Genpanel NMD-Ataxi-HSP (v.72)
956 gener (inkl. 29 repeatexpansioner)

Genpanel NMD-Ataxi-HSP

AAAS, AARS1, AARS2, ABAT, ABCA1, ABCB7, ABCD1, ABHD12, ABHD16A, ACAD9, ACADM, ACADVL, ACBD5, ACOX1, ACP5, ACTA1, ACTB, ACTN2, ADAMTS10, ADAR, ADCY5, ADCY6, ADGRG1, ADGRG6, ADSS1, AFG3L2, AGL, AGRN, AGTPBP1, AHI1, AIFM1, AIMP1, ALDH18A1, ALDH3A2, ALDH5A1, ALDOA, ALG1, ALG11, ALG12, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALS2, AMPD2, ANG, ANO10, ANO3, ANO5, ANTXR2, ANXA11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APP, APTX, AR, ARG1, ARL13B, ARL6IP1, ARMC9, ARSA, ASAH1, ASCC1, ASPA, ASXL1, ATAD3A, ATCAY, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D2, BAG3, BCAP31, BCAS3, BCKDHB, BCS1L, BEAN1, BICD2, BIN1, BLTP1, BOLA3, BRF1, BSCL2, C19orf12, C2CD3, C9orf72, CA8, CACNA1A, CACNA1E, CACNA1G, CACNA1S, CACNA2D2, CACNB4, CAMK4, CAMTA1, CAPN1, CAPN3, CASK, CAV3, CC2D2A, CCDC115, CCDC78, CCNF, CD59, CENPF, CEP104, CEP290, CEP41, CFAP276, CFL2, CHAT, CHCHD10, CHCHD2, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1, CIC, CILK1, CLCN1, CLCN2, CLN3, CLN5, CLN6, CLP1, CLPB, CNBP, CNTNAP1, COA7, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL12A1, COL13A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COLGALT1, COLQ, COQ2, COQ8A, COX10, COX15, COX20, COX6A1, CP, CPLANE1, CPOX, CPT1C, CPT2, CRB2, CRLF1, CRPPA, CRYAB, CSF1R, CSGALNACT1, CSPP1, CSTB, CTBP1, CTC1, CTDP1, CTSA, CTSF, CWF19L1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAB1, DAG1, DARS1, DARS2, DBH, DCAF17, DCTN1, DDB2, DDC, DDHD1, DDHD2, DDX59, DEGS1, DES, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DKC1, DLAT, DLD, DMD, DMPK, DNAJB2, DNAJB6, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DST, DYNC1H1, DYSF, EARS2, EBP, ECEL1, ECHS1, EGR2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ELP1, EMD, ENO3, ENTPD1, EPG5, EPM2A, EPRS1, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERGIC1, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAH, FAM20C, FAR1, FARS2, FARSA, FARSB, FBLN5, FBN2, FBXO7, FGD4, FGF14, FGFR2, FGFR3, FHL1, FIG4, FITM2, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLNC, FLVCR1, FLVCR2, FMR1, FOLR1, FOXG1, FOXP2, FOXRED1, FTL, FUCA1, FUS, FUT8, FXN, FXR1, GAA, GALC, GALNT2, GALNT3, GAN, GARS1, GBA1, GBA2, GBE1, GBF1, GCDH, GCH1, GDAP1, GFAP, GFM1, GFPT1, GIPC1, GJA1, GJB1, GJC2, GLA, GLB1, GLDN, GLE1, GLI3, GLRA1, GLRB, GLRX5, GLS, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNB1, GNB4, GNE, GOSR2, GPAA1, GPT2, GRID2, GRIN1, GRM1, GRN, GSN, GTF2H5, GTPBP2, GYG1, GYS1, HACE1, HADHA, HADHB, HARS1, HCFC1, HEPACAM, HEXA, HEXB, HIBCH, HIKESHI, HINT1, HK1, HMBS, HMGCL, HNRNPA1, HNRNPDL, HNRNPH1, HPCA, HPDL, HPRT1, HSD17B4, HSPB1, HSPB8, HSPD1, HSPG2, HTRA1, HTRA2, HTT, HYCC1, HYLS1, IARS2, IBA57, IFIH1, IGHMBP2, IMPDH2, INF2, INPP5E, INPP5K, IRF2BPL, IRF6, ISCA2, ISCU, ISG15, ITGA7, ITM2B, ITPR1, IVD, JAM3, JPH3, KAT6B, KBTBD13, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNJ2, KCNMA1, KCNQ2, KCTD17, KDM5C, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KIF5A, KIF7, KLHL40, KLHL41, KLHL7, KMT2B, L1CAM, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS2, LDB3, LDHA, LGI4, LIG3, LITAF, LMNA, LMNB1, LMOD3, LMX1B, LPIN1, LRP12, LRP4, LRPPRC, LRRK2, LRSAM1, LYRM7, LYST, MACF1, MAGEL2, MAN1B1, MAOA, MAP3K20, MAPK8IP3, MAPT, MARS1, MARS2, MATR3, MCM3AP, MCOLN1, MECR, MED12, MED27, MEF2C, MEGF10, MFN2, MGAT2, MICU1, MKS1, MLC1, MMACHC, MME, MMUT, MOGS, MORC2, MPDU1, MPI, MPLKIP, MPV17, MPZ, MRE11, MSTO1, MT-ATP6, MT-ATP8, MT-ND1, MT-ND6, MT-RNR1, MT-TL1, MTFMT, MTHFR, MTM1, MTMR2, MTRFR, MTTP, MUSK, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYL1, MYMK, MYO18B, MYO9A, MYORG, MYOT, MYPN, NAGA, NALCN, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEB, NEFH, NEFL, NEK9, NGF, NGLY1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NOP56, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NRROS, NT5C2, NTRK1, NUBPL, NUP188, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPTN, ORAI1, PABPN1, PACS2, PAFAH1B1, PAH, PANK2, PARK7, PAX6, PAX7, PCCA, PCCB, PCYT2, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PDHX, PDP1, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6,

