

IEM-panel (972 st):
Gener för medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild

AARS2, AASS, ABAT, ABCB7, ABCC8, ABCD1, ABCD4, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD5, ACER3, ACO2, ACOX1, ACSF3, ACY1, ADA, ADAR, ADCK2, ADCY5, ADK, ADPRS, ADSL, ADSS1, AFG3L2, AGA, AGK, AGL, 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, AP1S1, AP3B1, AP3D1, AP5Z1, APOO, APRT, ARG1, ARL3, ARNT2, ARSA, ARSB, ASAH1, ASL, ASNS, ASPA, ASS1, ASXL3, ATAD3A, ATAD3B, ATAD3C, ATIC, ATP13A2, ATP1A1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATPAF2, AUH, B4GALNT1, B4GALT1, B4GALT7, BBOX1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BLOC1S3, BLOC1S6, BOLA3, BSCL2, BTD, C12orf65, C19orf12, C1QBP, CA5A, CACNA1A, CACNA1E, CACNA1S, CAD, CARS1, CARS2, CAT, CBLIF, CBS, CCDC115, CCDC174, CCDC88C, CD320, CEP89, CERS1, CHCHD10, CHKB, CISD2, CLCN2, CLDN16, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CLPX, CLTC, CLTRN, CMPK2, CNKSR2, COA3, COA5, COA6, COA7, COA8, COASY, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COLGALT1, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX16, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6A2, COX6B1, COX7B, COX8A, CP, CPOX, CPS1, CPT1A, CPT2, CRAT, CRPPA, CSF1R, CTCF, CTH, CTNS, CTSB, CTSF, CTSK, CUBN, CWF19L1, CYC1, CYP7B1, D2HGDH, DARS1, DARS2, DBH, DBT, DCAF17, DDC, DEGS1, DGAT1, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHPS, DHTKD1, DIAPH1, DKC1, DLAT, DLD, DMGDH, DNA2, DNAJC12, DNAJC19, DNAJC3, DNAJC30, DNAJC5, DNMT1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DTNBP1, DTYMK, EARS2, EBF3, EBP, ECHS1, ECM1, ECSIT, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELAC2, ELOVL4, ELOVL5, ENO3, EPG5, EPHX1, EPM2A, EPRS1, ERAL1, ERCC6, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC5, EXOSC8, EXT1, EXT2, FA2H, FAH, FAM126A, FAM149B1, FAR1, FARS2, FARSA, FARSB, FASTKD2, FBP1, FBXL4, FCSK, FDF1, 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, GFER, GFM1, GFM2, GGT1, GJC2, GK, GLA, GLB1, GLDC, GLRA1, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAO1, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOT2, GPD1, GPHN, GPR161, GRHRP, GRIN2B, GRN, GSS, GSX2, GTPBP3, GUSB, GYG1, GYG2, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAL, HARS1, HCFC1, HEPACAM, HEXA, HEXB, HGD, HGSNAT, HIBCH, HIKESHI, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HPD, 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, IRF2BPL, ISCA1, ISCA2, ISCU, ITPA, ITPR1, IVD, JAG1, JAM2, KARS1, KCNA4, KCNJ11, KIF5A, KYNU, L2HGDH, LAMA1, LAMP2, LARS1, LARS2, LBR, LDHA, LHX4, LIAS, LIPA, LIPE, LIPT1, LIPT2, LMBRD1, LNPK, LONP1, LPIN1, LRP2, LRPPRC, LYRM4, LYRM7, MADD, MAG, MAGED2, MAGEL2, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MARS1, MARS2, MAT1A, MBD5, MBOAT7, MCAT, MCCC1, MCCC2, MCEE, MCOLN1, MDH1, MECP2, MECR, MFF, MFN2, MFSD8, MGAT2, MGME1, MICOS13, MICU1, MICU2, MIEF2, MIPEP, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOCS3, MOGS, MPC1, MPDU1, MPI, MPV17, MRM2, MRPL12, MRPL13, MRPL44, MRPL57, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS34, MRPS36, MRPS7, MSL3, MSTO1, MTFMT, MTHFD1, MTHFR, MTO1, MTPAP, MTR, MTRR, MVK, MYO5A, MYORG, NAA10, NADK2, NAGA, NAGLU, NAGS, NALCN, NANS, 