LabCorp

Patient Report

Specimen ID: 190-990-5101-0
Control ID:

SAMPLE1, TEST1

Acct #: 32300041 **Phone:** (919) 123-8888 **Rte:** 90
CMBP-QA-PRO-TEST-ACCt-BETTYSCO
tHIS-IS-ACCOUNT-cmbp-BETTYSCOT
RTP-UPDATE-cmbp-miami BLVD
DURHAM NC 27703

Patient Details
DOB:
Age(y/m/d): 000/00/00
Gender: F **SSN:**
Patient ID:

Specimen Details
Date collected: 07/09/2019 0000 Local
Date received: 07/09/2019
Date entered: 07/09/2019
Date reported: 07/09/2019 0000 ET

Physician Details
Ordering:
Referring:
ID:
NPI:

Ordered Items
Fragile X, PCR reflex Southern; Fragile X Southern Blot

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Fragile X, PCR reflex Southern					
Fragile X DNA					01
Fragile X Analysis by Southern Blot is indicated for this sample. Final report will follow under separate cover.					
Comment:					01
This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.					
Fragile X Syndrome, PDF	.				01
Fragile X Southern Blot					
Premutation, Female Abnormal					01

RESULTS: PCR and Southern Blot: 30 and 55 CGG repeats.

INTERPRETATION:
Premutation carrier of fragile X syndrome. Premutation allele has2 AGG interruptions: (CGG)5 AGG (CGG)5 AGG (CGG)45. Risk of expansion to full mutation in offspring is less than 1% (Domniz, PMID:30619448). This individual may be at risk for primary ovarian insufficiency (POI), late-onset fragile X-associated tremor/ataxia syndrome (FXTAS), and for having children with fragile X syndrome. See comments.

COMMENTS:
Approximately 2% of women with primary ovarian insufficiency (POI) and 14% with familial ovarian insufficiency have a premutation (Wittenberger MD, et al. Fertil Steril 2007;87:456-465). After the age of 50, approximately 16% of women with a premutation have late-onset fragile X-associated tremor/ataxia syndrome (FXTAS) (Rodriguez-Revenga, L et al. Eur J Hum Genet 2009;1-4). Genetic counseling is recommended for discussion of the clinical implications of this result for this individual and for at-risk family members. Prenatal diagnosis is available.

Fragile X syndrome is an X-linked disorder of intellectual

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TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

disability with variable severity. Expansions of CGG repeat sequences in the FMR1 gene account for 99% of mutations causing fragile X syndrome. The interpretation is based on the following ranges of repeat sequences:

Negative:	less than 45 repeats
Intermediate:	45-54 repeats
Premutation:	55-200 repeats with normal methylation pattern
Full Mutation:	greater than 200 repeats with abnormal methylation pattern

The risk for a premutation allele of 55-90 repeats to expand to a full mutation in offspring, when transmitted by a carrier female, is reduced with increasing number of AGG interruptions in the CGG repeat sequence. (Yrigollen, PMID:22498846; Nolin, PMID:25210937). Greater than 99% of males and approximately 50% of females with the full mutation are intellectually disabled. Other signs and symptoms may include delayed speech and language skills, autism, hyperactivity, developmental delay, increased susceptibility to seizures,macroorchidism in males, a long, narrow face with prominent ears, and joint laxity. Individuals with a premutation do not have fragile X syndrome, but may have an increased risk for fragile X-related disorders. Females may have fragile X-associated primary ovarian insufficiency(FXPOI), which can cause infertility or early menopause. Most males with a premutation and some females are at risk for fragile X-associated tremor and ataxia syndrome (FXTAS), which can affect balance and is associated with tremor and memory problems in older individuals. Treatment is supportive and focuses on educational and behavioral support and management of symptoms. (Santoro, PMID:22017584).
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.

METHODS/LIMITATIONS:
DNA is amplified by the polymerase chain reaction (PCR) to determine the size of the CGG repeat region 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 Eagl-digested DNA. The analytical sensitivity of both Southern blot and PCR analyses is 99% for expansion mutations in the FMR1 gene. Reported CGG repeat sizes may vary as follows: +/- one for repeats less than 60, and +/- two to

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TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB

four for repeats in the 60-120 range. For repeats greater than 120, the accuracy is +/- 10%. If 55-90 trinucleotide repeats are detected in females (excluding prenatal specimens), a PCR assay targeting AGG sequences within the CGG repeats is perfomed to assess the number and position of AGG interruptions.

REFERENCES:

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

Results Released By: EMILY SULLIVAN , SQE-A
Report Released By: EMILY SULLIVAN

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For inquiries, the physician may contact **Branch: 800-762-4344 Lab: 336-222-7566**

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July 9, 2019

LabCorp
Laboratory Corporation of America

CMBP QA TEST ACCOUNT
4512 S. MIAMI BLVD
SUITE 150
RTP, NC 27215-5820

Test Results of: SAMPLE1, TEST1
DOB: Age: Sex: F
Collected on: 07/09/2019
Received on: 07/09/2019
Reported on: 07/09/2019

Branch Number: POE00
Account Number: 32300041
Specimen Number: 190-990-5101-0
Specimen Type: Blood

Patient ID#:

Physician:

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