Pacific Reproductive Services

# Donor 6377-PRS

## Genetic Testing Summary

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

Last Updated: 05/20/21

Donor Reported Ancestry: Scottish, 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 gene by genotyping of 97 mutations in the CFTR gene | 1/343 |
| Spinal Muscular Atrophy (SMA) carrier screening | Negative for deletions of exon 7 in the SMN1 gene | 1/632 |
| Fragile X, PCR DNA Analysis | Normal Male | |
| 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.

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Pacific Reproductive Svcs [redacted]

[redacted]

| | | | |
|---|---|---|---|
| **LCLS Specimen Number:** | **036-229-4790-0** | Account Number: | 04316522 |
| Patient Name: | **6377, DONOR** | Ordering Physician: | **X UNKNOWN** |
| Date of Birth: | [redacted] | Specimen Type: | **BLOOD** |
| Gender: | M | Date Collected: | 02/05/2010 |
| Patient ID: | | Date Received: | 02/07/2010 |
| Lab Number: | (J10-791 L | CoPath Number: | |
| Indications: | FRAGILE X WORKUP | Client Reference: | |

Test: **Chromosome, Blood** Date Reported: **02/15/2010**

| | | | |
|---|---|---|---|
| Cells Counted: | 15 | Cells Karyotyped: | 2 |
| Cells Analyzed: | 5 | Band Resolution: | 650 |

**CYTOGENETIC RESULT: 46,XY**

**INTERPRETATION: NORMAL MALE KARYOTYPE**

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

Molecular results of fragile X testing will be reported separately.

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[redacted]

| | | | |
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| Date of Birth: | [redacted] | Specimen Type: | **BLOOD** |
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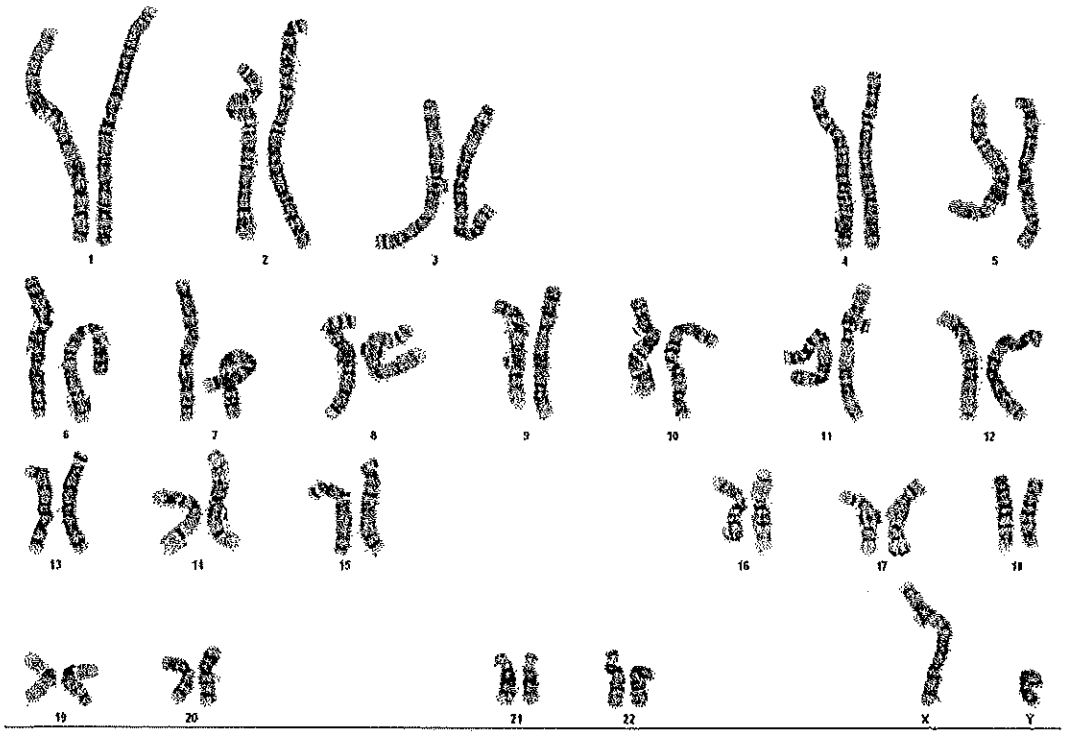

