MAYO CLINIC LABORATORIES

Test Definition: PMPDD

PMP22 Gene, Large Deletion/Duplication Analysis, Varies

Overview

Useful For

Diagnosis of Charcot-Marie-Tooth type 1A or hereditary neuropathy with liability to pressure palsies

Genetics Test Information

This test assesses for large deletions and duplications only.

Testing Algorithm

For more information see Hereditary Peripheral Neuropathy Diagnostic Algorithm.

Special Instructions

- Informed Consent for Genetic Testing
- Hereditary Peripheral Neuropathy Diagnostic Algorithm
- Molecular Genetics: Neurology Patient Information
- Informed Consent for Genetic Testing (Spanish)

Highlights

This test may aid in the diagnosis of Charcot-Marie-Tooth type 1A or hereditary neuropathy with liability to pressure palsies.

Method Name

Dosage Analysis by Polymerase Chain Reaction (PCR)/Multiplex Ligation-Dependent Probe Amplification (MLPA)

NY State Available

Yes

Specimen

Specimen Type

Varies

Shipping Instructions

Specimen preferred to arrive within 96 hours of collection.

Specimen Required

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Molecular Genetics: Neurology Patient Information in Special Instructions
3. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

Specimen Minimum Volume

1 mL

Reject Due To

All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Frozen		
	Refrigerated		

Clinical & Interpretive

Clinical Information

This test is appropriate for individuals with clinical features suggestive of Charcot-Marie-Tooth type 1A (CMT1A) and hereditary neuropathy with liability to pressure palsies (HNPP).

CMT1A is a dominantly inherited disease characterized progressive distal muscle weakness and atrophy, sensory loss, and slow nerve conduction velocity starting early in life. Duplications of the *PMP22* gene are associated with CMT1A.

Deletions of *PMP22* are associated with hereditary neuropathy with liability to pressure palsies (HNPP), a dominantly inherited disease resulting in peripheral neuronal demyelination. HNPP is characterized clinically by recurrent focal motor and sensory neuropathy in a single nerve that can manifest as numbness, muscular weakness, and atrophy.

Reference Values

An interpretive report will be provided.

Interpretation

All detected alterations are evaluated according to American College of Medical Genetics and Genomics recommendations.(1) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions

In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

This test may not detect deletions/duplications present in very low levels of mosaicism.

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.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

Clinical Reference

1. Richards S, Aziz N, Bale S, et al: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med 2015 May;17(5):405-424
2. van Paassen BW, van der Kooi AJ, van Spaendonck-Zwarts KY, et al: PMP22 related neuropathies: Charcot-Marie-Tooth disease type 1A and Hereditary Neuropathy with liability to Pressure Palsies. Orphanet J Rare Dis 2014 Mar 19;9:38
3. Li J, Parker B, Martyn C, et al: The *PMP22* Gene and Its Related Diseases. Mol Neurobiol 2013 April;47(2):673-698

Performance

Method Description

Multiple ligation-dependent probe amplification (MLPA) is utilized to test for the presence of large deletions and duplications within the *PMP22* gene.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Batched 1 time per week

Report Available

14 to 21 days

Specimen Retention Time

Whole Blood: 2 weeks (if available) Extracted DNA: Indefinitely

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification

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

CPT Code Information

81324

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
PMPDD	PMP22 Gene, Deletion/Duplication	75384-8

Result ID	Test Result Name	Result LOINC® Value
113371	Result Summary	50397-9
113374	Result	75384-8
113375	Interpretation	69047-9
113376	Additional Information	48767-8
113377	Specimen	31208-2
113378	Source	31208-2
113379	Released By	18771-6