
Table S1: Extended gene panel (n=185) used for NGS study

Gene list corresponding to the main clinical entry groups	Additional genes associated to other entry diagnostic categories included in the panel
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|---|---|
| - Nemaline Congenital myopathies | - Alpha-dystroglycanopathies |
| - Congenital myopathies excluding nemaline associated genes | - Metabolic myopathies |
| - Distal myopathies | - Myopathies associated with joint contractures |
| - Myofibrillar myopathies and inclusion body myopathies | - Congenital myasthenias |
| - Limb-Girdle Muscular Dystrophy | - Non-dystrophic myotonia |
| - Congenital Muscular Dystrophies except for alpha-dystroglycanopathies | - Periodic paralysis |
| | - Fetal and neonatal arthrogryposes |
| | - Other myopathies |

(official gene symbols approved by the HGNC) in red genes added during NGS panel update from 135 to 185 genes.

<i>ACAD9</i>	<i>ABHD5</i>
<i>ACTA1</i>	<i>ACADVL</i>
<i>ADSSL1</i>	<i>ADGRG1</i>
<i>ANO5</i>	<i>AGL</i>
<i>BAG3</i>	<i>AGRN</i>
<i>BIN1</i>	<i>ALG14</i>
<i>BVES</i>	<i>ALG13</i>
<i>CACNA1S</i>	<i>ALG2</i>
<i>CAPN3</i>	<i>AMPD1</i>
<i>CASQ1</i>	<i>ATP2A1</i>
<i>CAV3</i>	<i>B3GALNT2</i>
<i>CCDC78</i>	<i>B4GAT1</i>
<i>CFL2</i>	<i>CACNA1A</i>
<i>CHKB</i>	<i>CAVIN1</i>
<i>CNTN1</i>	<i>CHAT</i>
<i>COL12A1</i>	<i>CHRNA1</i>
<i>COL6A1</i>	<i>CHRNA1</i>
<i>COL6A2</i>	<i>CHRNA1</i>
<i>COL6A3</i>	<i>CHRNA1</i>
<i>CRYAB</i>	<i>CHRNA1</i>
<i>DAG1</i>	<i>CHRNA1</i>
<i>DES</i>	<i>CHRNA1</i>
<i>DMD</i>	<i>CHRNA1</i>
<i>DNAJB6</i>	<i>CHRNA1</i>
<i>DNM2</i>	<i>CHRNA1</i>

DPM3	CPT2
DYSF	DOK7
FLAD1	DOLK
FHL1	DPAGT1
FKRP	DPM1
FKTN	DPM2
FLNC	ECEL1
GAA	EMD
GMPPB	ENO3
GNE	ETFA
GOLGA2	ETFB
HACD1	ETFDH
HNRNPA1	FBN2
HNRNPDL	GBE1
ISPD	GFPT1
ITGA7	GYG1
KBTD13	GYS1
KLHL40	HSPG2
KLHL41	ISCU
KLHL9	KCNA1
KY	KCNE3
LAMA2	KCNJ18
LDB3	KCNJ2
LIMS2	LAMB2
LMNA	LAMP2
LMOD3	LDHA
MATR3	LARGE1
MEGF10	LARGE2
MTM1	LPIN1
MYBPC3	LRP4
MYH2	MUSK
MYH7	MYH3
MYO18B	MYH8
MYOT	MYO9A
MYPN	NALCN
NEB	NEK5
ORAI1	PABPN1
PLEC	PFKM
PNPLA2	PIEZO2
POMGNT1	PGAM2
POMT1	PGK1
POMT2	PGM1
PYROXD1	PHKA1
RYR1	PHKB

SELENON	PNPLA8
SGCA	POMGNT2
SGCB	POMK
SGCD	PREPL
SGCG	PRKAG2
SPEG	PYGM
SQSTM1	RAPSN
STAC3	RBCK1
STIM1	SCN4A
TCAP	SLC22A5
TIA1	SLC25A1
TNNT1	SLC25A20
TNPO3	SLC25A32
TOR1AIP1	SLC35A1
TPM2	SLC35A2
TPM3	SLC5A7
TRAPPC11	SNAP25
TRIM32	SYT2
TRIM54	TMEM5
TRIM63	TNNI2
TRIP4	TNNT3
TTN	TUBB2b
VCP	VMA21
	ZC4H2
