

Hereditary Neuropathies: Deletion/Duplication Panel

Test Code: MD350

Turnaround time: 2 weeks

CPT Codes: 81324 x1, 81404 x1, 81405 x1, 81479 x1

Condition Description

Hereditary neuropathies are a collection of inherited disorders affecting the peripheral nervous system. The hereditary neuropathies are divided into four major subcategories: hereditary motor and sensory neuropathy, hereditary sensory neuropathy, hereditary sensory and autonomic neuropathy, and hereditary motor neuropathy. Charcot-Marie-Tooth disease, is of the most common types of the hereditary motor and sensory neuropathies. Clinical presentation typically includes sensory symptoms like pain in the feet and hands, motor symptoms such as weakness in the lower leg and feet muscles. Some hereditary neuropathies can affect the autonomic nerves, resulting in impaired sweating, postural hypotension, or insensitivity to pain.

The estimated prevalence of hereditary neuropathies is about 1 in 2500 individuals. A myriad of genes are associated with hereditary neuropathies. Genetic testing has therefore become an important tool in the diagnosis of neuropathies.

References:

- Sagnelli et al. (2013) *J Neurol.* 260(10):2684-90.
- Auer-Grumbach M. (2013) *Handb Clin Neurol.* 115:893-906.
- Murphy et al. (2013) *Handb Clin Neurol.* 2013;115:213-32.

Genes

[AARS1](#), [ALDH3A2](#), [APTIX](#), [ATL1](#), [ATM](#), [ATP7A](#), [BSCL2](#), [CACNB4](#), [COQ8A](#), [CTDP1](#), [DCTN1](#), [DNAJB2](#), [DNM2](#), [DNMT1](#), [DYNC1H1](#), [EGR2](#), [ELP1](#), [FGD4](#), [FGF14](#), [FIG4](#), [FXN](#), [GAN](#), [GARS1](#), [GDAP1](#), [GJB1](#), [GLA](#), [HOXD10](#), [HSPB1](#), [HSPB8](#), [IGHMBP2](#), [ITPR1](#), [KCNA1](#), [KCNC3](#), [KIF1A](#), [KIF1B](#), [KIF5A](#), [L1CAM](#), [LITAF](#), [LMNA](#), [LRSAM1](#), [MED25](#), [MFN2](#), [MPZ](#), [MRE11](#), [MTMR2](#), [MTTP](#), [NDRG1](#), [NEFL](#), [NGF](#), [NIPA1](#), [NTRK1](#), [PEX7](#), [PHYH](#), [PLEKHG5](#), [PLP1](#), [PMP22](#), [PNPLA6](#), [POLG](#), [PRKCG](#), [PRPS1](#), [PRX](#), [RAB7A](#), [REEP1](#), [RETREG1](#), [SACS](#), [SBF2](#), [SCN9A](#), [SH3TC2](#), [SIL1](#), [SLC12A6](#), [SLC1A3](#), [SPART](#), [SPAST](#), [SPG11](#), [SPG21](#), [SPG7](#), [SPTBN2](#), [SPTLC1](#), [SPTLC2](#), [TDP1](#), [TRPV4](#), [TTBK2](#), [TPA](#), [TTR](#), [TWNK](#), [WASHC5](#), [WNK1](#), [YARS1](#), [ZFYVE26](#), [ZFYVE27](#)

Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of a neuropathy.

Methodology

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Detection

Deletion/Duplication: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Specimen Requirements

Submit only 1 of the following specimen types

Type: DNA, Isolated

Specimen Requirements:

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

Type: Whole Blood (EDTA)

Specimen Requirements:

EDTA (Purple Top)

Infants and Young Children (2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

Specimen Collection and Shipping:

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

Related Tests

- Hereditary Neuropathies: Sequencing Panel