

LASTNAME, FIRSTNAME

DOB: mm/dd/yyyy

Account Number: 00000000



Patient ID:

Specimen ID: 000-000-0000-0

Age: 00

Ordering Physician:

Sex: Female

Date Collected: mm/dd/yyyy

Date Received: mm/dd/yyyy

Date Reported: mm/dd/yyyy

Date Entered: mm/dd/yyyy

Specimen Type: Whole Blood

Ethnicity: Not Provided

Indication: Carrier Test / Screening

Fragile X Syndrome, Carrier

Summary: ● NEGATIVE

SAMPLE REPORT

Negative Results

Disorder (Gene)	Result	Interpretation
Fragile X syndrome (<i>FMR1</i>) NM_002024.5	NEGATIVE PCR: 29 and 29	Not a carrier of a fragile X expansion. Risk: NOT at an increased risk for an affected pregnancy.

Recommendations

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of positive results, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Labcorp Genetic Counselors please visit <https://womenshealth.labcorp.com/genetic-counseling> or call (855) GC-CALLS (855-422-2557).

Additional Clinical Information

Fragile X syndrome is an X-linked disorder of intellectual disability with variable severity. Expansions of CGG repeat sequences in the *FMR1* gene account for 99% of variants causing fragile X syndrome. The risk of expansion from a premutation allele of 55-90 repeats 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).

Comments

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder(s) tested. Information about the disorder(s) tested is available at <https://womenshealth.labcorp.com>.

Methods/Limitations

Fragile X Syndrome: PCR analysis is used to detect the number of CGG repeats on each allele of the *FMR1* gene. The reportable range is 5-200 repeats. Alleles with expansions above 200 repeats are reported as >200. In females, excluding prenatal specimens, alleles between 55 and 90 repeats are assessed by a PCR assay to determine the number and position of AGG interruptions within the CGG repeats. If indicated, methylation status is determined by PCR analysis based on methylation-specific immunoprecipitation. Interpretation of repeat expansion results is based on the following ranges: Negative: < 45 repeats; intermediate: 45-54 repeats; premutation: 55-200 repeats; full mutation: >200 repeats. The analytical sensitivity of this assay for the detection of expanded alleles in the *FMR1* gene is estimated to be >99%. Reproducibility of repeat numbers is typically ± 1 for alleles containing up to 60 repeats, ± 3 for alleles with 61-119 repeats, and ± 10 for alleles with >119 repeats. Low levels of mosaicism (<5%) and *FMR1* variants unrelated to trinucleotide expansion are not detected by this assay.

Limitations: Technologies used do not detect germline mosaicism and do not rule out the presence of large chromosomal aberrations including rearrangements and gene fusions, or variants in regions or genes not included in this test, or possible inter/intragenic interactions between variants, or repeat expansions. Variant classification and/or interpretation may change over time if more information becomes available. False positive or false negative results may occur for reasons that include: rare genetic variants, sex chromosome abnormalities, pseudogene interference, blood transfusions, bone marrow transplantation, somatic or tissue-specific mosaicism, mislabeled samples, or erroneous representation of family relationships.

References

Hunter JE, Berry-Kravis H, et al., *FMR1* Disorders. 1998 Jun16 (Updated 2019 Nov 21). In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews®[Internet]. PMID: 20301558

Spector E, Behlmann A, Kronquist K, et al. Laboratory testing for fragile X, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 23, 799 (2021). PMID: 33795824

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Age: 00

Sex: Female

Ordering Physician:

Fragile X Syndrome, Carrier

Performing Labs

Component Type	Performed at	Laboratory Director
Technical component, processing	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG
Technical component, analysis	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG
Professional component	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581-1771	Hui Zhu, PhD, FACMG

For inquiries, the physician may contact the lab at 800-255-7357

This test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. It has not been cleared or approved by the Food and Drug Administration.

Esoterix Genetic Laboratories, LLC is a subsidiary of Laboratory Corporation of America Holdings, using the brand Labcorp.

Patient Details

LASTNAME, FIRSTNAME

Phone:

Date of Birth: mm/dd/yyyy

Age: 00

Sex: Female

Patient ID:

Alternate Patient ID:

Physician Details

CLIENT NAME

CLIENT ADDRESS

Phone: 000000000

Account Number: 00000000

Physician ID:

NPI:

Specimen Details

Specimen ID: 0000000000

Control ID:

Alternate Control Number:

Date Collected: mm/dd/yyyy 0000 Local

Date Received: mm/dd/yyyy 1047 ET

Date Entered: mm/dd/yyyy 1019 ET

Date Reported: mm/dd/yyyy 1431 ET

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