

## Neurodegeneratív betegségek (Neurodegenerative disorders) génpanel v:2025-05-22

Panel tartalma 440 gén:

AARS1, AARS2, ABCB7, ABCD1, ABHD12, ACOX1, ADAR, ADCY5, ADPRS, AFG3L2, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALS2, ANG, ANO10, ANO3, ANXA11, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APP, APTX, AR, ARHGEF28, ARL6IP1, ARSA, ASPA, ATCAY, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6AP2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B4GALNT1, BCAP31, BOLA3, BSCL2, C19orf12, C9orf72, CA2, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CCDC88C, CCNF, CHCHD10, CHCHD2, CHMP2B, CIZ1, CLCN2, CLN3, CLN5, CLN6, CLN8, COA7, COA8, COASY, COL4A1, COL4A2, COL6A3, COLGALT1, COQ2, COQ8A, COX10, COX15, COX20, COX6B1, CP, CPT1C, CTBP1, CTC1, CTSA, CTSD, CTSF, CWF19L1, CYP27A1, CYP2U1, CYP7B1, CSF1R, CST3, CSTB, DARS1, DARS2, DCAF17, DCTN1, DDHD1, DDHD2, DNAJB2, DNAJC12, DNAJC13, DNAJC5, DNAJC6, DNAJC7, DNMT1, DSTYK, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL4, ELOVL5, EMC1, EPM2A, EPRS1, ERBB4, ERCC6, ERCC8, ERLIN2, EWSR1, FA2H, FAM126A, FARS2, FBXO7, FGF14, FIG4, FKR, FKTN, FLVCR1, FOLR1, FOXC1, FOXRED1, FRRS1L, FTL, FUCA1, FUS, FXN, GALC, GAN, GBA, GBA2, GBE1, GCDH, GCH1, GEMIN4, GFAP, GFM1, GIGYF2, GJC2, GLA, GLB1, GLRX5, GLT8D1, GMPPB, GNAL, GNAO1, GOSR2, GRID2, GRM1, GRN, GSN, HEPACAM, HEXA, HEXB, HIKESHI, HNRNPA1, HNRNPA2B1, HPCA, HSD17B4, HSPD1, HTRA1, HTT, IBA57, IDS, IFIH1, IRF2BPL, ISCA1, ISCA2, ITM2B, ITPR1, JPH3, KARS1, KCNA1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNT1, KCTD7, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2B, L1CAM, L2HGDH, LAMA2, LAMB1, LARGE1, LIPT2, LMNB1, LRPPRC, LRRK2, LYRM7, LYST, MAG, MAPT, MARS2, MATR3, MCOLN1, MECR, MFSD8, MLC1, MPV17, MRE11, MTFMT, MTHFS, MTRFR, MTPP, MYORG, NAA60, NACC1, NARS2, NAXE, NDUFS1, NDUFV1, NEK1, NEU1, NFU1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NOP56, NOTCH3, NPC1, NPC2, NR4A2, NT5C2, NUBPL, OCLN, OPTN, PANK2, PARK7, PC, PDE10A, PDGFB, PDGFRB, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PHGDH, PHYH, PINK1, PLA2G6, PLAA, PLEKHG2, PLP1, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMT1, POMT2, PPP2R2B, PPT1, PRICKLE1, PRKCG, PRKN, PRKRA, PRNP, PRPH, PRRT2, PSAP, PSAT1, PSEN1, PSEN2, PUM1, PYCR2, QARS1, RAB11B, RAB32, RARS1, RARS2, REEP1, REEP2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RTN2, SACS, SAMHD1, SCN1A, SCN2A, SCN8A, SCP2, SCYL1, SDHA, SDHAF1, SETX, SGCE, SIGMAR1, SIL1, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC25A12, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC6A3, SNCA, SNCB, SNORD118, SNX14, SOD1, SORL1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SS18L1, STN1, STUB1, SUMF1, SURF1, SYNE1, SYNJ1, TACO1, TAF1, TARDBP, TARS2, TBCD, TBCK, TBK1, TBP, TDP1, TDP2, TECPR2, TFG, TGFB1, TGM6, TH, THAP1, TMEM106B, TMEM240, TOR1A, TPP1, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TTBK2, TTC19, TTPA, TTR, TUBA4A, TUBB4A, TUFM, TWNK, TYMP, TYROBP, UBAP1, UBQLN2,

UBTF, UCHL1, UFC1, UFM1, UQCRC1, VAC14, VAMP1, VAPB, VARS1, VARS2, VCP, VLDLR, VPS11, VPS13A, VPS13C, VPS13D, VPS35, VRK1, WARS2, WASHC5, WDR45, WDR45B, WDR81, WFS1, WWOX, XK, XPR1, XRCC1, ZFYVE26, ZNHIT3

Forrás:

Adult onset neurodegenerative disorder (Version 8.1)

<https://panelapp.genomicsengland.co.uk/panels/474/>

Neurodegenerative Diseases Panel

<https://cegat.com/diagnostics/rare-diseases/neurodegenerative-diseases/>

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