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, NDUFB8, NDUFB9, NDUFC2, NDUF51, NDUF52, NDUF53, NDUF54, NDUF55, NDUF56, NDUF57, NDUF58, NDUFV1, NDUFV2, NDUFV3, NEU1, NFASC, NFE2L2, NFS1, NFU1, NGLY1, NKX6-2, NMNAT1, NOP10, NPC1, NPC2, NRROS, NSDHL, NSUN3, NT5C3A, NUBPL, NUP188, NUP214, NUS1, OAT, OCRL, OGDH, OPA1, OPA3, OPLAH, OSTC, OTC, OXA1L, OXCT1, OXR1, PAH, PAK1, PANK2, PARK7, PARS2, PAX7, PAXBP1, PC, PCBD1, PCCA, PCCB, PCK1, 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, PIGA, PIGM, PIGN, PIGO, PIGQ, PIGS, PIGT, PIGV, PIGW, PIK3R4, PITRM1, PLA2G6, PLAA, PLIN5, PMM2, PMPCA, PMPCB, PNP, PNPLA2, PNPLA4, PNPLA6, PNPLA8, PNPO, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POLRMT, POMGNT1, POMT1, PPA2, PPCS, PPM1K, PPOX, PPP1R15B, PPP1R21, PPP2R1A, PPP2R5D, PPT1, PRKAG2, PRKN, PRMT7, PRODH, PRODH2, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTC1, PTC3, PTRH2, PTS, PURA, PUS1, PUS7, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, QRSL1, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RALGAPA1, RANBP2, RARS1, RARS2, REPS1, RFT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF13, RNF213, RPIA, RRM2B, RTN4IP1, RYR1, SAMHD1, SARS1, SARS2, SC5D, SCN8A, SCO1, SCO2, SCP2, SCYL1, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SELENOI, SERAC1, SFXN4, SGSH, SHMT2, 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, SLC25A4, SLC25A42, SLC25A46, SLC27A4, SLC2A1, SLC2A2, SLC2A9, SLC30A10, SLC30A9, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC46A1, SLC4A10, SLC52A1, SLC52A2, SLC52A3, SLC5A1, SLC5A2, SLC5A7, SLC6A17, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC7A1, SLC7A2, SLC7A7, SLC7A9, SLC9A1, SLC9A7, SMARCD1, SMPD1, SMPD4, SMS, SNORD118, SNX14, SOD1, SOD2, SORD, SPG7, SPR, SPTBN4, SQOR, SQSTM1, SRD5A3, SSBP1, SSR4, ST3GAL5, STAT2, STT3A, STX3, STXB5L, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, SVBP, SYT1, SZT2, TACO1, TAF1, TALDO1, TANGO2, TAOK1, TARS2, TAT, TAZ, TBC1D20, TBCD, TBCK, TBL1X, TCN1, TCN2, TDO2, TFAM, TH, THAP11, TIMM22, TIMM50, TIMM8A, TIMMDC1, TK2, TKFC, TKT, TMEM106B, TMEM126A, TMEM126B, TMEM165, TMEM199, TMEM240, TMEM260, TMEM63A, TMEM65, TMEM70, TMLHE, TMTC3, TNNI3, TOMM70, TOP3A, TPI1, TPK1, TPP1, TRAPPC11, TRAPPC12, TRAPPC2L, TREX1, TRIAP1, TRIM37, TRIT1, TRMT1, TRMT10C, TRMT5, TRMU, TRNT1, TRPM6, TSEN54, TSFM, TTC19, TTPA, TUBB2A, TUBB3, TUBB4A, TUBB4B, TUFM, TWNK, TXN2, TYMP, UBQLN2, UBTF, UFM1, UMPS, UNC80, UPB1, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRH, UQCRCQ, UROC1, UROD, UROS, USP7, VAC14, VAMP2, VARS1, VARS2, WARS2, WDR45, WDR45B, WFS1, WIPI2, VMA21, VPS11, VPS13D, VPS33A, VPS4A, VPS51, VPS53, WWOX, XDH, XPR1, YARS1, YARS2, YIF1B, YME1L1, ZBTB42, ZNHIT3

