Spasztikus paraplegia (Spastic paraplegia) génpanel v:2023.10.12.

Panel tartalma 516 gén:

AARS2, ABCB7, ABCD1, ABHD12, ABHD16A, ABHD5, ACAD9, ACADVL, ACER3, ACO2, ACP33, ADAR, ADPRS, AFG3L2, AGK, AGTPBP1, AHI1, AIFM1, AIMP1, ALAS2, ALDH18A1, ALDH3A2, ALDH5A1, ALK, ALS2, AMACR, AMPD2, ANO10, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARL13B, ARL6, ARL6IP1, ARSA, ARSI, ATAD3A, ATCAY, ATL1, ATM, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B4GALNT1, B9D1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAS3, BCKDHA, BCKDHB, BCS1L, BICD2, BLOC1S1, BOLA3, BSCL2, BTD, C12orf65, C19orf12, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CASK, CC2D2A, CCDC88C, CCT5, CDK16, CEP290, CEP41, CHMP1A, CHP1, CLCN2, CLN5, CLN6, CLPB, CLPP, COA6, COA7, COA8, COASY, COL4A1, COL4A2, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, CP, CPLANE1, CPT1C, CTNNB1, CWF19L1, CYC1, CYP27A1, CYP2U1, CYP7B1, CSPP1, CSTB, DAB1, DARS, DARS1, DARS2, DBT, DDHD1, DDHD2, DDX3X, DGUOK, DHPS, DLAT, DLD, DNA2, DNAJC19, DNAJC5, DNM1L, DNMT1, DOCK3, DSTYK, EARS2, EBF3, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELOVL1, ELOVL4, ELOVL5, ENTPD1, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, FA2H, FAR1, FARS2, FASTKD2, FAT2, FBXL4, FDX2, FDXR, FGF14, FH, FLAD1, FLVCR1, FOXRED1, FTL, FXN, GAD1, GALC, GARS1, GBA, GBA2, GBE1, GCDH, GCH1, GFAP, GFER, GFM1, GFM2, GJA1, GJB1, GJC2, GLRX5, GOSR2, GPT2, GRID2, GRM1, GSS, GTPBP3, HACE1, HARS2, HEPACAM, HEXA, HEXB, HIBCH, HIKESHI, HMGCL, HPDL, HSPD1, HTRA2, HTT, IARS2, IBA57, IFIH1, INPP5E, IRF2BPL, ISCA2, ISCU, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KDM5C, KIAA0196, KIDINS220, KIF1A, KIF1C, KIF5A, KIF7, KLC2, KLC4, KPNA3, L1CAM, L2HGDH, LAMA1, LAMP2, LARS2, LIAS, LIPT1, LMNB1, LRPPRC, LYRM7, LYST, MAG, MAPK8IP3, MARS, MARS1, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MKKS, MKS1, MLC1, MOCS1, MPC1, MPV17, MRE11, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MTFMT, MTO1, MTPAP, MTRFR, MTTP, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NIPA1, NKX6-2, NPC1, NPC2, NPHP1, NR2F1, NRCAM, NSRP1, NT5C2, NUBPL, OFD1, OPA1, OPA3, OPHN1, OTC, PAH, PANK2, PARS2, PAX6, PC, PCCA, PCCB, PCDH12, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX10, PEX2, PEX7, PGAP1, PGN, PHYH, PI4KA, PIK3R5, PLA2G6, PLP1, PMPCA, PNKD, PNKP, PNPLA6, PNPT1, POLG, POLG2, POLR3A, POLR3B, POLR3K, PPP2R2B, PRICKLE1, PRKCG, PRRT2, PSEN1, PUM1, QARS1, RAB3GAP2, RARS, RARS1, RARS2, REEP1, REEP2, RHOB, RMND1, RNASEH1, RNASEH2B, RNF170, RNF216, RNU7-1, RPGRIP1L, RRM2B, RTN2, RUBCN, SACS, SAMD9L, SARS2, SCN1A, SCN2A, SCO1, SCO2, SCYL1, SDHA, SDHAF1, SELENOI, SERAC1, SETX, SFXN4, SIL1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A3, SLC1A4,

SLC20A2, SLC22A5, SLC25A15, SLC25A19, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC9A6, SNX14, SPART, SPAST, SPATA5L1, SPG11, SPG14, SPG16, SPG19, SPG20, SPG21, SPG24, SPG25, SPG27, SPG29, SPG32, SPG34, SPG36, SPG37, SPG38, SPG41, SPG7, SPR, SPTAN1, SPTBN2, STN1, STUB1, SUCLA2, SUCLG1, SURF1, SYNE1, TACO1, TAF8, TARS2, TBC1D24, TBP, TCTN1, TCTN2, TCTN3, TDP1, TECPR2, TFG, TGM6, TH, TIMM8A, TK2, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM63C, TMEM67, TMEM70, TPK1, TPP1, TRIM32, TRIT1, TRMT10C, TRNT1, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC8, TTPA, TTR, TUBB4A, TUFM, TWNK, TYMP, UBA5, UBAP1, UBE3A, UBTF, UCHL1, UNC80, UQCC2, UQCRB, UQCRC2, UQCRQ, USP8, VAMP1, VARS2, VCP, VLDLR, VPS37A, VRK1, WASHC5, WDR45, WDR45B, WDR48, WDR81, WFS1, WWOX, YARS2, ZEB2, ZFYVE26, ZFYVE27, ZNF423

Forrás:

Childhood onset hereditary spastic paraplegia (Version 4.8)

https://panelapp.genomicsengland.co.uk/panels/568/ (Megtekintve: 2023.05.18.)

Spastic paraplegia - PS303350 - 83 Entries

https://www.omim.org/phenotypicSeries/PS303350?sort=geneSymbols (Megtekintve: 2023.05.18.)

Ataxia / Spastic paraplegia panel

https://www.centogene.com/diagnostics/our-tests/ngs-panels/neurology (Megtekintve: 2023.07.19.)

Spastic Paraplegia Panel

https://blueprintgenetics.com/tests/panels/neurology/spastic-paraplegia-panel/ (Megtekintve: 2023.05.18.)