPORTED: 11/14/2006 08:56

COMMENTS: 2712-061109

| Test Name | In Range | Out of Range | Reference Range | Lab |
|---|---|---|---|---|
| CHOLESTEROL,TOTAL* | | | | QBA |
| CHOLESTEROL | 141 | | <200 MG/DL | |
| AST | | | | QBA |
| AST | 26 | | 3-50 U/L | |
| ALT | | | | QBA |
| ALT | 32 | | 3-60 U/L | |
| CBC (INCLUDES DIFF-PLT) | | | | QBA |
| WHITE BLOOD CELL COUNT | 4.1 | | 3.8-10.8 THOUS/MCL | |
| RED BLOOD CELL COUNT | 4.87 | | 4.20-5.80 MILL/MCL | |
| HEMOGLOBIN | 15.0 | | 13.2-17.1 G/DL | |
| HEMATOCRIT | 43.5 | | 38.5-50.0 % | |
| MCV | 89 | | 80-100 FL | |
| MCH | 30.8 | | 27-33 PG | |
| MCHC | 34.4 | | 32-36 G/DL | |
| PLATELET COUNT | 228 | | 140-400 THOUS/MCL | |
| RDW | 11.5 | | 11.0-15.0 % | |
| MPV | 9.9 | | 7.5-11.5 FL | |
| ABSOLUTE NEUTROPHILS | 1943 | | 1500-7800 CELLS/MCL | |
| ABSOLUTE LYMPHOCYTES | 1296 | | 850-3900 CELLS/MCL | |
| ABSOLUTE MONOCYTES | 320 | | 200-950 CELLS/MCL | |
| ABSOLUTE EOSINOPHILS | 537 | | 15-550 CELLS/MCL | |
| ABSOLUTE BASOPHILS | 4 | | 0-200 CELLS/MCL | |
| NEUTROPHILS | 47.4 | | % | |
| LYMPHOCYTES | 31.6 | | % | |
| REACTIVE LYMPHOCYTES | 0.0 | | % | |
| MONOCYTES | 7.8 | | % | |
| EOSINOPHILS | 13.1 | | % | |
| BASOPHILS | 0.1 | | % | |
| COMMENT | Platelets appear adequate. | | | |
| BLOOD GROUP & RH | | | | QBA |
| BLOOD GROUP | A | | | |

Testing performed on a lavender top (EDTA) tube. In the future, please draw all specimens for routine blood bank testing in yellow top (ACD solution B) tubes (order number ASPB300352).
Previous results for the ABO Blood and Rhesus blood group systems tests are not routinely reviewed.

RH TYPE POSITIVE

Previous results for the ABO Blood and Rhesus blood group systems tests are not routinely reviewed.

ENTERED AT 1806

PATIENT INFORMATION
**2712**

REPORT STATUS **Final**

QUEST DIAGNOSTICS INCORPORATED

ORDERING PHYSICIAN [redacted]

DOB: Age: ?
GENDER: M

REPORTED: 11/14/2006 08:56
COLLECTED: 11/09/2006 00:00

| Test Name | In Range | Out of Range | Reference Range | Lab |
|---|---|---|---|---|
| HEMOGLOBINOPATHY EVALUATION | | | | QBA |
| RED BLOOD CELL COUNT | 4.87 | | 4.20-5.80 MILL/MCL | |
| HEMOGLOBIN | 15.0 | | 13.2-17.1 G/DL | |
| HEMATOCRIT | 43.5 | | 38.5-50.0 % | |
| MCV | 89 | | 80-100 FL | |
| MCH | 30.8 | | 27-33 PG | |
| | Platelets appear adequate. | | | |
| RDW | 11.5 | | 11.0-15.0 % | |
| HEMOGLOBIN A1 | 97.6 | | >96.0 % | |
| HEMOGLOBIN F | NONE DETECTED | | 0.0-1.9 | |
| HEMOGLOBIN A2 | 2.4 | | 1.8-3.5 % | |
| HGB SCREEN INTERPRETATION | | | | |
| | THE HEMOGLOBINOPATHY SCREEN IS NORMAL. | | | |
| ABNORMAL HEMOGLOBIN #1 %: | 0.0 | | % | |

**Performing Laboratory Information:**

QBA Quest Diagnostics Incorporated 1901 Sulphur Spring Road Baltimore MD 21227 Laboratory Director: Robert R. L. Smith, M.D.

