Integrated GENETICS
LabCorp Specialty Testing Group

Sample Report

Fragile X Analysis Carrier Screen

Patient Name:
Referring Physician: Ordering Doctor, MD
Specimen #: 112345678
Patient ID: 12345678-9

Client #: 654321
Case #: 87654321

DOB: 0/0/1980
Sex: F
SSN:

Date Collected: 02/17/2012
Date Received: 02/18/2012
LAB ID:
Hospital ID:
Specimen Type: **BLDPER**

Any Hospital
Main Street
Anytown, AA 12345
USA

Indication: Carrier Test / No family history

RESULTS: PCR: 25 and 49 repeats

INTERPRETATION:

Intermediate: not a carrier of a fragile X expansion mutation. This result is not associated with fragile X syndrome. See Comments.

COMMENTS:
Minor increases and decreases in the size of intermediate alleles may occur when they are passed to future generations. There is no measurable risk of this individual having a child with fragile X syndrome. Southern blot analysis is not indicated when PCR results are negative or intermediate and there is no family history of unexplained intellectual disability, ovarian dysfunction or ataxia tremor.

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.

on 02/24/2012