

Neuropathia (Neuropathy) génpanel v:2023.10.12.

Panel tartalma 347 gén:

AARS, AARS1, ABCA1, ABCC9, ABHD12, ACTC1, ACTN2, AGTPBP1, AGXT, AIFM1, ALDH3A2, AMACR, ANKRD1, AP1S1, APOA1, APTX, ARHGEF10, ARL6IP1, ARSA, ASAH1, ATAD3A, ATL1, ATL3, ATM, ATP1A1, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BAG3, BCKDHB, BICD2, BRAF, BSCL2, C12orf65, C19orf12, C1orf194, CACNB4, CASQ2, CAV3, CCT5, CD59, CHCHD10, CLTCL1, CNTNAP1, COA7, COQ8A, COX10, COX6A1, CPOX, CRYAB, CTDPI, CYP27A1, CYP7B1, CSRP3, DARS2, DCAF8, DCTN1, DEGS1, DES, DGUOK, DHH, DHTKD1, DMD, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DTNA, DYNC1H1, EGR2, ELP1, EMD, ERBB3, ERCC6, ERCC8, ETFDH, EXOSC9, FA2H, FAH, FAM126A, FAM134B, FBLN5, FBXO38, FGD4, FGF14, FIG4, FKTN, FLVCR1, FMR1, FXN, GAA, GALC, GAN, GARS, GARS1, GATAD1, GBA2, GDAP1, GJB1, GJC2, GLA, GLE1, GNB4, GNE, GSN, HADHA, HADHB, HARS, HEXA, HINT1, HK1, HMBS, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, IARS2, IGHMBP2, IKBKAP, INF2, ITPR1, JAG1, JPH2, JUP, KARS, KARS1, KCNA1, KCNA2, KCNC3, KIF1A, KIF1B, KIF5A, KLC2, KRAS, L1CAM, LAMA4, LAMP2, LAS1L, LDB3, LITAF, LMNA, LRSAM1, LYST, MAP1B, MAP2K1, MAP2K2, MARS, MARS1, MCM3AP, MED25, MEGF10, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTMR2, MTTP, MYBPC3, MYH14, MYH6, MYH7, MYL2, MYL3, MYOT, MYOZ2, MYPN, NAGA, NAGLU, NDRG1, NEBL, NEFH, NEFL, NEXN, NGF, NIPA1, NOP56, NRAS, NTRK1, OPA1, OPA3, PDHA1, PDK3, PDLIM3, PDYN, PEX10, PEX7, PHYH, PKP2, PLEKHG5, PLN, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLG2, POLR3A, PPOX, PPP2R2B, PRDM12, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, RAF1, RBM20, REEP1, RETREG1, RIT1, RYR2, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN5A, SCN9A, SCP2, SCYL1, SELENOI, SEPT9, SETX, SGCD, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC25A19, SLC25A46, SLC52A1, SLC52A2, SLC52A3, SLC5A7, SMAD3, SMN1, SMN2, SORD, SOS1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TAZ, TCAP, TDP1, TFG, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRIM2, TRPA1, TRPV4, TTBK2, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VAPB, VCL, VCP, VPS13A, VRK1, VWA1, WARS, WASHC5, WNK1, XK, XPA, XRCC1, YARS, YARS1, ZFYVE26, ZFYVE27

Forrás:

Hereditary neuropathy (Version 1.466)

<https://panelapp.genomicsengland.co.uk/panels/85/> (Megtekintve: 2023.05.18.)

Invitae Comprehensive Neuropathies Panel

<https://www.invitae.com/en/providers/test-catalog/test-03200> (Megtekintve: 2023.05.18.)

Comprehensive Neuropathy Panel

<https://www.preventiongenetics.com/testInfo?val=Comprehensive-Neuropathy-Panel>

(Megtekintve: 2023.05.18.)

Charcot-Marie-Tooth Neuropathy Panel

<https://blueprintgenetics.com/tests/panels/neurology/charcot-marie-tooth-neuropathy-panel/>

(Megtekintve: 2023.05.18.)