NBS-M-panel (46 st):

Gener för nyföddhetscreening för medfödda metabola sjukdomar

ABCD4, ACADM, ACADVL, ACAT1, ARG1, ASL, ASS1, BCKDHA, BCKDHB, BTBD, CBS, CPT1A, CPT2, DBT, DLD, DNAJC12, ETFA, ETFB, ETFDH, FAH, FLAD1, GALT, GCDH, GCH1, HADHA, HADHB, IVD, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MMUT, PAH, PCBD1, PCCA, PCCB, PPM1K, PTS, QDPR, SLC22A5, SLC25A20, SLC52A1, SLC52A2, SLC52A3

EP-panel (427 st)

Gener för epileptisk encefalopati, 136 ingår i listan för medfödda metabola sjukdomar

AARS1, ABAT, ABCA2, ACTL6B, ADAM22, ADAR, ADARB1, ADCY5, ADRA2B, ADSL, AFG3L2, AGTR2, AIMP1, AKT3, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALKBH8, AMT, ANKRD11, AP2M1, AP3B2, AP4B1, AP4S1, APP, ARHGEF9, ARID1B, ARV1, ARX, ASAH1, ASH1L, ASNS, ASXL3, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP6AP2, ATP6V1A, ATP7A, ATRX, ATXN2, BRAF, BRAT1, BRWD3, BSCL2, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA2D1, CACNA2D2, CACNG2, CAD, CAMK2A, CAMK2B, CASK, CDK19, CDKL5, CERS1, CERT1, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP2, COL4A1, COL4A2, CPA6, CPLX1, CRADD, CSNK2A1, CSNK2B, CSTB, CTSD, CTSF, CUX2, CYFIP2, DALRD3, DCX, DDX3X, DEAF1, DENND5A, DEPDC5, DHDDS, DMXL2, DNAJA1, DNAJC5, DNM1, DNM1L, DNMT3A, DOCK7, DOLK, DPM1, DPM2, DPYD, DYNC1H1, DYRK1A, EEF1A2, EHMT1, EIF2S3, EPM2A, FBXO28, FGF12, FGF13, FGFR3, FLNA, FOLR1, FOXG1, FOXP1, FRRS1L, FZR1, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GAL, GAMT, GATM, GCSH, GLDC, GLRA1, GLS, GLYCTK, GNAO1, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPHN, GPT2, GRIA2, GRIA3, GRIA4, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GUF1, HACE1, HCN1, HECW2, HIVEP2, HNRNP2, HNRNPU, HSD17B10, IQSEC2, IRF2BPL, ITPA, KANSL1, KAT5, KCNA1, KCNA2, KCNB1, KCNC1, KCND2, KCND3, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNN3, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM4B, KDM5C, KIF1A, KIF5A, KIF5C, KMT2A, KMT2E, LGI1, LIAS, LMNB2, LNPK, MADD, MAGEL2, MAGI2, MAPK10, MARCHF6, MBD5, MBOAT7, MDH1, MDH2, MECP2, MED13L, MED17, MED23, MEF2C, MFF, MFSB8, MID2, MOCS1, MOCS2, MTHFS, MTOR, NAA10, NACC1, NAPB, NARS1, NBEA, NECAP1, NEDD4, NEUROD2, NEXMIF, NF1, NGLY1, NHLRC1, NPRL2, NPRL3, NR2F1, NR4A2, NRROS, NRXN1, NTNG2, NTRK2, NUP214, NUS1, OPHN1, OTUD6B, OXR1, PACS2, PAFAH1B1, PAK1, PAK3, PARS2, PAX7, PCDH12, PCDH19, PDE2A, PHACTR1, PHF21A, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGW, PLCB1, PLP1, PLPBP, PNKP, PNPO, POLG, PPFIBP1, PPP1CB, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRODH, PRRT2, PRUNE1, PTPN23, PUM1, PURA, QARS1, RAB11A, RAB11B, RAB39B, RALGAPA1, RAPGEF2, RARS2, RBSN, RELN, RFT1, RHOBTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGD1, RORA, RORB, RUSC2, SAMD12, SAMHD1, SCAF4, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SGCE, SHANK3, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC1A4, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC39A8, SLC45A1, SLC6A1, SLC6A8, SLC6A9, SLC9A6, SMARCA2, SMC1A, SNAP25, SORCS3, SPATA5, SPG11, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STAMBP, STARD7, STRADA, STT3B, STX1B, STXBP1, STYXL1, SUMF1, SV2A, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBC1D24, TBC1D2B, TBCD, TBL1XR1, TCF4, TDP2, THOC2, TMTC3, TNRC6A, TPP1, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRPM3, TRPM6, TSC1, TSC2, TUBA1A, TUBB2A, TUBB2B, TUBGCP2, UBA5, UBE3A, UFC1, UGDH, UGP2, UNC80, USP7, VARS1, WASF1, WDR26, WDR37, WDR45, WDR45B, WDR62, WWOX, YEATS2, YIF1B, YWHAG, ZBTB18, ZEB2, ZNF142, ZNHIT3

