

Centrum för Medfödda Metabola Sjukdomar

Genpanel IEM version 20 (1046 gener)**Gener kopplade till medfödda metabola sjukdomar eller andra monogena sjukdomar med liknande klinisk bild (inborn errors of metabolism)**

AARS2, AASS, ABAT, ABCB7, ABCC8, ABCD1, ABCD4, ABHD12, ABHD16A, 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, 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, AP1S1, AP3B1, AP3D1, AP5Z1, APOO, APRT, ARF3, ARG1, ARL3, ARNT2, ARSA, ARSB, ARSK, ASAH1, ASL, ASNS, ASPA, ASS1, ASXL3, ATAD3A, ATAD3B, ATAD3C, ATIC, ATP13A2, ATP1A1, ATP1A3, ATP2B1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATP6AP1, ATP6V0A2, ATP7A, ATP7B, ATP9A, ATPAF2, AUH, B3GALNT2, B4GALNT1, B4GALT1, B4GALT7, BBOX1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCKDK, BCS1L, BLOC1S3, BLOC1S6, BOLA3, BSCL2, BTD, C12orf65, C19orf12, C1QBP, CA5A, CACNA1A, CACNA1E, CACNA1S, CAD, CAMLG, CAMSAP1, CARS1, CARS2, CAT, CBLIF, CBS, CCDC115, CCDC174, CCDC88C, CD320, CEP89, CERS1, CHCHD10, CHKA, CHKB, CISD2, CLCN2, CLCN6, CLDN11, 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, COX11, COX14, COX15, COX16, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6A2, COX6B1, COX7B, COX8A, CP, CPOX, CPS1, CPT1A, CPT2, CRAT, CRLS1, 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, DNMI1, 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, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC5, EXOSC8, EXT1, EXT2, FA2H, FAH, FAM126A, FAM149B1, FAR1, FARS2, FARSA, FARSB, FASTKD2, FBP1, FBXL4, 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, 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, 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, HPDL, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HSD17B10, HSD17B4,

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