Hereditary Peripheral Neuropathy Diagnostic Algorithm

Inherited neuropathy suspected
The following tests should be performed in all adult patients:
HbA1c or glucose tolerance, vitamin B12, monoclonal proteins.

Nerve conduction studies show:
Ulnar motor forearm nerve conduction velocity <38 m/s (compound muscle action potential test >0.5 mV)
AND/OR
blink reflex R1 >13 ms
OR
Hereditary neuropathy with pressure palsies (HNPP) is suspected (diffuse sensory nerve conduction velocity slowing, motor conduction blocks at points of compression, prolonged motor nerve distal latencies)

Order PMPDD / PMP22 Gene, Large Deletion and Duplication Analysis

EITHER <50 years of age at onset with no obvious acquired cause OR
Definitive family history

Order as appropriate:
■ For peripheral neuropathy, order PEPAN / Comprehensive Peripheral Neuropathy Gene Panel
■ For peripheral neuropathy and distal myopathy, order DWFPAN / Comprehensive Distal Weakness Gene Panel
■ For peripheral neuropathy and/or neuromuscular junction and/or muscle involvement, order CGPH for PEPAN plus MUPAN (Gene List ID: Neurology-LXM917)
OR
See Tests To Consider box for more options

Tests To Consider
■ IMSN / Inherited Motor and Sensory Neuropathy Gene Panel
■ IMNP / Inherited Motor Neuropathy Gene Panel
■ ISNP / Inherited Sensory Neuropathy Gene Panel
■ CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing; stating disease state: Neurology Disorder

Negotiate:
EITHER <50 years of age at onset with no obvious acquired cause OR
Definitive family history

Disorder-specific management

Predominant motor neuropathy (length dependent)

Type of neuropathy

Common acquired sensory neuropathy excluded
Consider ordering one of the following:
■ AIAES / Axonal Neuropathy, Autoimmune/Paraneoplastic Evaluation, Serum
■ PEPAN / Comprehensive Peripheral Neuropathy Gene Panel
■ Testing for RFC1 - CANVAS / Spectrum Disorder

Disorder-specific management

Consider: WESDX / Whole Exome Sequencing for Hereditary Disorders or whole genome sequencing

Disorder-specific management

Diagnostic of chronic idiopathic axonal polyneuropathy (CIAP)

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