Frederick W. Luthardt PhD, FACMG
Board Certified Cytogeneticist

David Corwin, M.D.
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Test Site: Dynacare Laboratories
550 17th Ave. Suite 200 , SEATTLE , WA , 98122-5789 (206) 861-7050

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LabCorp San Diego
13112 Evening Creek Dr So Ste 200
San Diego, CA 92128
Phone: 858-668-3700

| Specimen Number | Patient ID | Control Number | Account Number | Account Phone Number | Route |
|---|---|---|---|---|---|
| 344-395-7036-0 | | 60111024357 | 04316522 | 415-487-2288 | 00 |

Patient Last Name: DONOR 6377
Patient First Name: X
Patient Middle Name:
Patient SS#:
Patient Phone:
Total Volume:
Age (Y/M/D):
Date of Birth:
Sex: M
Fasting: No
Patient Address:

Account Address: Pacific Reproductive Svcs [redacted]
[redacted]

Additional Information:

| Date and Time Collected | Date Entered | Date and Time Reported | Physician Name | NPI | Physician ID |
|---|---|---|---|---|---|
| 12/10/09 15:00 | 12/11/09 | 12/18/09 18:11ET | UNKNOWN , X | | |

Tests Ordered: CBC With Differential/Platelet; Hgb Frac. w/o Solubility; Tay-Sachs Disease Leukocytes

| TESTS | RESULT | FLAG | UNITS | REFERENCE INTERVAL | LAB |
|---|---|---|---|---|---|
| **CBC With Differential/Platelet** | | | | | |
| WBC | 6.0 | | x10E3/uL | 4.0 - 10.5 | 01 |
| RBC | 5.11 | | x10E6/uL | 4.10 - 5.60 | 01 |
| Hemoglobin | 16.1 | | g/dL | 12.5 - 17.0 | 01 |
| Hematocrit | 45.6 | | % | 36.0 - 50.0 | 01 |
| MCV | 89 | | fL | 80 - 98 | 01 |
| MCH | 31.5 | | pg | 27.0 - 34.0 | 01 |
| MCHC | 35.3 | | g/dL | 32.0 - 36.0 | 01 |
| RDW | 13.3 | | % | 11.7 - 15.0 | 01 |
| Platelets | 224 | | x10E3/uL | 140 - 415 | 01 |
| Neutrophils | 53 | | % | 40 - 74 | 01 |
| Lymphs | 35 | | % | 14 - 46 | 01 |
| Monocytes | 10 | | % | 4 - 13 | 01 |
| Eos | 2 | | % | 0 - 7 | 01 |
| Basos | 0 | | % | 0 - 3 | 01 |
| Neutrophils (Absolute) | 3.2 | | x10E3/uL | 1.8 - 7.8 | 01 |
| Lymphs (Absolute) | 2.1 | | x10E3/uL | 0.7 - 4.5 | 01 |
| Monocytes(Absolute) | 0.6 | | x10E3/uL | 0.1 - 1.0 | 01 |
| Eos (Absolute) | 0.1 | | x10E3/uL | 0.0 - 0.4 | 01 |
| Baso (Absolute) | 0.0 | | x10E3/uL | 0.0 - 0.2 | 01 |
| **Hgb Frac. w/o Solubility** | | | | | |
| **Hgb A** | **98.1** | **High** | % | 94.0 - 98.0 | 02 |
| Hgb S | 0.0 | | % | 0.0 | 02 |
| Hgb C | 0.0 | | % | 0.0 | 02 |
| Hgb A2 | 1.9 | | % | 0.7 - 3.1 | 02 |
| Hgb F | 0.0 | | % | | 02 |
| | | | 1 day | 69.7 - 84.3 | |
| | | | 5 days | 71.0 - 82.6 | |
| | | | 3 weeks | 62.7 - 77.3 | |
| | | | 6- 9 weeks | 41.9 - 63.9 | |
| | | | 3- 4 months | 7.2 - 39.2 | |
| | | | 6 months | 2.5 - 6.9 | |
| | | | 8-12 months | 0.6 - 2.6 | |
| | | | > 1 year | 0.0 - 2.0 | |