PEX7, PFKM, PFN1, PGAM2, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHKA1, PHKB, PHOX2B, PHYH, PI4KA, PIBF1, PIEZO2, PIGA, PIGB, PIGL, PIGN, PIGO, PIGS, PIGT, PIGV, PINK1, PLA2G6, PLEC, PLEKHG5, PLOD1, PLOD2, PLP1, PMM2, PMP2, PMP22, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, POLG, POLG2, POLH, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PPOX, PPP2R2B, PPP2R5D, PRDM12, PRDM13, PRG4, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRPS1, PRRT2, PRX, PSAP, PSEN1, PSEN2, PTEN, PTF1A, PTPN11, PTRH2, PTRHD1, PTS, PUM1, PYCR2, PYGM, PYROXD1, QDPR, RAB11B, RAB39B, RAB7A, RAPSN, RARS1, RARS2, RBCK1, REEP1, REEP2, RELN, RETREG1, RFC1, RFT1, RIPK4, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RNU7-1, ROBO3, RPGRIP1L, RPS6KA3, RRM2B, RTN2, RXYLT1, RYR1, RYR3, SACS, SAMD9L, SAMHD1, SAR1B, SBF1, SBF2, SCAF4, SCARF2, SCLT1, SCN10A, SCN11A, SCN1A, SCN4A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SEC23B, SELENON, SEPSECS, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIGMAR1, SIL1, SKI, SLC12A6, SLC16A2, SLC17A5, SLC18A2, SLC18A3, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A4, SLC25A46, SLC29A3, SLC2A1, SLC30A10, SLC35A1, SLC35C1, SLC35D1, SLC39A14, SLC39A8, SLC52A2, SLC52A3, SLC5A7, SLC6A3, SLC6A8, SLC6A9, SLC9A6, SMAD3, SMAD4, SMN1, SMPD4, SNCA, SNORD118, SNX14, SOD1, SORD, SOX10, SPART, SPAST, SPATA5L1, SPEG, SPG11, SPG21, SPG7, SPR, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAC3, STIM1, STN1, STT3A, STUB1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNGAP1, SYNJ1, SYT2, TACO1, TAF1, TANGO2, TARDBP, TARS2, TBK1, TBP, TCAP, TCTN1, TCTN2, TCTN3, TERT, TFG, TGFB2, TGFB3, TGFBR1, TGFBR2, TGM6, TH, THAP1, TIA1, TIMM8A, TINF2, TK2, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM67, TNNI2, TNNT1, TNNT3, TNPO3, TOE1, TOR1A, TOR1AIP1, TPK1, TPM2, TPM3, TPP1, TRAPPC11, TREM2, TREX1, TRIM2, TRIM32, TRIP4, TRPA1, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTN, TTPA, TTR, TUBA1A, TUBB2B, TUBB3, TUBB4A, TUSC3, TWNK, TXNDC15, TYMP, TYROBP, UBA1, UBA5, UBAP1, UBQLN2, UBTF, UCHL1, USP18, VAC14, VAMP1, VAMP2, VAPB, VCP, VIPAS39, VLDLR, VMA21, VPS11, VPS13A, VPS13B, VPS13D, VPS16, VPS33B, VPS35, VPS41, VPS4A, VPS53, VRK1, VWA1, WARS1, WASHC5, WDR45, WDR45B, WDR73, WDR81, WFS1, WNK1, WWOX, XK, XPA, XPC, XPR1, XRCC1, XYLT1, XYLT2, YARS1, YIF1B, YY1, ZC4H2, ZFYVE26, ZMPSTE24, ZSWIM6

Varav 29 repeatexpansioner (STR)

AR, ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, C9ORF72, CACNA1A, CNBP, CSTB, DMPK, FMR1, FXN, GIPC1, GLS, HTT, JPH3, LRP12, NIPA1, NOP56, PABPN1, PHOX2B, PPP2R2B, RFC1, TBP, XYLT1