

Panels used for analysis of Neuromuscular gene panel (Muscle v6)

Channelopathy

CACNA1S
CLCN1
KCNA1
KCNJ2
SCN4A

Congenital muscular dystrophy (CMD)

B3GALNT2	LAMA2
B4GAT1	LARGE1
CHKA	LMNA
CHKB	MSTO1
COL12A1	MTMR14
COL6A1	MYBPC3
COL6A2	PIGY
COL6A3	PLOD1
CRPPA	POMGNT1
DAG1	POMGNT2
DMD	POMK
DPM1	POMT1
DPM2	POMT2
DPM3	RXYLT1
FKRP	RYR1
FKTN	SELENON
GMPPB	SLC28A2
GOLGA2	TRAPPC11
ITGA7	TTN
ITGA9	

Congenital myasthenic syndrome (CMS)

AGRN	GMPPB
ALG14	LAMB2
ALG2	LRP4
CHAT	MUSK
CHD8	MYO9A
CHRNA1	NGLY1
CHRNB1	PREPL
CHRNA1	PYROXD1
CHRNA1	RAPSN
CHRNA1	SCN4A
COL13A1	SLC18A3
COLQ	SLC25A1
DOK7	SNAP25
DPAGT1	SYT2
GFPT1	

Distal arthrogryposis (DA)

ADCY6	CRPPA	KCNA1	NALCN	SELENON
ABCA7	DHCR24	KIAA1109	NEB	SEMA3A
ACTA1	DNM2	KIF5C	NEK9	SLC13A4
ADGRG6	DOK7	KY	NMNAT2	SLC18A3
ASB5	DST	L1CAM	NUP88	SLC35A3
ASCC1	DYNC1H1	LAMA2	NVL	SLC5A7
ASXL1	ECEL1	LAMA2	P4HA1	SLC6A9
B3GALT6	EGR2	LARGE1	PAX3	SMPD4
BAZ1B	ERBB3	LGIA	PFKM	SOX10
BICD2	ERCC5	LIFR	PI4KA	STAC3
BIN1	ERGIC1	LMNA	PIEZO2	SYNE1
CACNA1E	FBN2	LMX1B	PIGS	TGFB3
CACNA1S	FHL1	LOXL4	PIP5K1C	TNNI2
CHAT	FKBP10	MACF1	PLOD2	TNNT3
CHRNA1	FKRP	MAGEL2	PLXDC2	TOR1A
CHRNA1	FKTN	MAP2K1	PMP22	TPM2
CHRNA1	FLNA	MPZ	POMGNT2	TRIP4
CHRNA1	FLNB	MTM1	POMT1	TRPV4
CHRNA1	FOXP3	MUSK	POMT2	TTN
CNTN1	GBE1	MYBPC1	PRX	UBA1
CNTNAP1	GLDN	MYH2	RAPSN	UNC50
COL12A1	GLE1	MYH3	RYR1	UNC79
COL1A1	GPR153	MYH8	RYR3	UNC80
COL6A1	HS2ST1	MYLPF	SCN1A	ZBTB42
COL6A2	HSPG2	MYOD1	SCN4A	ZC4H2
COL6A3	KAT6B	MYOM2	SCYL2	

Glycogen storage disease (GSD) / Rhabdomyolysis

ACADM	LDHA
ACADS	LPIN1
ACADVL	MAGT1
AGL	MLIP
ASPH	OBSCN
CACNA1S	PCCA
CAV3	PCCB
COX6A2	PFKM
CPT2	PGAM2
ENO3	PGM1
ETFA	PHKA1
ETFB	PHKB
ETFDH	PYGM
FLAD1	RYR1
G6PC1	SLC25A32
GAA	SLC25A4
GBE1	TANGO2
GYG1	TBCK
GYS1	TMEM126B

Hypotonia

ASXL1
ASXL2
ASXL3
CACNA1E
DOCK3
EBF3
HECW2
IARS1
MICU2
MPZ
NKX6-2
PAX7
PAXBP1
PIGS
PLA2G6
PPP3CA
SH3TC2
SLC16A2
SLC25A46
SLC52A2
SLC52A3

