Patient ID	Patient Name		Birth Date	Gender	Age
SA00006122	TESTINGRNV, REPORTS		1982-01-01	F	33
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00006122	SA00006122	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		17 Dec 2015 00:00			

Fragile X Syndrome, Mol. Analysis

Result Summary

MCR

FULL MUTATION IDENTIFIED

Result

MCR

Number of CGG repeats: >200 (Full Mutation) and 30
Methylation status: Abnormal

Interpretation

1 MCR

Results indicate that this individual is a carrier of fragile X syndrome with an expansion in the full mutation (>200) range. Individuals with a full mutation in the FMR1 gene show a wide range of phenotypic variability. Physical, cognitive, and behavioral characteristics are generally milder in females due to random X inactivation. Cognitive functioning in females can range from normal intelligence to mental retardation.

Since an expansion has been identified within the FMR1 gene, genetic testing of other at risk family members could be considered.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen.

Test results should be interpreted in the 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 false-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.

Multiple in-silico evaluation tools were used to assist in the interpretation of these results. These tools are updated regularly, therefore changes to these algorithms may result in different predictions for a given alteration. However, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Reason for Referral

MCR

Carrier screen for fragile X syndrome. Test for the presence of an expansion within the FMR1 gene.

Specimen

MCR

WB Whole Blood

Method

MCR

PCR-based assays were used to determine the size and methylation status of the CGG repeat in the FMR1 gene. The reported number of CGG repeats is estimated to be correct to within ±5%.

Normal:5–44
Intermediate (Gray Zone): 45–54
Premutation: 55–200
Full mutation: >200

Released By

MCR

W. Edward Highsmith, Jr., Ph.D.

Received: 17 Dec 2015 08:41 **Reported:** 18 Dec 2015 08:49

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

Patient ID	Patient Name		Birth Date	Gender	Age
SA00006122	TESTINGRNV, REPORTS		1982-01-01	F	33
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
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Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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