

**Neurodegeneratív betegségek (Neurodegenerative disorders) génpanel**  
**v:2023.10.12.**

Panel tartalma 557 gén:

AP4E1, AP4S1, SIL1, TTC19, AAAS, AARS, AARS1, AARS2, ABCB7, ABCD1, ABHD12, ACOX1, ACTB, ADAR, ADCY5, ADPRHL2, AFG3L2, AIMP1, AIMP2, ALAS2, ALDH18A1, ALDH3A2, ALS2, AMPD2, ANG, ANO10, ANO3, ANXA11, AP1S2, AP4B, AP4B1, AP4M, AP4M1, AP5Z1, APOE, APOPT1, APP, APTX, AR, ARG1, ARHGEF28, ARL6IP1, ARSA, ARSI, ARX, ASPA, ATCAY, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP6AP2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8, AUH, B4GALNT1, BCAP31, BEAN1, BOLA3, BSCL2, C12orf65, C19orf12, C9orf72, CA2, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CARS2, CASK, CCDC88C, CCNF, CCT5, CDK16, CHCHD10, CHCHD2, CHMP1A, CHMP2B, CIZ1, CLCN2, CLN3, CLN5, CLN6, CLN8, CLP1, COA7, COASY, COG5, COL4A1, COL4A2, COL6A3, COLGALT1, COQ2, COQ8A, COX10, COX15, COX20, COX6B1, CP, CPT1C, CTBP1, CTC1, CTSA, CTSD, CTSF, CWF19L1, CYP27A1, CYP2U1, CYP7B1, CSF1R, CST3, CSTB, DAB1, DAO, DARS, DARS1, DARS2, DCAF17, DCTN1, DDC, DDHD1, DDHD2, DLAT, DMXL2, DNAJB2, DNAJC12, DNAJC13, DNAJC19, DNAJC5, DNAJC6, DNAJC7, DNMT1, DRD2, DRD5, DSTYK, DYNC1H1, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL4, ELOVL5, EMC1, ENTPD1, EPM2A, EPRS1, ERBB4, ERCC6, ERCC8, ERLIN1, ERLIN2, EWSR1, EXOSC3, FA2H, FAM126A, FARS2, FASTKD2, FBXO7, FGF14, FIG4, FKRP, FKTN, FLVCR1, FMR1, FOLR1, FOXC1, FOXG1, FOXRED1, FRRS1L, FTL, FUCA1, FUS, FXN, GAD1, GALC, GAMT, GAN, GBA, GBA2, GBE1, GCDH, GCH1, GEMIN4, GFAP, GFM1, GIGYF2, GJC2, GLA, GLB1, GLRA1, GLRB, GLRX5, GLT8D1, GMPPB, GNAL, GNAO1, GOSR2, GPAA1, GRID2, GRM1, GRN, GSN, HACE1, HEPACAM, HEXA, HEXB, HFE, HIKESHI, HNRNPA1, HNRNPA2B1, HPCA, HPRT1, HSD17B4, HSPD1, HTRA1, HTRA2, HTT, IBA57, IDS, IFIH1, IPPK, IRF2BPL, ISCA1, ISCA2, ITM2B, ITPR1, JPH3, KARS1, KCNA1, KCNC3, KCND3, KCNJ10, KCNK18, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIAA1161, KIDINS220, KIF1A, KIF1C, KIF5A, KLC4, KMT2B, L1CAM, L2HGDH, LAMA2, LAMB1, LARGE1, LIPT2, LMNB1, LRPPRC, LRRK2, LYRM7, LYST, MAG, MAPT, MARS, MARS2, MAT1A, MATR3, MCOLN1, MECR, MFSD8, MLC1, MMACHC, MMADHC, MPV17, MR1, MRE11, MT-ATP6, MT-ND6, MTFMT, MTHFS, MTPAP, MTPP, MVK, MYORG, NACC1, NAGLU, NARS2, NAXE, NDUFA12, NDUFS1, NDUFV1, NEFH, NEK1, NEU1, NFU1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NOP56, NOTCH3, NPC1, NPC2, NR4A2, NT5C2, NUBPL, OCLN, OPA3, OPHN1, OPTN, PANK2, PARK7, PAX2, PAX6, PC, PCDH12, PCLO, PDE10A, PDGFB, PDGFRB, PDHX, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PGAP1, PHGDH, PHYH, PIK3R5, PINK1, PLA2G6, PLAA, PLEKHG2, PLP1, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, PNPT1, POLG, POLR1C, POLR3A, POLR3B, POLR3K, POMGNT1, POMT1, POMT2, PPP2R2B, PPT1, PRICKLE1, PRKCG, PRKN, PRKRA, PRNP, PRPH, PRRT2, PSAP, PSAT1, PSEN1, PSEN2, PTEN, PTS, PUM1, PYCR2, QARS1, QDPR, RAB11B, RAB39B, RAB3GAP2, RARS1, RARS2,

REEP1, REEP2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RTN2, RUBCN, SACS, SAMHD1, SAR1B, SCN1A, SCN2A, SCN8A, SCN9A, SCP2, SCYL1, SDHA, SDHAF1, SEPSECS, SERAC1, SETX, SGCE, SIGMAR1, SLC16A2, SLC17A5, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC25A12, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC41A1, SLC46A1, SLC52A1, SLC52A2, SLC52A3, SLC6A3, SLC6A5, SLC9A6, SNCA, SNCAIP, SNCB, SNORD118, SNX14, SOD1, SORL1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SQSTM1, SRD5A3, SS18L1, STN1, STUB1, SUCLA2, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, SYT14, TACO1, TAF1, TAF15, TARDBP, TARS2, TBCD, TBCK, TBK1, TBP, TDP1, TDP2, TECPR2, TET2, TFG, TGFB1, TGM6, TH, THAP1, TIA1, TIMM8A, TMEM106B, TMEM240, TOR1A, TPK1, TPP1, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TSEN2, TSEN34, TSEN54, TTBK2, TTPA, TTR, TUBA4A, TUBB4A, TUFM, TWNK, TYMP, TYROBP, UBAP1, UBQLN2, UBR4, UBTF, UCHL1, UFC1, UFM1, UNC13A, UQCRC1, USP8, VAC14, VAMP1, VAPB, VARS1, VARS2, VCP, VEGFA, VLDLR, VPS11, VPS13A, VPS13C, VPS13D, VPS35, VPS37A, VPS53, VRK1, WARS2, WASHC5, WDR45, WDR45B, WDR48, WDR73, WDR81, WFS1, WWOX, XK, XPR1, XRCC1, YY1, ZEB2, ZFYVE26, ZFYVE27, ZNF592, ZNHIT3

Forrás:

Adult onset neurodegenerative disorder (Version 4.22)

<https://panelapp.genomicsengland.co.uk/panels/474/> (Megtekintve: 2023.07.19.)

Neurodegenerative Diseases

<https://cegat.com/diagnostics/rare-diseases/neurodegenerative-diseases/> (Megtekintve: 2023.07.19.)