
Lab Dept: **Anatomic Pathology**

Test Name: **PMP22 DUPLICATION/DELETION**

General Information

Lab Order Codes: PMPD

Synonyms: HNPP, Hereditary Neuropathy with liability to Pressure Palsies, Charcot-Marie-Tooth Disease 1A

CPT Codes: 81324 – PMP22 gene analysis; deletion/duplication analysis

Test Includes: PMP22 Deletion/Duplication analysis with an interpretive report.

Logistics

Test Indications: Use to detect rearrangements in the PMP22 gene. This test performs both duplication and deletion analysis to detect the cause of CMT1A and HNPP.

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: Athena Diagnostics (Athena Test: 131)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Results are reported in 14-28 days

Special Instructions: Note if patient is symptomatic or asymptomatic. If the patient is asymptomatic, complete and submit "[Informed Consent form](#)" with the specimen.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 8 mL (Minimum: 1 mL) blood

Processed Volume: Same as Draw Volume

Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature. Forward promptly. Minimum specimen volume makes it impossible to repeat the test or perform confirmatory or perform reflex testing. In some situations, a minimum specimen volume may result in a QNS (quantity not sufficient) result, requiring a second specimen to be collected.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	N/A
Methodology:	Multiplex Ligation-dependent Probe Amplification
References:	Athena Diagnostics March 2018 (800) 394-4493 Fax: (508) 753-5601
Updates:	2/7/2013: CPT update 4/7/2015: Volume/TAT update