PANEL NMG4
DEFECTOS DE LA CADENA RESPIRATORIA MITOCONDRIAL ASOCIADOS A GENES NUCLEARES

GENES QUE CODIFICAN PARA EL COMPLEJO I MITOCONDRIAL	
ACAD9	ACAD9 DEFICIENCY
C20ORF7	MITOCHONDRIAL COMPLEX 1 DEFICIENCY
C8ORF38	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX I DEFICIENCY
FOXRED1	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFA1	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFA2	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFA3	NAP
NDUFA4	NAP
NDUFA4L2	NAP
NDUFA5	NAP
NDUFA9	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFA10	LEIGH SYNDROME
NDUFA11	MITOCHONDRIAL COMPLEX I DEFICIENCY
RS2	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX 1 DEFICIENCY
NDUFA13	{THYROID CARCINOMA, HURTHLE CELL}
NDUFAF1	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFAF2	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFAF4	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFB1	NAP
NDUFB3	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFB2	NAP
NDUFB4	NAP
NDUFB5	NAP
NDUFB6	NAP
NDUFB7	NAP
NDUFB8	NAP
NDUFB9	?MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFB10	NAP
NDUFB11	NAP
NDUFS1	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFS2	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFS3	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFS4	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFS5	NAP
NDUFS6	COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF
NDUFS7	LEIGH SYNDROME
NDUFS8	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFV1	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFV2	MITOCHONDRIAL COMPLEX I DEFICIENCY
NDUFV3	NAP
NUBPL	MITOCHONDRIAL COMPLEX I DEFICIENCY
GENES QUE CODIFICAN PARA EL COMPLEJO II	
SDHA	CARDIOMYOPATHY, DILATED, 1GG
SDHB	COWDEN SYNDROME 2
SDHC	GASTROINTESTINAL STROMAL TUMOR
SDHD	CARCINOID TUMORS, INTESTINAL
SDHAF1	MITOCHONDRIAL COMPLEX II DEFICIENCY
SDHAF2	PARAGANGLIOMAS 2
GENES QUE CODIFICAN PARA EL COMPLEJO III	
BCS1L	MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 1
UQCRQ	MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 4
UQCRB	MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 3
UQCR10	NAP
UQCR11	NAP
UQCRC1	NAP
UQCRC2	MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 5
UQCRFS1	NAP
UQCRH	NAP
UQCRHL	NAP
TTC19	MITOCHONDRIAL COMPLEX III DEFICIENCY, NUCLEAR TYPE 2
GENES QUE CODIFICAN PARA EL COMPLEJO IV	
COX10	ENCEPHALOPATHY, PROGRESSIVE MITOCHONDRIAL, WITH PROXIMAL RENAL TUBULOPATHY DUE TO CYTOCHROME C
COX11	NAP

COX15	LEIGH SYNDROME DUE TO CYTOCHROME C OXIDASE DEFICIENCY
COX16	NAP
COX17	NAP
COX18	NAP
COX19	NAP
COX6A1	NAP
COX6A2	NAP
COX6B1	CYTOCHROME C OXIDASE DEFICIENCY
COX6B2	NAP
SCO1	HEPATIC FAILURE, EARLY ONSET, AND NEUROLOGIC DISORDER