CP 12/19/09

marginally CP

Normal adult hemoglobin
See page 2

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Phone: 858-668-3700

| Patient Name | | | | | Specimen Number | | |
|---|---|---|---|---|---|---|---|
| DONOR 6377, X | | | | | 344-395-7036-0 | | |
| Account Number | Patient ID | Control Number | Date and Time Collected | Date Reported | Sex | Age(Y/M/D) | Date of Birth |
| 04316522 | | 60111024357 | 12/10/09 15:00 | 12/18/09 | M | | |

| TESTS | RESULT | FLAG | UNITS | REFERENCE INTERVAL | LAB |
|---|---|---|---|---|---|
| Interpretation | | | | | |
| Normal adult hemoglobin present. ✓ CP | | | | | 02 |
| **Tay-Sachs Disease Leukocytes** | | | | | |
| % Hex A | 60.7 | | % | | 03 |
| Total Activity | 1205.0 | | nmol/hr/mg p | | 03 |
| Tot Act of Norm Ctrl | 997.6 | | nmol/hr/mg p | | 03 |
| Interpretation | | | | | |
| Result: NON-CARRIER ✓ CP 12/14/09 | | | | | 03 |

The above biochemical results are consistent with this individual being a non-carrier for Tay-Sachs disease.

Tay-Sachs disease (TSD) is an autosomal recessive lysosomal storage disorder that causes progressive neurological deterioration. TSD is more common in the Ashkenazi Jewish population where approximately 1 in every 25 individuals is a carrier. Both parents must be carriers of TSD in order to have an affected child. When two people who are both carriers for Tay-Sachs disease have children, the couple has a 25% chance with each pregnancy to have an affected child.

% Hex A Ranges:

| | |
|---|---|
| Non-carrier | >59% |
| Inconclusive | 53 - 59% |
| Carrier | 35 - 52% |

Director Review
Suzette M. Huguenin, Ph.D., FACMG 03
Director, Biochemical and Molecular Genetics

Methodology 03

Total hexosaminidase (A and B) and hexosaminidase B activities were measured by a modification of the heat inactivation method of Kaback using a synthetic fluorogenic substrate. The activity of hexosaminidase A was calculated as the difference between these activities. Kaback MM (1972) thermal fractionation of serum hexosaminidases: applications to heterozygote detection and diagnosis of Tay-Sachs disease. Methods Enzymol 28:862-867. LabCorp Genetics Customer Service, RTP, NC:1-800-345-GENE.

| | | | |
|---|---|---|---|
| 01 | SO | LabCorp San Diego<br>13112 Evening Creek Dr So Ste 200, San Diego, CA 92128 | Dir: Kelli Hanson, MD |
| 02 | BN | LabCorp Burlington<br>1447 York Court, Burlington, NC 27215-3361 | Dir: William F Hancock, MD |
| 03 | TG | LabCorp RTP<br>1912 Alexander Drive, RTP, NC 27709-9998 | Dir: Arundhati Chatterjee, MD |

For inquiries, the physician may contact **Branch: 800-888-1113 Lab: 858-668-3700**

genzyme

# Cystic Fib. .sis Mutation Analysis

**Patient Name:** Donor 6377, .
**Referring Physician:** Madelyn Kahn, MD
**Specimen #:** 61487807 **Client #:** 880107
**Patient ID:** 61236634-17 **Case #:** 61372694

Pacific Reproductive Services
[redacted]

DOB: [redacted]
Sex: M
SSN:

Date Collected: 02/05/2010
Date Received: 02/08/2010
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

**Ethnicity:** Caucasian

**Indication:** Carrier test / Gamete donor

**RESULTS: Negative for the 97 mutations analyzed**

**INTERPRETATION**

This individual's risk to be a carrier is reduced from 1/25 (4%) to 1/343 (0.3%), based on these results and a negative family history.

**COMMENTS:**

**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 |

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

**METHOD / LIMITATIONS:**

DNA is isolated from the sample and tested for the 97 CF mutations listed. Regions of the *CFTR* gene are amplified enzymatically and subjected to a solution-phase multiplex allele-specific primer extension with subsequent hybridization to a bead array and fluorescence detection. Some mutations are specifically identified by bi-directional dideoxysequencing. The assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

Under the direction of:

Narasimhan Nagan, Ph.D., FACMG, DABCC

Date: 02/17/2010



Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357

genzyme
GENETICS

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

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment, when available.

