

4.15 Patologie Mitocondriali

Intero genoma mitocondriale (37 geni + D-LOOP)	
	MT-ATP6
	MT-ATP8
	MT-CO1
	MT-CO2
	MT-CO3
	MT-CYB
	MT-ND1
	MT-ND2
	MT-ND3
	MT-ND4
	MT-ND4L
	MT-ND5
	MT-ND6
	MT-RNR1
	MT-RNR2
	MT-TA
	MT-TC
	MT-TD
	MT-TE
	MT-TF
	MT-TG
	MT-TH
	MT-TI
	MT-TK
	MT-TL1
	MT-TL2
	MT-TM
	MT-TN
	MT-TP
	MT-TQ
	MT-TR
	MT-TS1
	MT-TS2
	MT-TT
	MT-TV
	MT-TW

254 Geni nucleari associati a mitocondriopatie	
	AARS1
	AARS2
	AASS
	ABAT

ABCA4
ABCB6
ABCB7
ACAD8
ACAD9
ACADM
ACADS
ACADSB
ACADVL
ACAT1
ACO2
ACSL4
AFG3L2
AGK
AGXT
AIFM1
AK2
ALAS2
ALDH18A1
ALDH4A1
ALDH5A1
ALDH6A1
AMACR
AMT
APTX
ATP5F1A
ATP5F1E
ATP7B
ATPAF2
AUH
BCKDHA
BCKDHB
BCS1L
BRIP1
CAVIN1
CISD2
COQ2
COQ8A
COQ9
COX10
COX15
COX4I2
COX6B1
CPOX
CPS1
CPT1A

CPT2
CRBN
CYC1
CYCS
CYP11A1
CYP11B1
CYP11B2
CYP24A1
CYP27A1
CYP27B1
DARS2
DBT
DGUOK
DHODH
DIABLO
DLAT
DLD
DMGDH
DNA2
DNAJB2
DNAJC19
DNAJC30
DNM1L
ECHS1
ETFA
ETFB
ETFDH
ETHE1
FASTKD2
FBP1
FBXL4
FECH
FH
FOXRED1
FXN
G6PC1
GARS1
GATM
GCDH
GCSH
GDAP1
GFER
GFM1
GK
GLDC
GLUD1

GSR
GYS2
HADH
HADHA
HADHB
HAX1
HK1
HLCS
HMGCL
HMGCS2
HSD3B2
HSPA9
HSPB1
HSPB8
HSPD1
HTRA2
IDH2
IDH3B
IGHMBP2
ISCU
IVD
JPH1
KIF1B
LARS2
LIAS
LMNA
LRPPRC
LRSAM1
LYRM7
MAOA
MARS1
MCCC1
MCCC2
MCEE
MED25
MFN2
MGME1
MIP
MLYCD
MMAA
MMAB
MME
MMUT
MPV17
MPZ
MRPL3

MRPS16
MRPS22
MTRFR
MTRR
NDUFA1
NDUFA10
NDUFA11
NDUFA12
NDUFA13
NDUFA2
NDUFA9
NDUFAF1
NDUFAF2
NDUFAF3
NDUFAF4
NDUFAF5
NDUFAF6
NDUFB3
NDUFB9
NDUFS1
NDUFS2
NDUFS3
NDUFS4
NDUFS6
NDUFS7
NDUFS8
NDUFV1
NDUFV2
NEFL
NUBPL
OAT
OPA1
OPA3
OTC
OXCT1
PANK2
PARK7
PC
PCCA
PCCB
PDHA1
PDHB
PDHX
PDP1
PDSS1
PDSS2

PDX1
PINK1
POLG
POLG2
PPOX
PPP2R2B
PRODH
PUS1
RARS2
RNASEH1
RNASEL
RRM2B
SARDH
SARS2
SCO1
SCO2
SCP2
SDHA
SDHAF1
SDHB
SDHC
SDHD
SLC19A3
SLC25A12
SLC25A13
SLC25A15
SLC25A19
SLC25A20
SLC25A22
SLC25A3
SLC25A4
SLC9A6
SOD2
SPG7
STAR
SUCLA2
SUCLG1
SUOX
SURF1
TACO1
TAFAZZIN
TFAM
TIMM44
TIMM8A
TIMMDC1
TK2

TMEM126A
TMEM126B
TMEM70
TMLHE
TRMU
TRPV4
TSFM
TTC19
TUFM
TWNK
TYMP
UNG
UQCC2
UQCC3
UQCRB
UQCRC2
UQCRQ
WFS1
WWOX
YARS2