MAYO CLINIC LABORATORIES

1-800-533-1710

DMITO

Mitochondrial DNA Deletion Heteroplasmy, ddPCR, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
321	TESTRNV, TESTING ATLAS		1968-02-19	M	55
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100427261	X100427261	Test,Atlas			
Account Information		**Collected**			
C7028846 DLMP Rochester		17 Aug 2023 08:33			

Result Summary

Positive

Result

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A mitochondrial DNA (mtDNA) deletion was detected with approximate heteroplasmy level-56%

Interpretation

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A large mitochondrial DNA (mtDNA) deletion was detected. Large deletions are consistent with an increased risk of a mitochondrial deletion syndrome. Mitochondrial DNA deletion syndromes include Kearns-Sayre syndrome (KSS), progressive external ophthalmoplegia (PEO), Pearson syndrome, mitochondrial myopathy, and more rarely, Leigh syndrome. Results should be interpreted in the context of clinical findings, family history, and other laboratory testing.

Since a deletion was detected, genetic testing of at risk family members could be considered. Please contact the Genomics Laboratory at 1–800–533–1710 with questions about genetic testing for family members.

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

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Specimen

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WB Whole Blood

Method

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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 15:25 **Reported:** 30 Aug 2023 09:47

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.

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