

Centrum för Medfödda Metabola Sjukdomar

Genpanel MIT version 20 (286 gener)

Nukleära gener kopplade till mitokondriella sjukdomar. Alla gener ingår i IEM-panelen version 20.

AARS2, ACAD9, ADCK2, AFG3L2, AGK, AIFM1, ALDH1L2, ALKBH1, ANTXR2, APOO, APOPT1, ARNT2, ATAD3A, ATAD3B, ATAD3C, ATP5D, ATP5E, ATP5F1A, ATP6AP1, ATPAF2, BCS1L, BOLA3, C12ORF65, C19orf70, C1QBP, CA5A, CARS2, CEP89, CHCHD10, CISD2, CLPB, CLPP, CMPK2, COA3, COA5, COA6, COA7, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ9, COX10, COX11, COX14, COX15, COX16, COX20, COX4I1, COX4I2, COX5A, COX6A1, COX6B1, COX7B, COX8A, CRAT, CRLS1, CYC1, DARS2, DCAF17, DGUOK, DNA2, DNAJC19, DNAJC30, DNMI1, DTYMK, EARS2, ECHS1, ECSIT, ELAC2, EPG5, ERAL1, ETHE1, FARS2, FASTKD2, FBXW7, FDX1L, FDXR, FOXRED1, FTL, FXN, GARS, GATB, GATC, GFER, GFM1, GFM2, GTPBP3, HSPD1, HSPE1, IARS2, IBA57, ISCA1, ISCA2, ISCU, ITPR1, KARS, KIF5A, LARS2, LETM1, LIG3, LONP1, LRPPRC, LYRM7, MCAT, MECR, MFF, MFN2, MGME1, MICU1, MICU2, MIEF2, MIPEP, MPC1, MPV17, MRM2, MRPL12, MRPL24, MRPL3, MRPL44, MRPL57, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS34, MRPS7, MSTO1, MTO1, MTPAP, NADK2, NARS2, NAXD, NAXE, 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, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFS1, NFU1, NSUN3, NUBPL, NUP62, OGDHL, OPA1, OPA3, OXA1L, P4HTM, PARK7, PARS2, PDE12, PDE2A, PDHA1, PDHB, PDHX, PDK3, PDSS1, PDSS2, PET100, PET117, PITRM1, PMPCB, PNPLA4, PNPLA8, PNPT1, POLG, POLG2, POLRMT, PPA2, PRORP, PTCDD1, PTCDD3, PUS1, QRSL1, RARS2, REPS1, RMND1, RNASEH1, RRM2B, RTN4IP1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SFXN4, SHMT2, SLC25A1, SLC25A10, SLC25A19, SLC25A21, SLC25A24, SLC25A3, SLC25A32, SLC25A4, SLC25A42, SLC25A46, SLIRP, SPG7, SQOR, SUCLA2, SUCLG1, SURF1, TACO1, TAMM41, TAOK1, TARS2, TAZ, TFAM, TIMM22, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM65, TMEM70, TNNI3, TOMM70, TOP3A, TRIAP1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRCFS1, UQCRH, UQCRQ, USMG5, VARS2, VMA21, VPS13C, VPS13D, WARS2, YARS2.