

PANEL NMG8_1
MIOPATÍAS ESTRUCTURALES

CENTRAL CORE DISEASE OF MUSCLE	RYR1
DISTROFIA MUSCULAR DE DUCHENE	DMD
DISTROFIAS MUSCULARES DE CINTURAS (HERENCIA AUTOSOMICO RECESIVA) (LGMD2):	
LGMD2A	CAPN3
LGMD2B / Miyoshi muscular dystrophy-1	DYSF
LGMD2C	SGCG
LGMD2D	SGCA
LGMD2E	SGCB
LGDM2F	SGCD
LGMD2G	TCAP
LGMD2H	TRIM32
LGMD2J	TTN
LGMD2L / Miyoshi muscular dystrophy-3	ANO5
LGMD2Q	PLEC
LGMD2R	DES
LGMD2K /MDDGA1 /MDDGC1/ MDDGB1 / Síndrome de Walker-Warburg	POMT1
LGMD2M / MDDGA4 / Síndrome de Walker-Warburg	FKTN
MDDGA14/ Síndrome de Walker-Warburg	GMPPB
MDDGA11/ Síndrome de Walker-Warburg	B3GALNT2
LGMD2N / MDDGA2/ MDDGB2 / Síndrome de Walker-Warburg	POMT2
MDDGA6 / Síndrome de Walker-Warburg	LARGE
LGMD2I / MDDGA5/ Síndrome de Walker-Warburg	FKRP
MDDGA7 Síndrome de Walker-Warburg	ISPD
LGMD2O / MDDGA3/ MDDGB3 / Síndrome de Walker-Warburg	POMGNT1
MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, 7; MDDGC7	DAG1
CONGENITAL MUSCULAR DYSTROPHY AND HYPOGLYCOSYLATION OF A-DYSTROGLYCAN	B3GALNT2
DISTROFIAS MUSCULARES DE CINTURAS (HERENCIA AUTOSOMICO DOMINANTE) (LGMD1)	
LGMD1D/1E OR DISTAL-PREDOMINANT MYOPATHY	DNAJB6
LGMD1B	LMNA
LGMD1C	CAV3
MIOPATÍAS MIOFIBRILARES (MFM) (INCLUYE MIOPATIAS CON INCLUSIONES DE DESMINA (DRM))	
MYOPATHY, MYOFIBRILLAR, 1	DES
MYOPATHY, MYOFIBRILLAR, 4	LDB3
RIGID SPINE MUSCULAR DYSTROPHY 1; RSMD1	SEPN1
MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION; CFTD	
MYOPATHY, MYOFIBRILLAR ALPHA-B CRYSTALLIN-RELATED	CRYAB
MYOPATHY, MYOFIBRILLAR FATAL INFANTILE HYPERTROPHY, ALPHA-B CRYSTALLIN-RELATED	
MYOPATHY, MYOFIBRILLAR 6	BAG3
MIOPATIAS NEMALÍNICAS	
NEMALINE MYOPATHY 1: NEM1	TPM3
MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION; CFTD	
NEMALINE MYOPATHY 2 NEM2	NEB
NEMALINE MYOPATHY 3 NEM3	ACTA1
MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION; CFTD	
NEMALINE MYOPATHY 4 NEM4	TPM2
NEMALINE MYOPATHY, AMISH TYPE: NEM5	TNNT1
NEMALINE MYOPATHY 6 NEM6	KBTBD13
NEMALINE MYOPATHY 7 NEM7	CFL2
MIOPATÍAS CENTRONUCLEARES	
MYOTUBULAR MYOPATHY, X-LINKED	MTM1
MINICORE MYOPATHY, WITH EXTERNAL OPHTHALMOPLÉGIA	BIN1
CENTRONUCLEAR MYOPATHY, AUTOSOMAL, MODIFIER OF	MTMR14
MYOPATHY, CENTRONUCLEAR, 1	DNM2
	MYF6
DISTROFIA MUSCULAR DE EMERY-DREIFUSS (EDMD)	
EDMD1	EMD
EDMD2/3	LMNA
EDMD4	SYNE1
EDMD5	SYNE2
EDMD6	FHL1
MYOPATHY, X-LINKED, WITH POSTURAL MUSCLE ATROPHY; XMPMA	
MYOPATHY, REDUCING BODY, X-LINKED, CHILDHOOD-ONSET	
MYOPATHY, REDUCING BODY, X-LINKED, EARLY-ONSET, SEVERE	

MIOPATÍA DE ULLRICH/BETHLEM	
ULLRICH/BETHLEM MYOPATHY	COL6A1
	COL6A2
	COL6A3
MIOPATÍAS POR INCLUSIONES CORPUSCULARES	
INCLUSION BODY MYOPATHY -3 IBM3	MYH2
OTRAS MIOPATÍAS	
MUSCULAR DYSTROPHY, CONGENITAL, MEROSIN-DEFICIENT	LAMA2
MYOPATHY CONGENITAL, COMPTON-NORTH	CNTN1
MYOPATHY, DISTAL, 1; MPD1	MYH7
MYOPATHY MYOSIN STORAGE	
MUSCULAR DYSTROPHY DUE TO INTEGRIN ALPHA-7 DEFICIENCY	ITGA7
MUSCULAR DYSTROPHY, CONGENITAL, MEGACONIAL TYPE	CHKB
EARLY ONSET MYOPATHY, AREFLEXIA, RESPIRATORY DISTRESS AND DYSPHAGIA (EMARDD)	MEGF10
Mutations in GDP-mannose pyrophosphorylase B cause congenital and limb-girdle muscular dystrophies associated with hypoglycosylation of α -dystroglycan.	GMPPB
Mutations in the collagen XII gene define a new form of extracellular matrix-related myopathy.	COL12A1
Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in NemaLine Myopathy.	KLHL41
Exome sequencing identifies a novel SMCHD1 mutation in facioscapulohumeral muscular dystrophy 2.	SMCHD1
B3GALNT2 is a gene associated with congenital muscular dystrophy with brain malformations.	B3GALNT2
Mutations in the satellite cell gene MEGF10 cause a recessive congenital myopathy with minicores	MEGF10
Novel CLN3 mutation causing autophagic vacuolar myopathy	CLN3
Exome sequencing identifies a DNAJB6 mutation in a family with dominantly-inherited limb-girdle muscular dystrophy.	DNAJB6
A defect in the RNA-processing protein HNRNPD1 causes limb-girdle muscular dystrophy 1G (LGMD1G).	HNRNPD1
Mutation in TOR1AIP1 encoding LAP1B in a form of muscular dystrophy: a novel gene related to nuclear envelopathies	TOR1AIP1