

## Genpanel IEM (Inborn Errors of Metabolism), version 22 – 1099 gener

Gener kopplade till medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild

*AARSI, AARS2, AASS, ABAT, ABCB7, ABCC8, ABCD1, ABCD4, ABHD12, ABHD16A, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACER3, ACO2, ACOX1, ACSF3, ACY1, ADA, ADAR, ADCK2, ADCY5, ADK, ADPRS, ADSL, ADSSI, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGL, AGO1, AGPS, AGTPBP1, AGXT, AHCY, AIFM1, AIMP1, AIMP2, ALAD, ALAS2, ALDH18A1, ALDH1L2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH1, ALMS1, ALPL, ALS2, AMACR, AMN, AMPD1, AMPD2, AMT, ANO10, ANTXR2, APIS1, AP3B1, AP3D1, AP5Z1, APOO, APRT, ARF3, ARG1, ARL3, ARNT2, ARSA, ARSB, ARSK, ASAH1, ASL, ASNS, ASPA, ASS1, ASXL3, ATAD3A, ATAD3B, ATAD3C, ATIC, ATP11A, ATP13A2, ATP1A1, ATP1A3, ATP2B1, ATP2B2, ATP5F1A, ATP5F1B, ATP5F1D, ATP5F1E, ATP5MC3, ATP5MK, ATP5PO, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATP9A, ATPAF2, AUH, B3GALNT2, B4GALNT1, B4GALT1, B4GALT7, BBOX1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BICRA, BICRAL, BLOC1S3, BLOC1S6, BOLA3, BSCL2, BTD, C19orf12, C1QBP, C2orf69, CA5A, CACNA1A, CACNA1E, CACNA1S, CAD, CAMLG, CAMSAP1, CARS1, CARS2, CAT, CBLIF, CBS, CCDC115, CCDC174, CCDC88C, CD320, CEP89, CERS1, CHCHD10, CHKA, CHKB, CIAO1, CISD2, CLCN2, CLCN6, CLCN7, CLDN11, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CLPX, CLTC, CLTRN, CMPK2, CNKSR2, COA3, COA5, COA6, COA7, COA8, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COLGALT1, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ9, COX10, COX11, COX14, COX15, COX16, COX18, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6A2, COX6B1, COX7B, COX8A, CP, CPOX, CPS1, CPT1A, CPT2, CRAT, CRLS1, CRNKL1, CRPPA, CSF1R, CTCF, CTH, CTNS, CTSA, CTSD, CTSF, CTSK, CTU2, CUBN, CWF19L1, CYC1, CYP7B1, D2HGDH, DARS1, DARS2, DBH, DBT, DCAF17, DDC, DDOST, DEGS1, DGAT1, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHPS, DHTKD1, DIAPH1, DKC1, DLAT, DLD, DMGDH, DNA2, DNAJC12, DNAJC19, DNAJC3, DNAJC30, DNAJC5, DNM1L, DOHH, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DTNBP1, DTYMK, EARS2, EBF3, EBP, ECHS1, ECM1, ECSIT, EDEM3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELOVL4, ELOVL5, ENO3, EPG5, EPHX1, EPM2A, EPRS1, ERAL1, ERCC2, ERCC6, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC5, EXOSC8, EXT2, FA2H, FAH, FAM126A, FAM149B1, FAR1, FARS2, FARSA, FARSB, FASTKD2, FBP1, FBXL4, FBXO11, FBXW7, FCSK, FDFT1, FDX2, FDXR, FECH, FH, FIG4, FLAD1, FMO3, FOLR1, FOXI1, FOXRED1, FTCD, FTL, FUCA1, FUT8, FXN, G6PC, GAA, GABBR2, GABRG2, GALC, GALE, GALK1, GALM, GALNS, GALNT2, GALNT3, GALT, GAMT, GARS1, GATB, GATC, GATM, GBA, GBA2, GBE1, GBF1, GCDH, GCH1, GCK, GCLC, GCSH, GET3, GET4, GFAP, GFER, GFM1, GFM2, GGT1, GJC2, GK, GLA, GLB1, GLDC, GLRA1, GLRA2, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAO1, GNBI, GNE, GNMT,*

GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GOT2, GPHN, GPR161, GPRC5B, GRHPR, GRIN2B, GRN, GSS, GSX2, GTPBP3, GUSB, GYG1, GYG2, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAL, HARS1, HCFC1, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGD, HGSNAT, HIBCH, HIKESHI, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HPD, HPDL, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HSD17B10, HSD17B4, HSPD1, HSPE1, HTRA2, HYAL1, HYOU1, IARS1, IARS2, IBA57, IDH2, IDH3A, IDI1, IDS, IDUA, IER3IP1, IFIH1, INPP5K, INSR, INTS11, IRF2BPL, ISCA1, ISCA2, ISCU, ITPA, ITPR1, IVD, JAG1, JAM2, KARS1, KCNA4, KCNJ11, KCNK3, KGD4, KIF5A, KYNU, L2HGDH, LAMA1, LAMP2, LARS1, LARS2, LBR, LDHA, LETM1, LHX4, LIAS, LIG3, LIPA, LIPE, LIPT1, LIPT2, LMBRD1, LMBRD2, LNPK, LONP1, LPIN1, LRP2, LRPPRC, LSM11, LYRM4, LYRM7, MADD, MAG, MAGED2, MAGEL2, MAGT1, MAL, MANIB1, MAN2B1, MAN2C1, MANBA, MAOA, MAPK8IP3, MARS1, MARS2, MAT1A, MBD5, MBOAT7, MCAT, MCCC1, MCCC2, MCEE, MCM7, MCOLN1, MDH1, MECP2, MECR, MED22, MED27, MFF, MFN2, MFSD8, MGAT2, MGME1, MICOS10, MICOS13, MICU1, MICU2, MIEF2, MIPEP, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMS19, MMUT, MOCOS, MOCS1, MOCS2, MOCS3, MOGS, MORC2, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL12, MRPL24, MRPL3, MRPL38, MRPL39, MRPL44, MRPL57, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS34, MRPS7, MSL2, MSL3, MSTO1, MTFMT, MTHFD1, MTHFR, MTO1, MTPAP, MTR, MTRFR, MTRR, MTSS2, MVK, MYH7, MYO5A, MYORG, NAA10, NADK2, NAE1, NAGA, NAGLU, NAGS, NALCN, NANS, NARS1, NARS2, NAXD, NAXE, NBAS, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB1, NDUFB10, NDUFB11, NDUFB3, NDUFB6, NDUFB7, NDUFB8, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NEU1, NFASC, NFE2L2, NFS1, NFUI, NGLY1, NKX6-2, NMNAT1, NOPI0, NOTCH1, NOTCH3, NPC1, NPC2, NR4A2, NRCAM, NRROS, NSDHL, NSRP1, NSUN3, NT5C3A, NUBPL, NUP188, NUP214, NUP62, NUS1, OAT, OCRL, OGDH, OGDHL, OPA1, OPA3, OPLAH, OSTC, OTC, OXAIL, OXCT1, OXR1, P4HTM, PAH, PAICS, PAK1, PANK2, PARK7, PARS2, PAX7, PAXBP1, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PCLO, PCYT1A, PDE10A, PDE12, PDE2A, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDXK, PEPD, PET100, PET117, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP2, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PI4KA, PIGA, PIGM, PIGN, PIGO, PIGQ, PIGS, PIGT, PIGV, PIGW, PIK3R4, PITRM1, PLA2G6, PLAA, PLP1, PMM2, PMPCA, PMPCB, PNP, PNPLA2, PNPLA4, PNPLA6, PNPLA8, PNPO, PNPT1, POLG, POLG2, POLR1A, POLRIC, POLR3A, POLR3B, POLRMT, POMGNT1, POMT1, PPA2, PPCDC, PPCS, PPFIBP1, PPMIK, PPOX, PPP1R15B, PPP1R21, PPP2R1A, PPP2R5D, PPT1, PRKAG2, PRKN, PRMT7, PRODH, PRODH2, PRORP, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTCD1, PTCD3, PTRH2, PTS, PURA, PUS1, PUS7, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, QRSL1, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RALGAP1, RANBP2, RARS1, RARS2, RBCK1, REPS1, RFT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF13, RNF213, RNF220, RNH1, RNU4-2, RNU7-1, RPIA, RRM1, RRM2B, RTN4IP1, RYR1, SAMHD1, SARS1, SARS2, SC5D, SCAF4, SCN8A, SCO1, SCO2, SCP2, SCYL1, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SELENOI, SEMA6B, SERAC1, SF3B1,

