

Quarterly HOTLINE: Effective November 12, 2018

2012155 Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, *PMP22*

CMT REFLEX

Deletion/Duplication with Reflex to Sequencing Panel

Performed: Varies

Reported: Within 2 weeks, if reflexed add 3-6 weeks

Note: Deletion/Duplication analysis is performed on all samples. If no large deletions or duplications are detected and/or results do not explain the clinical scenario, then sequencing of the Charcot-Marie-Tooth and Related Hereditary Neuropathy gene will be added. Additional charges apply. If reflexed, an additional 8-10 weeks is required to complete testing.

Genes sequenced: AARS, AIFMI, ARHGEF10, ATL1, ATP7A, BAG3, BICD2, BSCL2, CCT5, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HEXA, HINT1, HK1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LAS1L, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, MYH14, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRNP, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN9A, SETX, SH3TC2, SLC12A6, SLC5A7, SOX10, SPTLC1, SPTLC2, TDP1, TFG, TRIM2, TRPV4, WNK1, YARS.