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 the most common type of these 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:

### Genes

AARS, ALDH3A2, APTX, AT1, ATM, ATP7A, BSCL2, CACNB4, COQ8A, CTDP1, DCTN1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, ELP1, FGD4, FGF14, FIG4, FAN, GAN, GARS, GDAP1, GJB1, GLA, HOXD10, HSPB1, HSPB8, IGHEBP2, ITPR1, KCNA1, KCNC3, KIF1A, KIF1B, KIF5A, L1CAM, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MRE11, MTMR2, MTPP, NDRG1, NEFL, NGF, NIPA1, NTRK1, PEX7, PHYH, PLEKHG5, PLP1, PMP22, PNPLA6, POLG, PRKCS, PRPS1, PRX, RAD27A, REEP1, RETREG1, SAC53, SPB2, SCN8A, SH3TC2, SIL1, SLC12A6, SLC1A3, SPART, SPAST, SPG11, SPG21, SPG7, SPTBN2, SPTLC1, SPTLC2, TDP1, TRPV4, TTBK2, TTPA, TTR, TWNK, WASHC5, WNK1, YARS, 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. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

### 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: Whole Blood**

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

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Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Hereditary Neuropathies: Sequencing Panel