*SFXN4, SGSH, SHMT2, SHQ1, SIL1, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A1, SLC1A4, SLC20A2, SLC22A12, SLC22A5, SLC25A1, SLC25A10, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A32, SLC25A36, SLC25A4, SLC25A42, SLC25A46, SLC27A4, SLC2A1, SLC2A2, SLC2A9, SLC30A10, SLC30A5, SLC30A9, SLC31A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC38A3, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC44A1, SLC46A1, SLC4A10, SLC4A11, SLC52A1, SLC52A2, SLC52A3, SLC5A1, SLC5A2, SLC5A7, SLC6A17, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC7A2, SLC7A7, SLC7A9, SLC9A1, SLC9A7, SLIRP, SMARCD1, SMG9, SMPD1, SMPD4, SMS, SNAPC4, SNIP1, SNORD118, SNX14, SOD1, SOD2, SORD, SPG7, SPR, SPTBN4, SPTSSA, SQOR, SQSTMI, SRD5A3, SSBP1, SSR4, ST3GAL5, STAT2, STT3A, STT3B, STX3, STXP5L, SUCLA2, SUCLG1, SUFU, SUGCT, SUMF1, SUOX, SURF1, SVBP, SYT1, SZT2, TACO1, TAF1, TAF8, TALDO1, TAMM41, TANGO2, TAOK1, TARS2, TAT, TAZ, TBC1D20, TBCD, TBCK, TBLIX, TCN1, TCN2, TDO2, TEFM, TFAM, TFE3, TH, THAP11, THUMPD1, TIAM1, TIMM22, TIMM50, TIMM8A, TIMMDC1, TK2, TKFC, TKT, TMEM106B, TMEM126A, TMEM126B, TMEM163, TMEM165, TMEM184B, TMEM199, TMEM240, TMEM260, TMEM63A, TMEM65, TMEM70, TMLHE, TMTC3, TNNI3, TOMM70, TOP3A, TP11, TPK1, TPPI, TPR, TRAPPC11, TRAPPC12, TRAPPC2L, TRAPPC9, TREX1, TRIAP1, TRIM37, TRIT1, TRMT1, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TSEN54, TSFM, TTC19, TTII, TTPA, TUBA1A, TUBB, TUBB2A, TUBB3, TUBB4A, TUBB4B, TUFM, TWNK, TXN2, TYMP, UBQLN2, UBTF, UFM1, UMPS, UNC80, UPB1, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRH, UQCRQ, UROCI, UROD, UROS, USP7, VAC14, VAMP2, VARS1, VARS2, VMA21, VPS11, VPS13C, VPS13D, VPS16, VPS33A, VPS4A, VPS50, VPS51, VPS53, WARS1, WARS2, WDR45, WDR45B, WFS1, WIPI2, WWOX, XDH, XPRI, YARS1, YARS2, YIF1B, YME1L1, ZBTB42, ZNF526, ZNHIT3.*