Patient ID	Patient Name		Birth Date	Gender	Age
SA00001056	SAMPLEREPDMGLM, VLD20150713A0318		1981-01-01	M	34
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00001056	SA00001056	CLIENT, CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		12 Jul 2015 19:53			

Y Microdeletion

Result Summary

MCR

NEGATIVE

Result

MCR

None of the listed markers spanning the AZFa, AZFb, and AZFc regions are deleted.

Interpretation

(1) MCR

No deletion of the AZFa, b, or c regions was identified. This result decreases the likelihood that a microdeletion of the Y chromosome is the cause of infertility in this individual. This assay is expected to detect 95% of AZFa, b, or c deletions thus far reported in the literature. The detection rate for each individual is case specific and dependent on clinical phenotype. For example, approximately 3–15% of males with non-obstructive azoospermia and 6–10% of males with severe oligospermia are predicted to have a microdeletion that would be detected by this assay. Among unselected infertile males, the overall frequency of Yq microdeletions is approximately 3%.

This assay does not exclude the possibility of other genetic causes for male infertility. Because other chromosomal abnormalities may have overlapping clinical features with non-obstructive azoospermia or severe oligospermia, a standard chromosome study is recommended. Also, testing for CFTR gene mutations may also provide additional useful diagnostic information in cases of obstructive azoospermia.

ADDITIONAL INFORMATION

Multiplex PCR is used to test DNA for the presence of microdeletions of the Y chromosome. The specific markers used in this assay are sY86 and sY84 (in region AZFa), sY127 and sY134 (in region AZFb), sY255 and sY254 (in region AZFc). 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. This may not be applicable for all tests.

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 Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Specimen

MCR

WB Whole Blood

Released By

MCR

EMILY LAUER

Received: 13 Jul 2015 20:30 **Reported:** 27 Jul 2015 09:57

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.

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