

1-800-533-1710

PATIENT NAME TESTING, FXPB		PATIENT NUMBER L3MRNG9156451		AGE 20Y	SEX M	ACCESSION # G9156451
ORDERING PHYSICIAN		CLIENT ORDER #			ACCOUNT # LIAISONS	
COLLECTION 09/22/10 10:47 A	RECEIVED 09/22/10 10:47 A	REPORT PRINTED 11/22/10 03:08 P		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	DATE TIME	DATE TIME				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI	LO	REF RANGE	PERFORM SITE *
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Fragile X Syndrome, Mol. Analysis

Specimen	Blood	MCR
Specimen ID	1034717	MCR
Order Date	22 Sep 2010 13:01	MCR
Reason For Referral		MCR

Test for the presence of an expansion within the FMR1 gene.

Method MCR

Southern blot and PCR-based assays were used to determine the size and methylation status of the CGG repeat within the FMR1 gene. The Southern blot assay utilized the DNA probe StB12.3 and an Eco RI/Nru I double digest. The reported number of CGG repeats is estimated to be correct to within +/-5%.

Normal: 5-44

Intermediate (Gray Zone): 45-58

Premutation: 59-200

Full mutation: >200

Result	CGG repeat: 29	MCR
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Methylation status: Normal

Final result: Normal

Interpretation MCR

This result is NOT consistent with a diagnosis of fragile X syndrome. The methods described detect >99% of patients affected with fragile X syndrome. Rarely, individuals with fragile X syndrome may have a non-expansion type mutation (e.g., point mutation) not detected by this assay.

Because some chromosome abnormalities have overlapping clinical features with fragile X syndrome, a standard chromosome study is recommended. If one has already been performed, please refer to that separate report for details. A genetic consultation may be of benefit.

* Perform Site Legend on last page of report

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CAUTIONS:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Reviewed By: Heather Lynn Owen
 Release Date: 22 Sep 2010 13:13

MCR
 MCR

* PERFORMING SITE

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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