SCO2	CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME C OXIDASE DEFICIENCY 1
SURF1	LEIGH SYNDROME, DUE TO COX DEFICIENCY
TACO1	MITOCHONDRIAL COMPLEX IV DEFICIENCY
C2ORF64	MITOCHONDRIAL COMPLEX IV DEFICIENCY
C12ORF62	MITOCHONDRIAL COMPLEX IV DEFICIENCY
FASTKD2	MITOCHONDRIAL COMPLEX IV DEFICIENCY
COX6B1	CYTOCHROME C OXIDASE DEFICIENCY
LRPPRC	LEIGH SYNDROME, FRENCH-CANADIAN TYPE
COX4I1	NAP
COX4I2	EXOCRINE PANCREATIC INSUFFICIENCY, DYSERYTHROPOIETIC ANEMIA, AND CALVARIAL HYPEROSTOSIS
COX4NB	NAP
CYCS	THROMBOCYTOPENIA 4
GENES QUE CODIFICAN PARA EL COMPLEJO V	
ATPAF1	NAP
ATPAF2	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 1
TMEM70	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 2
ATP5E	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 3
ATP5D	NAP
ATP5B	NAP
ATP5A1	?MITOCHONDRIAL COMPLEX (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 4
ATP5C1	NAP
ATP5O	NAP
ATP5F1	NAP
ATP5G1	NAP
ATP5G2	NAP
ATP5G3	NAP
GENES QUE CODIFICAN PARA LA PIRUVATO DESHIDROGENASA	
DLD	DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY
PDHA1	PYRUVATE DEHYDROGENASE E1-ALPHA DEFICIENCY
DLAT	PYRUVATE DEHYDROGENASE E2 DEFICIENCY
PDHX	LACTICACIDEMIA DUE TO PDX1 DEFICIENCY
PDHB	PYRUVATE DEHYDROGENASE E1-BETA DEFICIENCY
PDHA2	NAP
PDP1	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY
PDP2	NAP
LIAS	PYRUVATE DEHYDROGENASE LIPOIC ACID SYNTHETASE DEFICIENCY
GENES ASOCIADOS A DEFICIENCIAS PRIMARIAS DE COENZYMA Q10	
COQ2	COQ10D1 (COENZYME Q10 DEFICIENCY, PRIMARY, 1)
COQ3	NAP
COQ4	NAP
COQ5	NAP
PDSS2	COQ10D3
PDSS1	COQ10D2
CABC1	COQ10D4
COQ9	COQ10D5
COQ10A	NAP
COQ10B	NAP
COQ7	NAP
COQ6	COQ10D6
COQ4	COQ10DX
GENES ASOCIADOS A DEPLECIÓN DE ADN MITOCONDRIAL	
TYMP	MTDPS1 (MITOCHONDRIAL DNA DEPLETION SYNDROME-1)
TK2	MTDPS2
DGUOK	MTDPS3
POLG	MTDPS4A MTDPS4B
SUCLA2	MTDPS5
MPV17	MTDPS6

C10ORF2	MTDPS7
RRM2B	MTDPS8A MTDPS8B
SUCLG1	MTDPS9
AGK	MTDPS10
POLG2	PEOA4 (PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, AUTOSOMAL DOMINANT
SLC25A4	MTDPS12
FBXL4	MTDPS13
MGME1	MTDPS11
GENES ASOCIADOS A DEFICIENCIA COMBINADA DE FOSFORILACIÓN OXIDATIVA	
GFM1	COXPD1
MRPS16	COXPD2
TSFM	COXPD3
TUFM	COXPD4
MRPS22	COXPD5
AIFM1	COXPD6
C12ORF65	COXPD7
AARS2	COXPD8
GFER	COXPDX
FARS2	COXPD14
MRPL3	COXPD9
MRPL44	COXPD16
ELAC2	COXPD17
MTFMT	COXPD15
PNPT1	COXPD13
EARS2	COXPD12
RMND1	COXPD11
MTO1	COXPD10
LYRM4	COXPDX
CRIF1	COXPDY
OTROS	
ISCU	MIOPATIA MITOCONDRIAL
YARS2	MIOPATIA MITOCONDRIAL
TAZ	MIOPATIA MITOCONDRIAL
PUS1	MIOPATIA MITOCONDRIAL
SLC25A3	MIOPATIA MITOCONDRIAL: MITOCHONDRIAL PHOSPHATE CARRIER DEFICIENCY
DNA2	ADULT-ONSET INDIVIDUALS WITH A FORM OF MITOCHONDRIAL MYOPATHY FEATURING INSTABILITY OF MUSCLE