Limb girdle muscular dystrophy (LGMD)

ANO5	FKRP	POMT2
ACTN2	FKTN	POPD2
B4GAT1	GAA	POPD3
BVES	GAPDH	PYROXD1
CACNA1S	GGPS1	SELENON
CAPN3	GMPPB	SGCA
CAV3	GYG1	SGCB
CHD7	HMGCS1	SGCD
COL12A1	HNRNPDL	SGCG
COL6A1	JAG2	SMCHD1
COL6A2	LARGE1	SYNE1
COL6A3	LIMS2	SYNE2
CRPPA	LMNA	TCAP
DAG1	LRIF1	TMEM43
DES	MYOT	TNPO3
DMD	PABPN1	TNXB
DNAJB6	PLEC	TOR1AIP1
DNMT3B	POGLUT1	TRAPPC11
DYNC1H1	POMGNT1	TRIM32
DYSF	POMGNT2	TRPV4
EMD	POMK	TTN
FHL1	POMT1	VCP

Myopathy

ABHD5	DNAJB4	IGHMBP2	MYH2	SLC22A5
ACTA1	DNM2	INPP5K	MYH7	SLC25A20
ACTN2	DST	ISCU	MYL1	SLC25A42
ADSS1	ETFA	ITGA7	MYL2	SMPX
AMPD1	ETFB	KBTBD13	MYMK	SPEG
ANO5	FDX2	KIF21A	MYO18B	SPTAN1
ASB5	FHL1	KIF5C	MYOT	SPTBN4
ASCC1	FHL3	KLHL24	MYPN	SQSTM1
ATP2A1	FKBP14	KLHL29	NEB	SRPK3
BAG3	FLAD1	KLHL40	OPA1	STAC3
BIN1	FLNB	KLHL41	ORA1	STIM1
CACNA1S	FLNC	KLHL9	P4HA1	SVIL
CASQ1	FOXP3	KY	PAX7	SYNC
CAVIN1	FXR1	LAMP2	PGK1	STIM1
CCDC78	GAA	LDB3	PHOX2A	TIA1
CFL2	GAPDH	LIFR	PIEZO2	TNNC2
CHST14	GNE	LMNA	PLCB1	TNNT1
CNTN1	GYG1	LMOD3	PNPLA2	TNNT3
COL12A1	HACD1	MAP3K20	POLG2	TPM2
COL6A1	HADHA	MB	PUS1	TPM3
COL6A2	HADHB	MEGF10	RBCK1	TTN
COL6A3	HNRNPA1	MICU1	RYR1	UNC45B
CRYAB	HNRNPA2B1	MSTN	RYR3	VAMP1
DCST2	HRAS	MTM1	SELENON	VCP
DES	HS2ST1	MYF6	SIL1	VMA21
DHCR24	HSPB8			

Nuclear mitochondrial

AARS2	MTFMT	RMND1
ACAD9	NDUFA1	RRM2B
AGK	NDUFA10	SCO2
AIFM1	NDUFA12	SDHA
ATP5F1A	NDUFA2	SDHAF1
BCS1L	NDUFA4	SLC19A3
C1QB	NDUFA9	SLC25A4
CEP89	NDUFAF1	SLC25A42
COX10	NDUFAF2	SUCLA2
COX14	NDUFAF6	SUCLG1
COX15	NDUFS1	SURF1
COX6A2	NDUFS2	TACO1
COX6B1	NDUFS3	TARS2
CYC1	NDUFS4	TK2
DGUOK	NDUFS7	TMEM126B
DNA2	NDUFS8	TRMU
FBXL4	NDUFV1	TSMF
FDX2	NFU1	TTC19
FOXRED1	OPA1	TWNK
LRPPRC	PDHA1	TYMP
MGME1	PET100	UQCRC2
MPV17	POLG	UQCRCQ
MRPL3	POLG2	VARS2

Visceral myopathy

ACTG2
LMOD1
MYH11
MYLK
SGOL1