PEDHEP-panel (156 st):

Gener för leversjukdomar hos barn, 60 ingår i listan för medfödda metabola sjukdomar

ABCA1, ABCB11, ABCB4, ABCC2, ABCD3, ACOX2, AGL, AKR1D1, ALAS2, ALDOB, ALG3, ALG6, ALG8, ALG9, ALMS1, AMACR, ANKS6, ARG1, ASAH1, ASS1, ATP6AP1, ATP7B, ATP8B1, BAAT, BBS1, BCS1L, CC2D2A, CCDC115, CEP164, CEP290, CEP41, CEP83, CLDN1, COG2, COG4, COG5, COG6, COG7, CSPP1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DNAJB11, DNAJC21, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, EFL1, EIF2AK3, FAH, FBP1, FECH, G6PC, G6PD, GALE, GALT, GANAB, GATA6, GBE1, GDF1, GPD1, GPI, GUCY2D, GYS2, HAMP, HFE, HJV, HSD17B4, HSD3B7, IFT140, IFT172, IFT43, IFT52, IFT81, INPP5E, INTU, JAG1, KIF12, KRT18, KRT8, LARS1, LIPA, LRP5, LSR, MPI, MPV17, MYO5B, NBAS, NEK1, NGLY1, NOTCH2, NPC1, NPC2, NPHP3, NR1H4, OFD1, PGM1, PHKA2, PHKB, PHKG2, PKD1, PKD2, PKHD1, PKLR, PMM2, POLG, PPM1F, PRF1, PRKCSH, PYGL, RINT1, RPGRIP1L, SBDS, SC5D, SCYL1, SEC63, SERPINA1, SKIV2L, SLC10A1, SLC10A2, SLC25A13, SLC25A20, SLC27A5, SLC2A2, SLC37A4, SLCO1B1, SLCO1B3, SMPD1, SRP54, TALDO1, TCTN2, TFR2, TJP2, TMEM107, TMEM199, TMEM216, TMEM67, TRAF3IP1, TRMU, TTC21B, TTC26, TTC37, TWNK, UGT1A1, UNC13D, UNC45A, USP53, WDR19, WDR35, WDR83OS, VIPAS39, VPS33B, XPNPEP3

MOVE-panel (323 st):

Gener för medfödda rörelsesjukdomar hos barn, 183 ingår i listan för medfödda metabola sjukdomar

