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**Lab Dept:**                    **Anatomic Pathology**

**Test Name:**                **PMP22 DUPLICATION/DELETION**

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***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.

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***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.

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***Specimen***

**Specimen Type:**            Whole blood

**Container:**                 Lavender top (EDTA) tube

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

**Processed Volume:**       Same as Draw Volume

<b>Collection:</b>	Routine blood collection
<b>Special Processing:</b>	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.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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### ***Interpretive***

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