MAYO CLINIC LABORATORIES

1-800-533-1710

PMPDD

PMP22 Gene, Large Deletion/Duplication Analysis, Varies

Patient ID	Patient Name		Birth Date	Gender	Age
SA00099741	TESTING, REPORTS		1986-01-01	F	31
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00099741	SA00099741	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		26 Oct 2017 00:00			

PMP22 Gene, Deletion/Duplication

Result Summary MCR

POSITIVE

Result MCR

The following deletion was identified:
Exon(s): 1–5
Classification: PATHOGENIC

Interpretation [1] MCR

The deletion of exon(s) 1–5 encompasses the entire PMP22 gene and is consistent with a diagnosis of hereditary neuropathy with liability to pressure palsies (HNPP) for this individual.

Since a mutation has been identified, genetic testing of at risk family members could be considered. Mutation specific testing is available at Mayo Medical Laboratories by ordering FMTT / Familial Mutation, Targeted Testing. Please contact the Genomics Laboratory at 1–800–533–1710 with questions about this test.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

Gene dosage analysis (MLPA) was used to test for the presence of large deletions and duplications in the PMP22 gene, including the common, recurrent whole gene deletions and duplications associated with hereditary neuropathy with liability to pressure palsies (HNPP) and Charcot-Marie-Tooth disease type 1A (CMT1A), respectively (GenBank accession number NM_00304; build GRCh37 (hg19)). Please note, this assay will not detect small deletions or insertions outside of the probe binding regions, single base changes, splicing mutations, or rearrangements outside of targeted regions. Somatic mosaicism and complex higher copy number changes may not quantified by the assay. 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

Niu, Zhiyv, Ph.D.

Received: 26 Oct 2017 13:31 **Reported:** 13 Nov 2017 09:21

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