AARS1, AARS2, ABCD1, ABHD12, ACER3, ACOX1, ACP5, ACTB, ADAR, ADCY5, ADPRS, AFG3L2, AGTPBP1, AHI1, ALDH18A1, ALDH3A2, ALDH6A1, ALG6, ALS2, ANO10, ANO3, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARL13B, ARSA, ARV1, ARX, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATXN10, AUH, B4GALT1, BCAP31, BCS1L, BSCL2, C19orf12, CA8, CACNA1A, CACNA1B, CACNA1E, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CARS2, CASK, CDKL5, CEP104, CHMP1A, CLCN2, CLN3, CLN5, CLP1, COA7, COASY, COG4, COG5, COL6A3, COQ2, COQ8A, COX10, COX15, COX20, CP, CSF1R, CSPP1, CSTB, CTBP1, CTC1, CTD1P1, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DARS2, DCAF17, DDC, DDHD1, DDHD2, DEGS1, DHCR24, DHFR, DKC1, DLAT, DLD, DNAJC12, DNAJC19, DNAJC5, DNAJC6, EARS2, ECHS1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, ELOVL4, EMC1, ENTPD1, ERLIN1, ERLIN2, EXOSC9, FA2H, FAM126A, FARS2, FBXL4, FBXO7, FGF14, FLVCR1, FOXG1, FRRS1L, FTL, FUS, FXN, GABBR2, GALT, GAMT, GAN, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJB1, GJC2, GLRA1, GM2A, GNAL, GNAO1, GNB1, GOSR2, GPAA1, GRID2, GRIN1, GRIN2B, GRM1, GTPBP2, HARS1, HEPACAM, HEXA, HPCA, HPRT1, HSPD1, HTRA2, HTT, IARS1, IFIH1, INPP5E, INPP5K, IREB2, ITPR1, JAM3, KCNA1, KCNA2, KCNC3, KCND3, KCNMA1, KCNQ2, KCTD17, KIF1A, KIF1C, KIF5A, KMT2B, L2HGDH, LIPT1, LIPT2, MARS2, MCOLN1, MDH2, MECP2, MECP2, MICU1, MLC1, MRE11, MSTO1, MTFMT, MYORG, NDUFAF2, NDUFAF6, NDUFS1, NDUFS8, NFASC, NIPA1, NKX2-1, NKX6-2, NPC1, NPC2, NT5C2, NUP62, OPA3, PANK2, PARK7, PCDH12, PCYT2, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PEX10, PEX2, PEX7, PGK1, PGM3, PINK1, PLA2G6, PLEKHG2, PLP1, PMPCA, PNKD, PNKP, PNPLA6, POLG, POLR1C, POLR3B, PPP2R2B, PRICKLE1, PRKCG, PRKN, PRKRA, PRPS1, PRRT2, PSAP, PTS, QDPR, RAB39B, RARS1, REEP1, RELN, RNASEH2B, RNASET2, RNF216, RORA, RTN2, SACS, SAMD9L, SAMHD1, SCN8A, SCP2, SCYL1, SERAC1, SETX, SGCE, SIGMAR1, SIL1, SLC16A2, SLC18A2, SLC19A3, SLC1A3, SLC20A2, SLC25A15, SLC2A1, SLC30A10, SLC39A14, SLC6A3, SLC6A8, SLC9A1, SLC9A6, SMPD1, SNCA, SNORD118, SPAST, SPR, SPTBN2, SPTLC2, SQSTM1, STAMBP, STUB1, STXBP1, SUMF1, SURF1, SYNE1, SYNJ1, TACO1, TAF1, TBC1D24, TGM6, TH, THAP1, TIMM8A, TMEM240, TOR1A, TPK1, TPP1, TRAPPC11, TREX1, TTC19, TTPA, TUBB4A, TWNK, UBA5, UBAP1, UBQLN2, UCHL1, VAC14, VAMP1, VAMP2, WDR45, WDR73, WDR81, WFS1, VLDLR, VPS13A, VPS13B, VPS13C, VPS13D, WWOX, XPR1, ZFYVE26, ZNF142

ENDO-panel (362 st):

Gener för medfödda endokrina sjukdomar, 45 ingår i listan för medfödda metabola sjukdomar

