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 PMP22 Gene, Large Deletion and Duplication Analysis

Disorder-specific management

Charcot-Marie-Tooth disease type 1A (CMT1A) – PMP22 duplication
HNPP – PMP22 deletion

HNPP missense mutations

Consider: PNPAN / Peripheral Neuropathy Expanded Panel by Next-Generation Sequencing (NGS)

Disorder-specific management

Consider: WES / Whole Exome Sequencing or whole genome sequencing

Diagnostic of chronic idiopathic axonal polyneuropathy (CIAP)
OR
Consider ordering 1 of the following:
• HSINP / Hereditary Sensory/Autonomic Neuropathy Panel by Next-Generation Sequencing (NGS)
• PNPAN / Peripheral Neuropathy Expanded Panel by Next-Generation Sequencing (NGS)