



Patient ID 321	Patient Name TESTRNV, TESTING ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100427260	Client Order Number X100427260	Ordering Physician 123	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 Aug 2023 06:33		

Result Summary

Negative

Result

MCR

A mitochondrial DNA (mtDNA) deletion was NOT detected.

Interpretation

MCR

This result decreases the likelihood but does not rule out the presence of a mitochondrial DNA (mtDNA) deletion.

Individuals may have a mtDNA deletion that is not detectable by the methods utilized (e.g. a deletion present at a low heteroplasmy level or confined to tissues that were not tested). This assay also does not rule out mitochondrial disease due to single nucleotide variants in mtDNA, mtDNA depletion, or the

presence of pathogenic variants in nuclear genes associated with mitochondrial disease.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing.

A genetic consultation may be of benefit.

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 321	Patient Name TESTRNV, TESTING ATLAS	Birth Date 1968-02-19	Sex M	Age 55
Order Number X100427260	Client Order Number X100427260	Ordering Physician 123	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 Aug 2023 06:33		

Specimen

MCR

WB Whole Blood

Method

MCR

Droplet digital polymerase chain reaction method for determining heteroplasmy for large mitochondrial genome deletions.

See www.mayocliniclabs.com (Test ID DMITO) for additional information about this test.

Disclaimer

1 MCR

CLINICAL CORRELATIONS:

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.

If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of at-risk individuals.

TECHNICAL LIMITATIONS:

This assay will not detect the breakpoints for large mitochondrial deletions or single nucleotide variants that cause mitochondrial

disease. Therefore, the absence of a detectable variant does not rule out the possibility that an individual is affected with mitochondrial disease. This test can only detect mitochondrial DNA (mtDNA) deletions that include the mt-ND4 or mt-ND2 genes.

Some individuals who have a mitochondrial deletion syndrome may have a deletion that is not identified by this assay. The absence of a deletion, therefore, does not eliminate the possibility of mitochondrial DNA deletion syndrome. For predictive testing of asymptomatic individuals, it is important to first document the presence of a deletion in an affected family member.

Of note, absence of a mitochondrial deletion does not rule out the presence of a deletion below the limits of detection of this assay (<10% heteroplasmy).

Rare variants exist that could lead to false-negative or false-positive results. If results obtained do not match clinical findings, additional testing should be considered.

Released By

MCR

Devin Oglesbee, Ph.D.

Received: 18 Aug 2023 15:24

Reported: 30 Aug 2023 09:46

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 US 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