Molecular Testing for Mitochondrial Diseases

Panels available and list of genes

Full Nuclear Gene Panel - 406 genes

Subpanels:

1. Mitochondrial Encephalopathy/Leigh Disease – 119 genes
2. Complex I deficiency – 35 genes
3. Complex IV deficiency – 21 genes
4. Complex II + III + V + Coenzyme Q + PDH deficiencies – 26 genes
5. mtDNA Depletion and Deletion Panel – 21 genes
6. Progressive External Ophthalmoplegia (PEO)/Optic Atrophy – 23 genes

Full panel - 406 genes associated with mitochondrial diseases

AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACACB, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACAT2, ACLY, ACO2, ACSL4, ACSL5, ACSM3, ADCK3, ADSL, AFG3L2, AGK, AGL, AGPS, AGXT, AGXT2, AIFM1, AK2, AKAP10, AKR7A2, ALAS2, ALDH18A1, ALDH1B1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMPD1, AMT, ANKRD26, APTX, ARG1, AS3MT, ATIC, ATP10D, ATP5A1, ATP5E, ATPAF2, AUH, BAX, BCAT1, BCAT2, BCKDHA, BCKDHB, BCL2, BCS1L, BOLA3, BTD, C12orf65, CASP8, CHAT, CHDH, CISD2, CLYBL, COA5, COMT, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6B1, COX7A1, COX7A2, CPOX, CPS1, CPT1A, CPT1B, CPT2, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, DARS2, DBT, DDAH1, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ECSIT, ELAC2, ELN, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASN, FASTKD2, FBP1, FBXL4, FECH, FH, FOXRED1, FPGS, FTH1, FXN, G6PC, GAA, GALC, GARS, GATM, GBE1, GCDH, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLS, GLUD1, GLYCTK, GPAM, GPD1, GPD2, GPX1, GPX4, GSR, GSTK1, GYS1, GYS2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HIGD2A, HK1, HK2, HLCS, HMGCL, HMGCS2, HSD17B10, HSD17B4, HSD3B1, HSPA9, HSPB7, HSPD1, HTRA2, IDE, IDH1, IDH2, IDH3B, IMMP2L, IMMT, ISCU, IVD, KARS, KIF1B, KRT5, KYNU, L2HGDH, LARS, LARS2, LDHA, LDHB, LETM1, LIAS, LMBRD1, LPIN1, LRPPRC, MAOA, MAOB, MARS2, MCCC1, MCCC2, MCEE, MDH1, MFN2, MGME1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MPC1, MPI, MPST, MPV17, MRPL3, MRPL40, MRPL44, MRPL48, MRPS16, MRPS18A, MRPS2, MRPS22, MRRF, MTCH2, MTFMT, MTHFD1, MTHFD1L, MTHFS, MTO1, MTPAP, MTRR, MUT, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB3, NDUFB6, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFU1, NIPSNAP1, NIPSNAP3A, NLRX1, NME1, NUBPL, OAT, OGG1, OPA1, OPA3, OTC, OXCT1, PACRG, PAK7, PANK2, PARK7, PARL, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX11B, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PINK1, PMM2, PMPCA, PNKD, PNPT1, POLG, POLG2, POLRMT, PPOX, PRODH, PTGES2, PTS, PUS1, PYCR1, PYGL, PYGM, QDPR, RAB11FIP5, RARS2, REEP1, RMND1, RPL35A, RRM2B, SARDH, SARS2, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHAF2, SDHB, SDHD, SECISBP2, SE-RAC1, SHMT1, SI, SIRT3, SIRT5, SLC16A1, SLC19A3, SLC22A4, SLC22A5, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A39, SLC25A4, SLC37A4, SOD1, SOD2, SPG7, SPR, SPTLC2, STAR, SUCLA2, SUCLG1, SUCLG2, SUGCT, SUOX, SURF1, TACO1, TAZ, TCIRG1, TCN2, TFAM, TFB1M, TIMM44, TIMM8A, TK2, TMEM126A, TMEM70, TOMM20, TOMM40, TOP1MT, TPK1, TRMU, TRNT1, TSFM, TSPO, TST, TTC19, TUFM, TUSC3, TXN2, TXNRD2, TYMP, TWNK, UCP1, UCP2, UCP3, UNG, UQCRB, UQCRQ, WFS1, YARS2.

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1. Mitochondrial Encephalopathy/Leigh Disease – 119 genes

AARS2, ACAD9, ACO2, ADCK3, AFG3L2, AIFM1, APTX, ATP5E, ATPAF2, BCS1L, BOLA3, COQ2, COQ9, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6B1, COX7A1, DARS2, DGUOK, DLAT, DLD, DNM1L, EARS2, ETFDH, ETHE1, FARS2, FASTKD2, FH, FOXRED1, GFER, GFM1, GFM2, HLCS, HSPD1, LARS2, LIAS, LMBRD1, LRPPRC, MARS2, MFN2, MPV17, MRPS16, MTFMT, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA7, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFU1, NUBPL, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PNPT1, POLG, RARS2, RMND1, RRM2B, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHD, SERAC1, SLC19A3, SUCLA2, SUCLG1, SUCLG2, SURF1, TACO1, TIMM44, TK2, TMEM70, TOMM20, TPK1, TRMU, TSFM, TTC19, TUFM, TUSC3, TYMP, TWNK, UQCRB, UQCRQ, YARS2.

2. Complex I deficiency – 35 genes

ACAD9, MTFMT, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA7, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB3, NDUFB6, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NUBPL, POLG, SUCLA2.

3. Complex IV deficiency – 21 genes

COA5, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6B1, COX7A1, ETHE1, FASTKD2, LRPPRC, OPA1, OPA3, POLG, RRM2B, SCO1, SCO2, SPG7, SURF1, TACO1.

4. Complex II + III + V + Coenzyme Q + PDH deficiencies – 26 genes

ADCK3, APTX, ATP5E, ATPAF2, BCS1L, COQ2, COQ6, COQ7, COQ9, DLAT, ETFDH, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, SDHA, SDHAF1, SDHAF2, SDHB, SDHD, TMEM70, TTC19, UQCRB, UQCRQ .

5. mtDNA Depletion and Deletion Panel – 21 genes

AGK, APTX, , DGUOK, DNA2, FBXL4, GFER, MFN2, MGME1, MPV17, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, SUCLG2, TK2, TYMP, TWNK.

6. Progressive External Ophthalmoplegia (PEO)/Optic Atrophy – 23 genes

ACO2, AUH, C12ORF65, CISD2, DGUOK, MFN2, MGME1, MTPAP, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC19A3, SLC25A4, SPG7, TIMM8A, TK2, TMEM126A, TYMP, TWNK, WFS1.

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