

Nukleáris mitokondriális betegségek (Nuclear mitochondrial disorders) génpanel v:2022.01.26.

Panel tartalma 432 gén:

AARS2, AASS, ABAT, ABCB7, ACACA, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ADAR, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH3A2, AMPD1, AMT, ANO10, APOPT1, APTX, ATAD3A, ATP5A1, ATP5B, ATP5C1, ATP5D, ATP5E, ATP5F1, ATP5G1, ATP5G2, ATP5G3, ATP5H, ATP5I, ATP5J, ATP5J2, ATP5L, ATP5L2, ATP5O, ATP7B, ATPAF1, ATPAF2, AUH, BAG3, BCS1L, BOLA3, BTBD, C12orf65, C19orf12, C19orf70, C1QBP, CA5A, CARS2, CEP89, CHAT, CHCHD10, CHKB, CISD2, CLPB, CLPP, COA1, COA3, COA4, COA5, COA6, COA7, COASY, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX11, COX14, COX15, COX16, COX17, COX18, COX19, COX20, COX4I1, COX4I2, COX5A, COX5B, COX6A1, COX6A2, COX6B1, COX6B2, COX6C, COX7A1, COX7B, COX7C, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS, DARS2, DCC, DES, DGUOK, DHTKD1, DLAT, DLD, DNA2, DNAJC19, DNM1L, DNM2, EARS2, ECHS1, ECSIT, ELAC2, ERAL1, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FH, FLAD1, FOXRED1, FXN, G6PC, GAMT, GARS, GATB, GATC, GATM, GCDH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GLUD1, GTPBP3, GYG2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HPDL, HSD17B10, HSPA9, HSPD1, HTRA2, HTT, IARS2, IBA57, IDH2, IDH3A, IDH3B, IER3IP1, IFIH1, ISCA1, ISCA2, ISCU, KARS, L2HGDH, LAMP2, LARS, LARS2, LIAS, LIG3, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM4, LYRM7, MARS2, MDH2, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MPC1, MPV17, MRM2, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34, MRPS7, MSTO1, MTFMT, MTHFD1, MTO1, MTPAP, NADK2, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA3, NDUFA4, NDUFA5, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAB1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFAF8, NDUFB1, NDUFB10, NDUFB11, NDUFB2, NDUFB3, NDUFB4, NDUFB5, NDUFB6, NDUFB7, NDUFB8, NDUFB9, NDUFC1, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFS1, NFU1, NGLY1, NNT, NR2F1, NSUN3, NUBPL, NUP62, OGDH, OPA1, OPA3, OTC, OXA1L, OXCT1, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDP2, PDPR, PDSS1, PDSS2, PET100, PET117, PINK1, PITRM1, PMPCA, PMPCB, PNKD, PNPLA4, PNPLA8, PNPT1, POLG, POLG2, POLRMT, POP1, PPA2, PPOX, PSAP, PTCD3, PUS1, PYCR1, QARS, QRSL1, RANBP2, RARS, RARS2, REEP1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, ROBO3, RRM2B, RTN4IP1, SACS, SAMHD1, SARS2, SCN1A, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHAF3, SDHAF4, SDHB, SDHC, SDHD, SERAC1, SFXN4, SIRT1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A40, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC52A3, SLC6A8, SLC7A13, SPAST, SPG7, SQOR, SRRT, SSBP1, STAT2, STXBP1, SUCLA2, SUCLG1, SUCLG2, SUGCT, SURF1, TACO1, TANGO2, TARS2, TAZ, TFAM, TIMM22, TIMM44, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM65, TMEM70, TOP1MT, TOP3A, TPK1, TRAP1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC1, UQCC2, UQCC3,

UQCR10, UQCR11, UQCRB, UQCRC1, UQCRC2, UQCRFS1, UQCRH, UQCRQ, VARS2, VPS13C, WARS2, WDR45, WFS1, XPNPEP3, YARS2, YME1L1.

Forrás:

Possible mitochondrial disorder - nuclear genes (Version 1.64)

<https://panelapp.genomicsengland.co.uk/panels/539/> (Megtekintve: 2022.01.26.)

Invitae Nuclear Mitochondrial Disorders Panel

<https://www.invitae.com/en/physician/tests/98002/> (Megtekintve: 2022.01.26.)