

## Ataxia génpanel v: 2025-05-11.

Panel tartalma 537 gén:

AAAS, AARS1, AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ACBD6, ACO2, ADCY5, ADGRG1, ADPRS, AFG3L2, AGTPBP1, AHI1, ALDH5A1, ALG1, ALG11, ALG12, ALG14, ALG3, ALG6, ALG8, ALG9, ALS2, AMPD2, ANO10, AP1S2, APTX, ARL13B, ARL3, ARL6, ARMC9, ARSA, ASAH1, ASL, ATAD3A, ATCAY, ATG7, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6AP1, ATP6V0A1, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, BRF1, BSCL2, C12orf4, C19orf12, C2CD3, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAD, CAMTA1, CAPN1, CAPRIN1, CASK, CBY1, CC2D2A, CCDC115, CCDC88C, CENPF, CEP104, CEP290, CEP41, CHCHD10, CHMP1A, CHP1, CHST14, CHST3, CHST6, CHSY1, CILK1, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, COA7, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COQ2, COQ4, COQ6, COQ8A, COQ9, COX20, CP, CPLANE1, CRB2, CRPPA, CTBP1, CTDPI, CTSA, CWF19L1, CYP27A1, CYP2U1, CYP7B1, CSGALNACT1, CSPP1, CSTB, DAG1, DAGLA, DARS2, DCC, DDHD2, DDOST, DDX59, DHCR7, DHDDS, DHPS, DHRSX, DKC1, DLG4, DNAJC19, DNAJC3, DNAJC5, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYSL5, DYNC1H1, EBF3, EDEM3, EEF2, EEFSEC, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EN1, EOGT, EPM2A, ERCC4, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAM149B1, FAT2, FBXL4, FCSK, FDXR, FEM1C, FGF14, FKRP, FKTN, FLVCR1, FMR1, FOLR1, FRMD5, FTH1, FUT8, FXN, G6PC3, GALC, GALNT2, GALNT3, GBA2, GDAP2, GEMIN5, GFAP, GFPT1, GJC2, GLI3, GLRA1, GLRB, GLS, GMPPA, GMPPB, GNE, GORAB, GOSR2, GPAA1, GRID2, GRM1, GRN, GSS, HARS1, HARS2, HEATR5B, HEPACAM, HEXA, HEXB, HIBCH, HMBS, HTT, HYLS1, IFT74, INPP5E, INTS11, IRF2BPL, ITM2B, ITPR1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNN2, KCNQ2, KCNQ3, KIAA0586, KIAA0753, KIF1A, KIF1C, KIF5A, KIF7, LAMA1, LARGE1, LARS2, LETM1, LFNG, LIG3, LMNB1, LNPBK, LRPPRC, LRSAM1, LSM7, MAG, MAGT1, MAN1B1, MAN2B2, MAPK8IP3, MARS2, MECP, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MKKS, MKS1, MLC1, MMACHC, MME, MOGS, MORC2, MPDU1, MPI, MRE11, MSTO1, MTCL1, MTFMT, MTPAP, MTRFR, MTPP, MVK, NAA60, NAXE, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEU1, NFASC, NGLY1, NHLRC1, NKX2-1, NKX6-2, NOL3, NOP56, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NPTX1, NUBPL, NUS1, OFD1, OGDHL, OPA1, OPA3, OPHN1, PACS2, PANK2, PAX6, 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, PITRM1, PLA2G6, PLP1, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, PNPT1, POC1B, POLG, POLR3A, POLR3B, POLR3K, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU4F1, PPP2R2B, PRDM13, PRDX3, PRKCG, PRNP, PRPS1, PRRT2, PTF1A, PTPMT1, PTRH2, PUM1, RAB3A, RARS2, RELN, RFT1, RFXANK, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RRM2B, RUBCN, RXYLT1, SACS, SAMD9L, SAR1B, SCLT1, SCN1A, SCN2A, SCN8A, SCYL1, SEC23B, SEPSECS, SERAC1, SETX, SH3TC2, SIL1, SLC16A2,

SLC17A5, SLC1A3, SLC20A2, SLC25A15, SLC25A46, SLC2A1, SLC35A1, SLC35A2, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC44A1, SLC52A2, SLC52A3, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPART, SPAST, SPG11, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STUB1, STXBP1, SUFU, SUOX, SVBP, SYNE1, SYNGAP1, SYT14, TANGO2, TBC1D23, TBP, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TERT, TGM6, THG1L, TINF2, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM199, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM67, TOE1, TOGARAM1, TPP1, TRAPPC11, TRIM32, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTC8, TTPA, TUBA1A, TUBA4A, TUBA8, TUBB2B, TUBB3, TUBB4A, TUSC3, TWNK, TXNDC15, TYMP, UBA5, UBTF, UCHL1, VAMP1, VLDLR, VPS13B, VPS13D, VPS41, VPS53, VRK1, VWA3B, WASHC5, WDPCP, WDR73, WDR81, WFS1, WWOX, XRCC1, XYLT1, XYLT2, ZFH3, ZFYVE26, ZNF423, ZSWIM6

Forrás:

**Hereditary ataxia and cerebellar anomalies - childhood onset (Version 22.7)**

<https://panelapp.genomicsengland.co.uk/panels/488/>

**Hereditary ataxia with onset in adulthood (Version 8.5)**

<https://panelapp.genomicsengland.co.uk/panels/466/>

**Hereditary ataxia (Version 1.342)**

<https://panelapp.genomicsengland.co.uk/panels/20/>

**Ataxia and cerebellar anomalies - narrow panel (Version 8.3)**

<https://panelapp.genomicsengland.co.uk/panels/477/>

Ataxia NGS Panel

<https://www.fulgentgenetics.com/Ataxia>

Ataxia Panel

<https://blueprintgenetics.com/tests/panels/neurology/ataxia-panel/>

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