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Phone: 206-861-7000

| Specimen Number | Patient ID | Control Number | Account Number | Account Phone Number | Route |
|---|---|---|---|---|---|
| 036-229-4790-0 | | 60111025438 | 04316522 | 415-487-2288 | 00 |

| Patient Last Name | Patient First Name | Patient Middle Name | Patient SS# | Patient Phone | Total Volume | Age (Y/M/D) | Date of Birth | Sex | Fasting |
|---|---|---|---|---|---|---|---|---|---|
| 6377 | DONOR | | | | | [redacted] | [redacted] | M | No |

Account Address: Pacific Reproductive Svcs [redacted]
[redacted]

Patient Address:

Additional Information: UPIN: XXXXX

| Date and Time Collected | Date Entered | Date and Time Reported | Physician Name | NPI | Physician ID |
|---|---|---|---|---|---|
| 02/05/10 15:27 | 02/06/10 | 02/15/10 20:09ET | UNKNOWN , X | | |

Tests Ordered
S.Muscular Atrophy Carrier; Chromosome, Blood, Routine; Fragile X, PCR Reflex Southern

| TESTS | RESULT | FLAG | UNITS | REFERENCE INTERVAL | LAB |
|---|---|---|---|---|---|

**S.Muscular Atrophy Carrier**

Specimen Type: See below. 01
Peripheral Blood

Clinical Data: See below. 01
Carrier Test/Screening

Results: See below. 01
SMN1 copy number : 2 (Reduced Carrier Risk)

2/18/10

Interpretation See below. 01
This individual has an SMN1 copy number of two. This result reduces but does not eliminate the risk to be a carrier of SMA. Ethnic specific risk reductions based on a negative family history and an SMN1 copy number of two are provided in the Comments section of this report.

Comment: 01
Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutation (most often deletions or gene conversions) in the survival motor neuron (SMN1) gene. Molecular testing assesses the number of copies of the SMN1 gene. Individuals with one copy of the SMN1 gene are predicted to be carriers of SMA. Individuals with two or more copies have a reduced risk to be carriers. (Affected individuals have 0 copies of the SMN1 gene.)

This copy number analysis cannot detect individuals who are carriers of SMA as a result of either 2 (or very rarely 3) copies the SMN1 gene on one chromosome and the absence of the SMN1 gene on the other chromosome or small intragenic mutations within the SMN1 gene. This analysis also will not detect germline mosaicism or mutations in genes other than SMN1. Additionally de novo mutations have been reported in approximately 2% of SMA patients.

---

Carrier Frequency and Risk Reductions for Individuals with No Family History of SMA

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Phone: 206-861-7000

| Patient Name | | | | | Specimen Number | | |
|---|---|---|---|---|---|---|---|
| 6377, DONOR | | | | | 036-229-4790-0 | | |
| Account Number | Patient ID | Control Number | Date and Time Collected | Date Reported | Sex | Age(Y/M/D) | Date of Birth |
| 04316522 | | 60111025438 | 02/05/10 15:27 | 02/15/10 | M | [redacted] | [redacted] |

| TESTS | RESULT | FLAG | UNITS | REFERENCE INTERVAL | LAB |
|---|---|---|---|---|---|

| Ethnicity | Detection Rate(1) | A prior Carrier Risk(1) | Reduced Carrier Risk for 2 copy result | Reduced Carrier Risk for 3 copy result |
|---|---|---|---|---|
| Caucasian | 94.9% | 1:35 | 1:632 | 1:3,600 |
| Ashkenzai Jewish | 90.2% | 1:41 | 1:350 | 1:4,000 |
| Asian | 92.6% | 1:53 | 1:628 | 1:5,000 |
| Hispanic | 90.6% | 1:117 | 1:1061 | 1:11,000 |
| African American | 71.1% | 1:66 | 1:121 | 1:3,000 |
| Mixed Ethnicities | For counseling purposes, consider using the ethnic background with the most conservative risk estimates. | | | |

