Integrated GENETICS
LabCorp Specialty Testing Group

Sample Report

Fragile X Analysis

Patient Name:
Referring Physician: Ordering Doctor, MD
Specimen #: 1345678 **Client #:**
Patient ID: 12345678 **Case #:**

City Hospital
123 Anywhere Avenue
Anywhere, ST 12345

DOB: 00/00/1982
Sex: F
SSN:

Date Collected: 03/07/2012
Date Received: 03/08/2012
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

Indication: Carrier test / No family history

RESULTS: Southern: 2.8, 3.1, 5.2 and 5.5 kb. PCR: 29 and 110 repeats

INTERPRETATION

Premutation carrier of fragile X syndrome. This individual is at risk for primary ovarian insufficiency, late-onset fragile X-associated tremor/ataxia syndrome (FXTAS), and for having children with fragile X syndrome.

COMMENTS:
Genetic counseling is recommended for discussion of the clinical implications of this result for this individual and for at-risk family members.

Fragile X syndrome is caused by an expansion of CGG repeat sequences in the FMR1 gene in 99% of cases. There are rare FMR1 mutations including missense mutations and gene deletions which cause fragile X syndrome. The interpretation is based on the following ranges of repeat sequences:

Negative:	< 45 repeats
Intermediate:	45-54 repeats
Premutation:	55-200 repeats with normal methylation pattern
Full Mutation:	>200 repeats with abnormal methylation pattern

Reported CGG repeat sizes may vary as follows: +/- one for repeats less than 60, and +/- two to four for repeats in the 60 - 120 range respectively. For repeats greater than 120, the accuracy is +/- 10%.

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition. Genetic counseling is recommended for any individual seeking additional information regarding interpretation of genetic test results.

METHOD / LIMITATIONS:
Isolated DNA is tested by the polymerase chain reaction (PCR) to determine the size of the CGG repeat within the FMR1 gene. PCR products are generated using a fluorescence labeled primer and sized by capillary gel electrophoresis. If indicated, Southern blot analysis is performed by hybridizing the probe StB12.3 to EcoRI- and EagI-digested DNA. The analytical sensitivity of both Southern blot and PCR analyses is 99% for expansion mutations in the FMR1 gene. False positive or negative results may occur for reasons that include somatic or tissue-specific mosaicism, rare genetic variants, blood transfusions, bone marrow transplantation, or erroneous representation of family history.

REFERENCES: Garber K et al. Eur J Hum Genet 2008;16:666-72. Sherman S et al. Genet Med 2005;7:584-87. Wittenberger MD et al. Fertil Steril 2007;87:456-65. Rodriguez-Revenga L et al. Eur J Hum Genet 2009;1-4.

The test was developed and its performance characteristics have been determined by Esoterix Genetic Laboratories, LLC. 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.

Signed:

Date: 03/16/2012

Testing Performed At Esoterix Genetic Laboratories, LLC 3400 Computer Drive Westborough, MA 01581 1-800-255-7357