AAAS, AARS2, ABCC8, ABCD1, AGPAT2, AIRE, AKT2, ALB, ALMS1, ALPL, AMH, AMHR2, AMMECR1, ANKRD11, ANOS1, AP2S1, APPL1, AQP2, AR, ARL6, ARMC5, ARX, ATRX, AVP, AVPR2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BLK, BLM, BMP15, BRAF, BSCL2, BSND, BTK, CACNA1H, CASR, CAVIN1, CBL, CCDC141, CCDC8, CDC6, CDC73, CDCA8, CDKN1B, CDKN1C, CDT1, CEL, CENPJ, CEP19, CHD7, CIDEA, CISD2, CLCN5, CLCNKA, CLCNKB, CLDN16, CLPP, CNOT1, CREBBP, CRIPT, CTLA4, CUL4B, CUL7, CYB5A, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A2, CYP24A1, CYP27B1, CYP2R1, DCAF17, DCC, DCHS1, DHCR7, DHH, DIO1, DIO2, DIO3, DLK1, DMP1, DNAJC3, DUOX1, DUOX2, DUOXA2, DYRK1B, EIF2AK3, EIF2B5, EIF2S3, ENPP1, EP300, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FEZF1, FGD1, FGF23, FGF8, FGFR1, FHL1, FOXA2, FOXE1, FOXL2, FOXP3, FSHB, FSHR, GALT, GATA3, GATA4, GATA6, GCK, GCM2, GH1, GH2, GHR, GHRH, GHRHR, GHSR, GLI2, GLI3, GLIS3, GLUD1, GNA11, GNAS, GNAS-AS1, GNRH1, GNRHR, GPC3, HADH, HAMP, HDAC8, HESX1, HFE, HFM1, HJV, HNF1A, HNF1B, HNF4A, HRAS, HS6ST1, HSD11B2, HSD17B3, HSD17B4, HSD3B2, IER3IP1, IGF1, IGF1R, IGFALS, IGSF1, IL17RD, IL2RA, INS, INSR, IRS4, ITCH, IYD, JAG1, KCNJ1, KCNJ11, KCNJ5, KDM6A, KIF14, KISS1R, KLF11, KMT2D, KRAS, LARS2, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LIG4, LIPE, LMNA, LRBA, MAFA, MAMLD1, MAP2K1, MAP2K2, MAP3K1, MC2R, MC4R, MCM8, MCM9, MEN1, MKKS, MKRN3, MKS1, MNX1, MRAP, MYRF, MYT1L, NBN, NDN, NEUROD1, NEUROG3, NIPBL, NKX2-1, NKX2-2, NKX2-5, NNT, NOBOX, NROB1, NR3C1, NR3C2, NR5A1, NRAS, NSMF, NTN1, NTRK2, OBSL1, ORC1, ORC4, ORC6, OTX2, PAPP2, PAX4, PAX6, PAX8, PCBD1, PCK1, PCNT, PCSK1, PCYT1A, PDE11A, PDE8B, PDX1, PDZK1IP1, PGM1, PHEX, PHF6, PIK3R1, PITX2, PLIN1, PMM2, PNPLA6, POLD1, POLG, POMC, POR, POU1F1, PPARG, PPP1R15B, PRKAR1A, PRMT7, PROK2, PROKR2, PROP1, PTF1A, PTH, PTH1R, PTPN11, RAD21, RAF1, RBBP8, RET, RFX6, RIT1, RNU4ATAC, ROBO2, ROR2, RPL10, RPS6KA3, RSPO1, SAMD9, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SECISBP2, SERPINA6, SERPINA7, SGPL1, SHOC2, SHOX, SIM1, SLC12A1, SLC12A3, SLC16A1, SLC16A2, SLC19A2, SLC26A4, SLC26A7, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC40A1, SLC5A2, SLC5A5, SMC1A, SMC3, SNRPN, SOHLH1, SOS1, SOX10, SOX2, SOX3, SOX9, SRA1, SRCAP, SRD5A2, SRY, STAG3, STAR, STAT3, STAT5B, STX16, TAC3, TACR3, TBCE, TBL1X, TBX1, TBX19, TCF12, TFR2, TG, THRA, THRB, TOE1, TPO, TRH, TRHR, TRIM37, TRMT10A, TRPM6, TSHB, TSHR, TTC8, TWNK, UCP2, VDR, WDR11, WFS1, WNK1, WNK4, VPS13B, WRN, WT1, XRCC4, ZBTB20, ZFP57, ZFPM2, ZMPSTE24

DIAB-panel (57 st):

Gener för monogen diabetes och kongenital hyperinsulinemi, alla ingår i listan för endokrina sjukdomar

ABCC8, AGPAT2, AIRE, AKT2, APPL1, BLK, BSCL2, CEL, CIDEA, CISD2, CNOT1, COQ2, COQ9, CTLA4, DCAF17, DNAJC3, DYRK1B, EIF2AK3, EIF2B1, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, IL2RA, INS, INSR, ITCH, KCNJ11, KLF11, LIPE, LMNA, MAFA, NEUROD1, NEUROG3, NKX2-2, PAX4, PAX6, PCBD1, PDX1, PIK3R1, POLD1, PPARG, PPP1R15B, PTF1A, RFX6, SLC19A2, SLC29A3, SLC2A2, TRMT10A, WFS1, ZFP57, ZMPSTE24