Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT
WX0000003039 M 12/05/1988 33 Y

Referral Testing

Collected: 02/19/2022 06:07 Received: 02/19/2022 06:07

Test Name	Result	Flag	Ref-Ranges	Units	Site
Fragile X (FMR1) with Reflex to Methylation Analysis					
Frag X Specimen	Whole Blood				ARRL
Fragile X Allele 1	15			CGG repeats	ARRL
Fragile X Allele 2	16			CGG repeats	ARRL
Fragile X Methylation Pattern	Normal				ARRL
Fragile X Interpretation	See Note				ARRL

This individual has a FMR1 allele with a CGG repeat size in the normal range; therefore, he is predicted to be neither affected with, nor a carrier of, fragile X syndrome (FXS). This test does not detect rare FMR1 variants causing less than 1% of FXS.

Methylation pattern is normal for gender.

This result has been reviewed and approved by Rong Mao, M.D.
BACKGROUND INFORMATION: Fragile X (FMR1) with Reflex to Methylation Analysis
CHARACTERISTICS OF FRAGILE X SYNDROME (FXS): Affected males have moderate intellectual disability, hyperactivity, perseverative speech, social anxiety, poor eye contact, hand flapping or biting, autism spectrum disorders and connective tissue anomalies in males. Females are usually less severely affected than males. FXS is caused by FMR1 full mutations.
CHARACTERISTICS OF FRAGILE X TREMOR ATAXIA SYNDROME (FXTAS): Onset of progressive ataxia and intention tremor typically after the fourth decade of life. Females also have a 21 percent risk for primary ovarian insufficiency. FXTAS is caused by FMR1 premutations.
Incidence of FXS: 1 in 4,000 Caucasian males and 1 in 8,000 Caucasian females.
INHERITANCE: X-linked.
PENETRANCE OF FXS: Complete in males; 50 percent in females.
PENETRANCE OF FXTAS: 47 percent in males and 17 percent in females >50 years of age.
CAUSE: Expansion of the FMR1 gene CGG triplet repeat.
  Full mutation: typically >200 CGG repeats (methylated).
  Premutation: 55 to approx 200 CGG repeats (unmethylated).
  Intermediate: 45-54 CGG repeats (unmethylated).
  Normal: 5-44 CGG repeats (unmethylated).
CLINICAL SENSITIVITY: 99 percent.
METHODOLOGY: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary

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electrophoresis. Methylation-specific PCR analysis is performed for CGG repeat lengths of >100 to distinguish between premutation and full mutation alleles.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent; estimated precision of sizing for intermediate and premutation alleles is within 2-3 CGG repeats.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Rare FMR1 variants unrelated to trinucleotide expansion will not be detected. A specific CGG repeat size estimate is not provided for full mutation alleles. AGG trinucleotide interruptions within the FMR1 CGG repeat tract are not assessed.

PHENOTYPE	NUMBER OF CGG REPEATS
Unaffected	<45
Intermediate	45-54
Premutation	55-200
Affected	>200

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.
Performed by ARUP Laboratories,
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www.aruplab.com, Tracy I. George, MD - Lab. Director

Performing Site:
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