Method: 01

Specimen DNA is isolated and ampilified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and two reference genes. A mathematical algorithm is used to calculate the number of copies of SMN1. Sequencing of the primer and probe binding sites for the SMN1 real-time PCR reaction is performed on all fetal samples and on samples from individuals with 1 copy of SMN1 on carrier testing, to rule out the presence of sequence variants which could interfere with analysis and interpretaion. False positive or negative results may occur for reasons that include genetic variants, blood transfusions bone marrow transplantation, erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

References: 01

1. Carrier frequency and detection rate are calculated based on analysis of allele frequencies among >1000 individuals from each ethnic group noted. (Genzyme Genetics data submitted for publication). 2. Online review of SMA: http:/www.genereviews.org/profiles/sma

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendment of 1988

February 15, 2010

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Laboratory Corporation of America

Pacific Reproductive Svcs [redacted]

Test Results of: 6377, DONOR
DOB: [redacted] Age: [redacted] Sex: M
Collected on: 02/05/2010
Received on: 02/06/2010
Reported on: 02/15/2010

Branch Number: CAB60
Account Number: 04316522
Specimen Number: 036-229-4790-0
Specimen Type: Blood

Patient ID#:

Physician: UNKNOWN X

**Test: Fragile X, PCR DNA Analysis**

**Result:**

| |
|---|
| **NORMAL, Male**<br>**33 CGG repeats identified** cp 2/19/10 |

**Interpretation:**
DNA studies by PCR analysis identified one allele. These results do not provide evidence of the common CGG repeat expansion observed in patients with Fragile X syndrome. Routine chromosome analysis is recommended in the diagnostic work-up for other causes of mental retardation. Due to the nature of the assay, small variations in reported repeat number may exist within and between laboratories.

Fragile X syndrome is one of the most common causes of inherited mental retardation. Some individuals with Fragile X have characteristic physical features and behaviors. There can be wide variability in phenotypic expression. Fragile X is most often caused by an expansion in the number of the CGG repeats in the Fragile X gene *(FMR1)*. People with fewer than 45 CGG repeats have alleles within the normal range. People with 45-54 repeats are considered normal but have alleles in the grey zone. Some increases and decreases in repeat number can occur in offspring of individuals with grey zone alleles, but the chance is small that grey zone alleles would expand to a full mutation in the next generation. Those with 55-200 repeats have alleles in the premutation range. These individuals are not expected to have Fragile X, but are at increased risk to have children with Fragile X syndrome. Individuals with more than 200 repeats have full mutations and are expected to be clinically affected. Exceptions can occur as there are rare forms of FMRP deficiency not caused by CGG expansion, which may not be detected by this analysis.

**Methodology:**
DNA analysis of the *FMR1* gene was performed by PCR amplification followed by agarose gel, as well as capillary electrophoresis. Southern blot analysis was not indicated due to the presence of one normal allele by PCR. The detection rate of this test is >99% for the common Fragile X expansion *(FRAXA)*. This test does not examine the *FRAXE* expansion. Molecular-based testing is highly accurate, but as in any laboratory test, rare diagnostic errors may occur. All test results must be combined with clinical information for the most accurate interpretation.

This test was developed and its performance characteristics determined by Laboratory Corporation of America Holdings (LabCorp). 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 research.

**References:**


1. Park V, Howard-Peebles P, Sherman S, Taylor A, and Wulfsberg E. (1994). Am J Med Genet 53:380-381.
2. Maddalena A, et al. (2001). Genet Med 3:200-205.
3. Jacquemont, S, Hagerman, RJ, Lechey, MA, Hall, DA, Levine, RA, Brunberg, JA, Zhang, L, Jardini, T, Gane, LW, Harris, SW, Herman, K, Grigsby, J, Greco, CM, Berry-Kravis, E, Tassone, F, and Hagerman, PJ. (2004) J Amer Med Assoc 291:460-469.
4. Hagerman PJ and Hagerman RJ. (2004) Am J Hum Genet 74:805-816.


**Results Released By:** Kenneth J. Friedman, Ph.D., Director
**Report Released By:** Lori A. Carpenter, MS, Genetic Counselor

Arundhati Chatterjee, M.D.
Medical Director


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1912 Alexander Drive, RTP, NC, 27709 (800) 345-GENE