

Ataxia génpanel v:2022.02.09.

Panel tartalma 543 gén:

AAAS, AARS, AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ABHD5, ACADVL, ACO2, ADCK3, ADCY5, ADGRG1, ADPRHL2, AFG3L2, AGTPBP1, AHI1, ALAS2, ALDH5A1, ALG1, ALG10, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALS2, AMACR, AMPD2, ANO10, AP1S2, APTX, ARL13B, ARL3, ARL6, ARMC9, ARSA, ASAH1, ATAD3A, ATCAY, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6AP1, ATP6V0A2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, BRF1, BSCL2, BTBD, C10ORF2, C12ORF4, C12orf65, C19orf12, C1GALT1C1, C2CD3, C5orf42, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAD, CAMTA1, CAPN1, CASK, CC2D2A, CCDC115, CCDC88C, CDK5, CENPF, CEP104, CEP290, CEP41, CHCHD10, CHMP1A, CHP1, CHST14, CHST3, CHST6, CHSY1, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, COA7, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COQ2, COQ4, COQ6, COQ8A, COQ9, COX20, CP, CPLANE1, CRB2, CTBP1, CTDPL1, CTSA, CWF19L1, CYP27A1, CYP2U1, CYP7B1, CSGALNACT1, CSPP1, CSTB, DAB1, DAG1, DARS2, DCC, DDHD2, DDOST, DDX59, DHCR7, DHDDS, DHPS, DKC1, DLAT, DMXL2, DNAJC19, DNAJC5, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYSL5, DYNC1H1, EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EOGT, EPM2A, ERCC4, EVC, EVC2, EXOC3L2, EXOC8, EXOSC1, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAM149B1, FBXL4, FDXR, FGF14, FKR, FKTN, FLVCR1, FMR1, FOLR1, FRMD4A, FTL, FUK, FUT8, FXN, G6PC3, GALT, GALNT12, GALNT2, GALNT3, GBA, GBA2, GDAP2, GEMIN5, GFAP, GFPT1, GJB1, GJC2, GLI3, GLRA1, GLRB, GLS, GMPPA, GMPPB, GNE, GORAB, GOSR2, GPAA1, GRID2, GRM1, GSS, HARS, HARS2, HEATR5B, HEPACAM, HEXA, HEXB, HIBCH, HTT, HYLS1, ICK, INPP5E, IRF2BPL, ISPD, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNK18, KCNN2, KCNQ2, KCNQ3, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LARGE1, LARS2, LFNG, LIG3, LMNB1, LNP, LRPPRC, LRSAM1, MAGT1, MAN1B1, MAPK8IP3, MARS2, MECP, MFN2, MGAT2, MGME1, MINPP1, MKKS, MKS1, MLC1, MMACHC, MME, MOGS, MORC2, MPDU1, MPI, MRE11, MRE11A, MSTO1, MTCL1, MTFMT, MTPAP, MVK, NAGLU, NAXE, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NGLY1, NHLRC1, NKX2-1, NKX6-2, NMNAT2, NOL3, NOP56, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NUBPL, NUS1, OFD1, OPA1, OPA3, OPHN1, PACS2, PANK2, PAX2, PAX6, PCLO, PDE6D, PDHX, PDSS1, PDSS2, PDYN, PEX10, PEX16, PEX2, PEX3, PEX6, PEX7, PGAP2, PGAP3, PGM1, PGM3, PHGDH, PHYH, PI4KA, PIBF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PIK3R5, PITRM1, PLA2G6, PLP1, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, POC1B, POLG, POLG2, POLR3A, POLR3B, POLR3K, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU4F1, PPP2R2B, PRDX3, PRICKLE1, PRKCG, PRNP, PRRT2, PTF1A, PTRH2, PUM1, RABL2A, RARS2, RELN, RFC1, RFT1, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RRM2B, RUBCN, SACS, SAMD9L, SAR1B, SCLT1, SCN1A, SCN2A, SCN8A, SCN9A, SCYL1, SEC23B, SEPSECS, SERAC1, SETX, SH3TC2,

SIL1, SLC16A2, SLC17A5, SLC1A3, SLC20A2, SLC25A15, SLC25A32, SLC25A46, SLC2A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A8, SLC44A1, SLC52A2, SLC52A3, SLC6A5, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPAST, SPG11, SPG20, SPG7, SPR, SPTBN2, SQSTM1, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, STUB1, STXBP1, SUFU, SUOX, SVBP, SYNE1, SYNGAP1, SYT14, TAPT1, TBC1D23, TBC1D32, TBP, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TERT, TGM6, THG1L, TINF2, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM5, TMEM67, TOE1, TPP1, TRAPPC11, TRIM32, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTC8, TTPA, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUSC3, TWNK, TXNDC15, TYMP, UBA5, UBR4, UBTF, UCHL1, VAMP1, VLDLR, VPS13B, VPS13D, VPS41, VPS53, VRK1, VWA3B, WASHC5, WDPCP, WDR63, WDR73, WDR81, WFS1, WWOX, XRCC1, XYLT1, XYLT2, ZFYVE26, ZNF423, ZNF592, ZSWIM6.

Forrás:

Hereditary ataxia and cerebellar anomalies - childhood onset (Version 6.388)
<https://panelapp.genomicsengland.co.uk/panels/488/> (Megtekintve: 2022.02.09.)

Hereditary ataxia - adult onset (Version 2.139)
<https://panelapp.genomicsengland.co.uk/panels/466/> (Megtekintve: 2022.02.09.)

Hereditary ataxia (Version 1.282)
<https://panelapp.genomicsengland.co.uk/panels/20/> (Megtekintve: 2022.02.09.)

Ataxia and cerebellar anomalies - narrow panel (Version 2.281)
<https://panelapp.genomicsengland.co.uk/panels/477/> (Megtekintve: 2022.02.09.)

Ataxia NGS Panel
<https://www.fulgentgenetics.com/Ataxia> (Megtekintve: 2022.02.09.)

Ataxia Panel
<https://blueprintgenetics.com/tests/panels/neurology/ataxia-panel/> (Megtekintve: 2022.02.09.)

Cerebellar Ataxia
<https://www.centogene.com/science/centopedia/ngs-panel-genetic-testing-for-cerebellar-ataxia.html> (Megtekintve: 2022.02.09.)