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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: 2 years Male

Specimen Collected: 05-Sep-23 17:36

Fragile X (FMR1) w/Reflex to Methylation **|Received: 05-Sep-23 17:37** **Report/Verified: 05-Sep-23 17:43**

Procedure	Result	Units	Reference Interval
FRAG X Specimen	Whole Blood		
Fragile X Allele 1	> 200		
Fragile X Allele 2	Not Applicable	CGG repeats	
Fragile X Methylation Pattern	**Full** *		
Fragile X Interpretation	See Note f1 i1		

Result Footnote

f1: Fragile X Interpretation

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

This individual has a loss-of-function FMR1 allele (typically greater than 200 CGG repeats that is fully methylated); thus, he is affected with fragile X syndrome. Genetic consultation is recommended.

The expanded allele is fully methylated and predicted to be nonfunctional.

This result has been reviewed and approved by [redacted]

Test Information

i1: Fragile X Interpretation

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.

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Jonathan R. Genzen, MD, PhD

ARUP Accession: 23-248-900235
Report Request ID: 18462947
Printed: 08-Sep-23 18:11

Test Information

i1: Fragile X Interpretation

METHODOLOGY: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary 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.