

## Genpanel avseende Intellectuell Funktionsnedsättning (IF)

### Version 9.0, 1314 gener

AAAS, AARS, AASS, ABAT, ABCA2, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ACAD9, ACADM, ACADS, ACER3, ACO2, ACOX1, ACSL4, ACTB, ACTG1, ACTL6A, ACTL6B, ACY1, ADAM22, ADAR, ADARB1, ADAT3, ADD1, ADD3, ADGRG1, ADK, ADNP, ADSL, AFF2, AFF3, AFF4, AGA, AGO1, AGO2, AHCY, AHDC1, AH11, AIFM1, AIMP1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG12, ALG13, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, AMER1, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANKRD17, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APOPT1, ARCN1, ARF1, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSE, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATG7, ATIC, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATP9A, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT7, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCS1L, BICRA, BLM, BLOC1S1, BMP4, BOLA3, BPTF, BRAF, BRAT1, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BTD, BUB1B, C12orf4, C12orf57, C12orf65, C2CD3, C5orf42, CA2, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D1, CAD, CAMK2A, CAMK2B, CAMK4, CAMTA1, CAPN15, CARS, CASK, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC22, CCDC32, CCDC47, CCDC88C, CCND2, CDC42, CDC6, CDH11, CDH2, CDK10, CDK13, CDK19, CDK5RAP2, CDK8, CDKL5, CDON, CELF2, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP55, CEP57, CEP83, CEP85L, CHAMP1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CIC, CIT, CKAP2L, CLCN3, CLCN4, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLTC, CNBP, CNKSR2, CNNM2, CNOT1, CNOT2, CNOT3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COL4A3BP, COLEC11, COPB2, COQ4, COQ8A, COX10, COX15, CPE, CPS1, CRADD, CRB2, CREBBP, CSDE1, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CSTB, CTBP1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTSA, CTSD, CTU2, CUL3, CUL4B, CUX1, CUX2, CWC27, CWF19L1, CXorf56, CYB5R3, CYC1, CYFIP2, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCHS1, DCPS, DCX, DDB1, DDC, DDHD2, DDX11, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHTKD1, DHX30, DHX37, DIAPH1, DIS3L2, DKC1, DLD, DLG3, DLG4, DLL1, DMD, DMPK, DMXL2, DNAJC12, DNAJC19, DNMI, DNMI1, DNMT3A, DNMT3B, DOCK3, DOCK6, DOCK7, DOLK, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPP6, DPYD, DPYS, DPYSL5, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, EBF3, EBP, EDEM3, EED, EEF1A2, EFTUD2, EHMT1, EIF2AK2, EIF2AK3, EIF2S3, EIF3F, EIF4A3, EIF5A, ELAC2, ELOVL4, ELP2, EMC1, EMC10, EML1, EMX2, EP300, EPG5, ERBB4, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC6L2, ERCC8, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXT2, EXTL3, EZH2, FAM111A, FAM126A, FAM20C, FAM50A, FAR1, FARS2, FARSA, FAT4, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXO31, FBXW11, FBXW7, FGD1, FGF12, FH, FIG4, FKRP, FKTN, FLNA, FLVCR2, FMN2, FMR1, FMR1, FOLR1, FOXP1, FOXP2, FOXRED1, FRMPD4, FTCD, FTSJ1, FUCA1, FUT8, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALE, GALNT2, GALT, GAMT, GATAD2B, GATM, GCDH, GCH1, GDII, GEMIN5, GFAP, GFER, GFM1, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLIS3, GLRA2, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAI1, GNAO1, GNAS, GNB1, GNB2, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GPAAI, GPC3, GPC4, GPT2, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM1, GTF2E2, GTF2H5, GTPBP2, GTPBP3, GUSB, H3F3A, H3F3B, HACE1, HADHA, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIST1H1E, HIST1H4C, HIVEP2, HK1,