LETM1	CRUCIAL FOR THE MAINTENANCE OF MITOCHONDRIAL TUBULAR NETWORKS AND FOR THE ASSEMBLY OF THE
DNM1L	DEFECTIVE MITOCHONDRIAL AND PEROXISOMAL FISSION
NFU1	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 1
BOLA3	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 2
IBA57	MULTIPLE MITOCHONDRIAL DYSFUNCTIONS SYNDROME 3
ADCK1	NAP
ADCK2	NAP
ADCK4	NAP
ADCK5	NAP
OTRAS CONDICIONES QUE PUEDEN SOLAPARSE CON UNA DEFICIENCIA DE OXPHOS	
UPB1	DEFICIENCIA DE BETA-UREIDOPROPIONASA
NAT8L	DEFICIENCIA DE N-ACETILASPARTATO
DARS2	LEUKOENCEPHALOPATHY WITH BRAIN STEM AND SPINAL CORD INVOLVEMENT AND LACTATE ELEVATION
ADSL	ADENYLOSUCCINASE DEFICIENCY
DPYD	DIHYDROPYRIMIDINE DEHYDROGENASE DEFICIENCY
DPYS	DIHYDROPYRIMIDINURIA
TPK1	THIAMINE METABOLISM DYSFUNCTION SYNDROME 5 (EPISODIC ENCEPHALOPATHY TYPE)
RPIA	RIBOSE 5-PHOSPHATE ISOMERASE DEFICIENCY
SERAC1	3-METHYLGLUTACONIC ACIDURIA WITH DEAFNESS, ENCEPHALOPATHY, AND LEIGH-LIKE SYNDROME
SLC19A3	THIAMINE METABOLISM DYSFUNCTION SYNDROME 2 (BIOTIN- OR THIAMINE-RESPONSIVE ENCEPHALOPATHY TYPE 2)
GATM	CEREBRAL CREATINE DEFICIENCY SYNDROME 3
GAMT	CEREBRAL CREATINE DEFICIENCY SYNDROME 2
SLC6A8	CEREBRAL CREATINE DEFICIENCY SYNDROME
Nuevos genes asociados a defectos en la función mitocondrial	
MRPL3	Exome sequencing identifies MRPL3 mutation in mitochondrial cardiomyopathy.
MRPL44	A. Whole-exome sequencing identifies a mutation in the mitochondrial ribosome protein MRPL44 to underlie mitochondrial infantile cardiomyopathy.
ELAC2	ELAC2 mutations cause a mitochondrial RNA processing defect associated with hypertrophic
MTFMT	Mutations in MTFMT underlie a human disorder of formylation causing impaired mitochondrial
	Mutation in PNPT1 impairs RNA import into mitochondria and causes respiratory-chain deficiency
PNPT1	

EARS2	Leukoencephalopathy with thalamus and brainstem involvement and high lactate 'LTBL' caused by EARS2 mutations.
RMND1	Infantile encephaloneuromyopathy and defective mitochondrial translation are due to a homozygous RMND1 mutation
MTO1	Mutations of the mitochondrial-tRNA modifier MTO1 cause hypertrophic cardiomyopathy and lactic acidosis.
MICU1	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling
PET100	A Founder Mutation in PET100 Causes Isolated Complex IV Deficiency in Lebanese Individuals with Leigh Syndrome
OPA1	Regulate mitochondrial fusion
VARS2	VARS2 and TARS2 Mutations in Patients with Mitochondrial Encephalomyopathies
TARS2	
ETHE1	Encephalopathy, ethylmalonic
TRMU	Use of WES to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies
GARS	
FLAD1	
PTCD1	
CHCHD10	A mitochondrial origin for frontotemporal dementia and amyotrophic lateral sclerosis through CHCHD10 involvement
SPG7	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance.