ARUP LABORATORIES
A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CHARCOT-MARIE-TOOTH (CMT) AND HEREDITARY NEUROPATHY TESTING

Patient Name: _____ **Date of Birth:** _____ **Sex:** ☐ **F** ☐ **M**

Physician: _____ **Physician Phone:** _____

Practice Specialty: _____ **Physician Fax:** _____

Genetic Counselor: _____ **Counselor Phone:** _____

Patient's Ethnicity (check all that apply)

☐ African American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Suspected Diagnosis; Age of Onset: _____

☐ Charcot-Marie-Tooth disease (CMT); type: _____ ☐ Hereditary sensory neuropathy (HSN)
☐ Hereditary motor neuropathy (HMN) ☐ Transthyretin amyloidosis
☐ Hereditary neuropathy with liability to pressure palsies (HNPP) ☐ Other: _____

Does the patient have symptoms of CMT/hereditary neuropathy? ☐ No ☐ Yes (check all that apply)

☐ Abnormal gait/difficulty walking
☐ Abnormal reflexes
☐ Carpal tunnel syndrome
☐ Foot drop
☐ Hearing loss
☐ High arches/pes cavus deformity
☐ Hip dysplasia
☐ Spastic paraplegia
☐ Muscle wasting; (specify) ☐ proximal ☐ distal
☐ Muscle weakness; (specify) ☐ progressive ☐ stable ☐ proximal ☐ distal
☐ Peripheral neuropathy; (specify) ☐ sensory ☐ motor
☐ Transient/recurring focal pressure neuropathy
☐ Other: _____

Diagnostic Studies:

☐ Electromyography (EMG): ☐ Normal ☐ Abnormal—describe: _____
☐ Nerve conduction velocity (NCV): ☐ Normal ☐ Demyelinating (<38 m/s) ☐ Axonal (>38 m/s) ☐ Intermediate (25–45 m/s)
☐ Other: _____

Has the patient undergone previous DNA testing for CMT/hereditary neuropathy? ☐ No ☐ Yes ☐ Unknown

☐ *PMP22* deletion/duplication; result: _____
☐ Chromosomal microarray; result: _____
☐ Other testing; describe test(s) and results: _____

Is there any relevant family history of neuropathy? ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset: _____

_____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown
If yes, attach a copy of the relative's DNA laboratory result (REQUIRED) for familial mutation testing).

PATIENT HISTORY FOR CHARCOT-MARIE-TOOTH (CMT) AND HEREDITARY NEUROPATHY TESTING

Check the test you intend to order.

Recommended testing for CMT1 or HNPP

☐ **2012160 Charcot-Marie-Tooth Type 1A (CMT1A)/Hereditary Neuropathy with Liability to Pressure Palsies (HNPP), *PMP22* Deletion/Duplication:** Clinical sensitivity 70–80% for CMT1; 80% for HNPP

☐ **2012155 Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, *PMP22* Deletion/Duplication with Reflex to Sequencing Panel:** *PMP22* deletion/duplication with reflex to sequencing of hereditary neuropathy genes

Recommended testing for CMT subtypes/hereditary neuropathies OTHER than CMT1A/HNPP

☐ **2012151 Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies Panel, Sequencing**

Follow-up targeted testing for family members (copy of relative's lab result is REQUIRED)

☐ **2012160 Charcot-Marie-Tooth 1A (CMT1A)/Hereditary Neuropathy with Liability to Pressure Palsies (HNPP), *PMP22* Deletion/Duplication**: For known familial deletion/duplication previously identified in a relative

☐ **2001961 Familial Mutation, Targeted Sequencing**: Tests for a sequence variant previously identified in a relative; a copy of relative's lab result is REQUIRED.

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.

Master Label

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