Recombine

CarrierMap™

**Ordering Practice:**

Practice Code: 926
Fairfax Cryobank
3015 Williams Drive, #110, Fairfax, VA, 22031, US
Physician: Suzanne Seitz
Report Generated: 2017-03-09

**Donor 2712**

DOB: [redacted]
Gender: Male
Ethnicity: Jewish
Procedure ID: 84995
Kit Barcode: [redacted]
Specimen: Sperm, #86018
Specimen Collection: 2007-05-11
Specimen Received: 2017-02-28
Specimen Analyzed: 2017-03-07

**Partner Not Tested**

## TEST INFORMATION

Test: CarrierMap$^{GEN}$ (Genotyping)
Panel: Custom Panel
Diseases Tested: 1
Genes Tested: 1
Mutations Tested: 62

## SUMMARY OF RESULTS: NO MUTATIONS IDENTIFIED

### Donor 2712 was not identified to carry any of the mutation(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. To speak with a Genetic Counselor, call 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

# 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.

**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.

## Diseases & Mutations Assayed

**Phenylalanine Hydroxylase Deficiency (PAH):** Mutations (62): ♂ Genotyping | c.1066-11G>A (IVS10-11G>A), c.1315+1G>A (IVS12+1G>A), c.1241A>G (p.Y414C), c.1222C>T (p.R408W), c.754C>T (p.R252W), c.1223G>A (p.R408Q), c.473G>A (p.R158Q), c.782G>A (p.R261Q), c.814G>T (p.G272X), c.143T>C (p.L48S), c.194T>C (p.I65T), c.896T>G (p.F299C), c.842C>T (p.P281L), c.838G>A (p.E280K), c.117C>G (p.F39L), c.3G>A (p.M1I), c.1A>G (p.M1V), c.611A>G (p.Y204C), c.721C>T (p.R241C), c.727C>T (p.R243X), c.1139C>T (p.T380M), c.926C>T (p.A309V), c.898G>T (p.A300S), c.734T>C (p.V245A), c.818C>T (p.S273F), c.997C>T (p.L333F), c.199T>C (p.S67P), c.1042C>G (p.L348V), c.136G>A (p.G46S), c.728G>A (p.R243Q), c.745C>T (p.L249F), c.581T>C (p.L194P), c.722G>T (p.R241L), c.829T>G (p.Y277D), c.899C>T (p.A300V), c.926C>A (p.A309D), c.1045T>C (p.S349P), c.1157A>G (p.Y386C), c.1169A>G (p.E390G), c.331C>T (p.R111X), c.241_256delACCCATTTGGATAAAC (p.T81fs), c.442-1G>A (IVS4-1G>A), c.463_464insTGTGTACC (p.R155fs), c.569T>G (p.V190G), c.682G>T (p.E228X), c.755G>A (p.R252Q), c.770G>T (p.G257V), c.781C>T (p.R261X), c.800A>G (p.Q267R), c.842+5G>A (IVS7+5G>A), c.856G>A (p.E286K), c.904delT (p.F302fs), c.913-7A>G (IVS8-7A>G), c.935G>T (p.G312V), c.1068C>G (p.Y356X), c.1238G>C (p.R413P), c.1301C>A (p.A434D), c.842+2T>A (IVS7+2T>A), c.764T>C (p.L255S), c.722G>A (p.R241H), c.533A>G (p.E178G), c.456_706+138del11653

## 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 |
|---|---|---|---|
| Phenylalanine Hydroxylase Deficiency | ♂ Arab: Unknown | 46.08% | Unknown |
| | ♂ Ashkenazi Jewish: 1/224 | 44.44% | 1/403 |
| | ♂ Brazilian: 1/71 | 56.41% | 1/163 |
| | ♂ Chinese: 1/51 | 76.57% | 1/218 |
| | ♂ Cuban: 1/71 | 69.64% | 1/234 |
| | ♂ European: 1/51 | 73.00% | 1/189 |
| | ♂ French Canadian: 1/80 | 76.27% | 1/337 |
| | ♂ Iranian: 1/31 | 66.94% | 1/94 |
| | ♂ Korean: 1/51 | 57.58% | 1/120 |
| | ♂ Non-Ashkenazi Jewish: Unknown | 63.64% | Unknown |
| | ♂ Slovakian Gypsy: 1/39 | >99% | <1/3,900 |
| | ♂ Spanish Gypsy: 1/4 | 93.75% | 1/64 |
| | ♂ Taiwanese: Unknown | 83.10% | Unknown |
| | ♂ US Amish: 1/16 | 86.84% | 1/122 |