

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform molecular genetic testing.
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 ____/____/____ Gender F M

Physician _____ Physician Phone (____) _____ Practice Specialty _____

Genetic Counselor _____ Counselor Phone (____) _____

Patient's Ethnicity (check all that apply)

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

Suspected diagnosis:

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

Age of onset: _____

Does the patient have SYMPTOMS of CMT/hereditary neuropathy? No Yes; please check all that apply

- Abnormal gait/difficulty walking Muscle wasting; proximal distal
 Abnormal reflexes Muscle weakness; progressive stable; proximal distal
 Foot drop Peripheral neuropathy; sensory motor
 Hearing loss Spastic paraplegia
 High arches/pes cavus deformity Transient/recurrent focal pressure neuropathy
 Hip dysplasia 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: _____

Does the patient have a FAMILY HISTORY of neuropathy? No Yes Unknown

If yes, attach a PEDIGREE or specify the relatives' RELATIONSHIP to the patient. List their symptoms & age of onset:

Has DNA testing been performed for these family member(s)? No Yes Unknown

If yes, describe test(s) and results (a copy of relative's report is required for targeted testing): _____

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

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

Circle the test you intend to order:

Recommended testing for CMT1A 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 78 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
Targeted testing for known mutation (laboratory report from family member REQUIRED)	
2001961	Familial Mutation, Targeted Sequencing- targeted testing for a known familial sequence mutation
2012160	Charcot-Marie-Tooth 1A (CMT1A)/Hereditary Neuropathy with Liability to Pressure Palsies (HNPP), PMP22 Deletion/Duplication - for known familial deletion/duplication

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

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