HLCS, HMGB1, HMGCL, HNMT, HNRNPH1, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPD, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA2, HUWE1, IARS, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IKBKG, IL1RAPL1, IMPDH2, INPP5E, INPP5K, INTS1, IQSEC2, IRF2BPL, IRX5, ISPD, ITPA, ITPR1, IVD, JAM3, JARID2, KANSL1, KARS, KAT5, KAT6A, KAT6B, KAT8, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNJ10, KCNJ11, KCNJ6, KCNK9, KCNMA1, KCNN2, KCNN3, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM1A, KDM3B, KDM4B, KDM5B, KDM5C, KDM6A, KDM6B, KIAA0586, KIAA1109, KIDINS220, KIF11, KIF14, KIF1A, KIF1BP, KIF21B, KIF2A, KIF5A, KIF5C, KIF7, KLF7, KLHL7, KMT2A, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, LICAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARP7, LARS, LIAS, LIG4, LINGO4, LINS1, LIPT1, LMBRD2, LMNB1, LONP1, LRP2, LRPPRC, LSS, LYRM7, LZTR1, MAB21L1, MAB21L2, MACF1, MADD, MAF, MAGEL2, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAPK1, MAPK8IP3, MAPRE2, MASP1, MAST1, MAT1A, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCM3AP, MCOLN1, MCPH1, MDH2, MECP2, MED12, MED13, MED13L, MED17, MED23, MED25, MED27, MEF2C, MEIS2, METTL23, METTL5, MFF, MFSD2A, MFSD8, MGAT2, MICU1, MID1, MINPP1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MNI, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPLKIP, MPP5, MRPS22, MRPS34, MSL3, MSMO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, MVK, MYCN, MYO5A, MYT1L, NAA10, NAA15, NACCI, NAGA, NAGLU, NALCN, NANS, NAPB, NARS, NBEA, NCDN, NCKAPI, NDE1, NDP, NDST1, NDUFA1, NDUFA2, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEDD4L, NEMF, NEU1, NEUROD2, NEXMIF, NF1, NFASC, NFIA, NFIX, NFU1, NGLY1, NHS, NIPBL, NKAP, NKX2-1, NLGN3, NONO, NOVA2, NPC1, NPC2, NPHP1, NR2F1, NR4A2, NRAS, NRCAM, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NSUN2, NT5C2, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUS1, OCLN, OCRL, ODC1, OFD1, OGDHL, OGT, OPA3, OPHN1, OSGEP, OTC, OTUD5, OTUD6B, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PARN, PAX6, PAX8, PBX1, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCGF2, PCNT, PCYT2, PDE4D, PDGFRB, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDZD8, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM2L1, PGM3, PHACTR1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PI4KA, PIBF1, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PITRM1, PLA2G6, PLAA, PLCB1, PLK4, PLP1, PLPBP, PMM2, PMPCB, PNKP, PNPLA6, PNPT1, POGZ, POLA1, POLG, POLR1C, POLR2A, POLR3A, POLR3B, POLRMT, POMGNT1, POMGNT2, POMT1, POMT2, PORCN, POU3F3, PPIL1, PPM1D, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM13, PRICKLE2, PRMT7, PRPF8, PRPS1, PRR12, PRSS12, PRUNE1, PSAP, PSMD12, PSPH, PTCHI, PTCHD1, PTDSS1, PTEN, PTF1A, PTPN11, PTPN23, PTPN4, PTRHD1, PTS, PUF60, PUM1, PURA, PUS1, PUS3, PUS7, PYCRI, PYCR2, QARS, QDPR, QRICHI, RAB11A, RAB11B, RAB18, RAB23, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAC3, RAD21, RAF1, RAI1, RALA, RALGAP1, RARB, RARS, RARS2, RBBP8, RBM10, RELN, RERE, RFT1, RFX3, RFX4, RFX7, RHOTB2, RIT1, RLIM, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNU7-1, ROGDI, ROR2, RORA, RPGRIP1L, RPIA, RPL10, RPS6KA3, RRM2B, RSRC1, RTEL1, RTN4IP1, RTTN, SAMD9, SAMHD1, SARS2, SATB1, SATB2, SBF1, SC5D, SCAF4, SCAMP5, SCAPER, SCN1A, SCN2A, SCN3A, SCN8A, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGPL1, SGSH, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SIAH1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SLC12A2, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A1, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A22, SLC2A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC38A3, SLC39A14, SLC39A8, SLC46A1, SLC4A4, SLC5A6, SLC6A1, SLC6A17, SLC6A19, 3(3)SLC6A3, SLC6A8, SLC6A9, SLC9A6, SLX4, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3,

*SMG8, SMOC1, SMPD1, SMPD4, SMS,SNAP25, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SNX27, SON, SOS1, SOS2, SOX10, SOX11,SOX2, SOX4, SOX5, SOX6, SOX9, SPART, SPATA5, SPATA5L1, SPECC1L, SPEN, SPG11, SPOP,SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SRCAP, SRD5A3, SRRM2, SSR4,ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STIL, STRA6, STRADA, STT3A, STX1B, STXBP1,SUCLG1, SUMF1, SUOX, SUPT16H, SURF1, SUZ12, SVBP, SYN1, SYNCRIP, SYNGAP1, SYNJ1,SYP, SYT1, SZT2, TAF1, TAF6, TAF8, TANC2, TANGO2, TAOK1, TASP1, TAT, TAZ, TBC1D20,TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBLIXR1, TBR1, TCF20, TCF4, TCF7L2, TCN2,TCTN2, TCTN3, TDP2, TECPR2, TELO2, TENM3, TET3, TFE3, TGIF1, TH, THOC2, THOC6,THRA, THUMPD1, TIAM1, TIMM50, TLK2, TMC01, TMEM106B, TMEM165, TMEM216, TMEM222, TMEM237, TMEM240, TMEM5, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNPO2, TNRC6B, TOE1, TOR1A, TP73, TPP1, TPP2, TRAF7, TRAIP, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIO, TRIP12, TRIT1, TRMT1, TRMT10A, TRNT1, TRPM3, TRRAP, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSPAN7, TTC19, TTC37, TTC5, TTC8, TTI2, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, TUSC3, TWIST1, UBA5, UBE2A, UBE3A, UBE3B, UBE4A, UBR1, UBR7, UBTF, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPF3B, UROC1, USP7, USP9X, VAMP2, VARS, VARS2, VLDLR, VPS11, VPS13B, VPS41, VPS4A, VPS53, VRK1, WAC, WARS2, WASF1, WDFY3, WDPCP, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WNK3, WNT1, WWOX, XRCC4, XYLT1, YIF1B, YIPF5, YWHAG, YY1, ZBTB18, ZBTB20, ZBTB24, ZBTB7A, ZC4H2, ZDHHC9, ZEB2, ZFHX4, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYND11, ZNF142, ZNF335, ZNF462, ZNF526, ZNF699, ZNF711, ZSWIM6*

## **Expansionsanalys**

*AFF2, CNBP, DMPK, EIF4A3, FMR1, XYLT1, ZIC2*