

Ataxia génpanel v:2023.10.12.

Panel tartalma 633 gén:

AAAS, AARS, AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADVL, ACO2, ADAR, ADCK3, ADGRG1, ADPRHL2, ADPRS, AFG3L2, AGK, AGTPBP1, AHI1, AIFM1, AIMP1, ALAS2, ALDH18A1, ALDH5A1, ALG1, ALG10, ALG11, ALG13, ALG14, ALG2, ALG6, ALG8, ALS2, AMACR, AMPD2, ANO10, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARSA, ASAH1, ATCAY, ATG7, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6V0A1, ATP7A, ATP7B, ATP8A2, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8, AUH, B3GALNT2, B3GLCT, B4GALNT1, B4GALT1, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BEAN1, BICD2, BOLA3, BRF1, BSCL2, BTB, C10ORF2, C12ORF4, C12orf65, C19orf12, C1GALT1C1, C2CD3, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAD, CAMLG, CAMTA1, CAPN1, CAPRIN1, CARS2, CASK, CBY1, CC2D2A, CCDC28B, CCDC88C, CDK5, CEP104, CEP290, CEP41, CHCHD10, CHMP1A, CHP1, CHST3, CHSY1, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPP, COA7, COASY, COG2, COG4, COG5, COG8, COQ2, COQ4, COQ6, COQ8A, COX20, CP, CTBP1, CTSA, CYP27A1, CYP2U1, CYP7B1, CSPP1, CSTB, DAB1, DAG1, DARS2, DCC, DDHD2, DDOST, DDX59, DHDDS, DHPS, DKC1, DMXL2, DNAJC19, DNAJC5, DNMT1, DOCK3, DOLK, DPM2, DPYSL5, DYNC1H1, EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EOGT, EPM2A, ERCC4, ETHE1, EVC, EVC2, EXOC3L2, EXOC8, EXOSC1, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAM149B1, FARS2, FASTKD2, FAT2, FBXL4, FDX2, FDXR, FGF14, FH, FKTN, FLAD1, FLVCR1, FMR1, FOLR1, FOXRED1, FRMD4A, FRMD5, FTL, FUK, FXN, GAD1, GALT, GALNT12, GARS1, GBA, GBA2, GCDH, GCH1, GFAP, GFER, GFM1, GFM2, GJB1, GJC2, GLRA1, GLRB, GLRX5, GLS, GMPPB, GOSR2, GPAA1, GRID2, GRM1, GRN, GSS, GTPBP3, HACE1, HARS, HARS2, HEATR5B, HEPACAM, HEXA, HEXB, HIBCH, HMGCL, HSPD1, HTRA2, HTT, IARS2, IBA57, IFT74, INPP5E, IRF2BPL, ISCA2, ISCU, ISPD, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNN2, KDM5C, KIAA0556, KIDINS220, KIF14, KIF1A, KIF1C, KIF5A, KIF7, L1CAM, LAMA1, LAMP2, LARGE1, LARS2, LFNG, LIAS, LIG3, LIPT1, LMNB1, LRPPRC, LRSAM1, LYRM7, LYST, MAG, MAPK8IP3, MARS1, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MKKS, MKS1, MLC1, MMACHC, MOCS1, MPC1, MPV17, MRE11, MRPL3, MRPL44, MRPS16, MRPS22, MSTO1, MT-ATP6, MTFMT, MTO1, MTPAP, MTRFR, MTPP, MVK, NAGLU, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUF3B, NDUF3S1, NDUF3S2, NDUF3S3, NDUF3S4, NDUF3S6, NDUF3S7, NDUF3S8, NDUFV1, NDUFV2, NFU1, NHLRC1, NIPA1, NKX2-1, NKX2-2, NMNAT2, NOL3, NOP56, NOTCH3, NPC1, NPC2, NPHP1, NPTX1, NR2F1, NT5C2, NUBPL, NUS1, OFD1, OGDHL, OPA1, OPA3, OPHN1, OSTC, OTC, PACS2, PANK2, PARS2, PAX2, PAX6, PC, PCCA, PCCB, PCLO, PDE6D, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX10, PEX16, PEX2, PEX6, PEX7, PGAP1, PHGDH, PHYH, PIGM, PIK3R5, PLA2G6, PLP1, PMPCA, PMPCB, PNKD, PNKP,

PNPLA6, PNPT1, POC1B, POLG, POLG2, POLR3A, POLR3B, POLR3K, POMGNT2, POMK, POMT1, POMT2, POU4F1, PPP2R2B, PRDM13, PRDX3, PRICKLE1, PRKCG, PRNP, PRPS1, PRRT2, PTF1A, PTRH2, PUM1, QARS1, RAB3GAP2, RABL2A, RARS1, RARS2, REEP1, REEP2, RELN, RFT1, RFXANK, RMND1, RNASEH1, RNASEH2B, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RRM2B, RTN2, RUBCN, SACS, SAMD9L, SAR1B, SARS2, SCLT1, SCN1A, SCN2A, SCN8A, SCO1, SCO2, SCYL1, SDHA, SDHAF1, SEC23B, SELENOI, SEPSECS, SERAC1, SETX, SFXN4, SIL1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A19, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC44A1, SLC52A2, SLC52A3, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, STUB1, SUCLA2, SUCLG1, SUFU, SURF1, SVBP, SYNE1, SYT14, TACO1, TAPT1, TARS2, TBC1D23, TBC1D24, TBC1D32, TBP, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TERT, TFG, TGM6, THG1L, TIMM8A, TINF2, TK2, TMEM106B, TMEM107, TMEM126B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM5, TMEM67, TMEM70, TOE1, TOGARAM1, TPK1, TPP1, TPR, TRAPPC11, TRIM32, TRIP4, TRIT1, TRMT10C, TRNT1, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC8, TTPA, TTR, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXNDC15, TYMP, UBA5, UBAP1, UBE3A, UBR4, UBTF, UCHL1, UNC80, UQCC2, UQCRB, UQCRC2, UQCRQ, USP8, VAMP1, VARS2, VCP, VLDLR, VPS13B, VPS13D, VPS37A, VPS41, VPS53, VRK1, VWA3B, WASHC5, WDPCP, WDR45, WDR45B, WDR63, WDR73, WDR81, WFS1, WWOX, XRCC1, XYLT1, XYLT2, YARS2, ZFYVE26, ZFYVE27, ZNF423, ZNF592, ZSWIM6

Forrás:

Hereditary ataxia and cerebellar anomalies - childhood onset (Version 12.17)

<https://panelapp.genomicsengland.co.uk/panels/488/> (Megtekintve: 2023.05.16.)

Hereditary ataxia with onset in adulthood (Version 4.9)

<https://panelapp.genomicsengland.co.uk/panels/466/> (Megtekintve: 2023.05.16.)

Ataxia and cerebellar anomalies - narrow panel (Version 4.7)

<https://panelapp.genomicsengland.co.uk/panels/477/> (Megtekintve: 2023.05.16.)

Ataxia NGS Panel

<https://www.fulgentgenetics.com/Ataxia> (Megtekintve: 2023.05.16.)

Ataxia Panel

<https://blueprintgenetics.com/tests/panels/neurology/ataxia-panel/> (Megtekintve: 2023.05.16.)

Neurology

<https://www.centogene.com/diagnostics/our-tests/ngs-panels/neurology> (Megtekintve:
2023.07.18.)