

**PANEL gR8C**  
**HIPOTONIA- TRASTORNOS DE LA NEURONA MOTORA**

SIGMAR1	AMYOTROPHIC LATERAL SCLEROSIS 16, JUVENILE
ALS2	AMYOTROPHIC LATERAL SCLEROSIS 2, JUVENILE
SETX	AMYOTROPHIC LATERAL SCLEROSIS 4, JUVENILE
SLC52A3	BROWN-VIALETTO-VAN LAERE SYNDROME 1/ FAZIO-LONDE DISEASE
SLC52A2	BROWN-VIALETTO-VAN LAERE SYNDROME 2
SLC52A1	BROWN-VIALETTO-VAN LAERE SYNDROME 3
IKBKAP	DYSAUTONOMIA, FAMILIAL (HIPOTONIA)
OCRL1	LOWE OCULOCEREBRORENAL SYNDROME (HIPOTONIA)
DES	SCAPULOPEONEAL SYNDROME, NEUROGENIC, KAESER TYPE
ASAH1	SPINAL MUSCULAR ATROPHY WITH PROGRESSIVE MYOCLONIC EPILEPSY
PLEKHG5	SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 4
DNAJB2	SPINAL MUSCULAR ATROPHY, DISTAL, AUTOSOMAL RECESSIVE, 5
REEP1	SPINAL MUSCULAR ATROPHY, DISTAL, TYPE VB; DSMAVB
ATP7A	SPINAL MUSCULAR ATROPHY, DISTAL, X-LINKED 3
UBA1	SPINAL MUSCULAR ATROPHY, X-LINKED 2, INFANTILE
SMN1	SPINAL MUSCULAR ATROPHY-1
SMN2	SPINAL MUSCULAR ATROPHY-1
DHTKD1	CHARCOT-MARIE-TOOTH DISEASE TYPE 2Q /2-AMINOADIPIC 2-OXOADIPIC ACIDURIA
DYNC1H1	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 20
HSPB1	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2F
GDAP1	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K
DNM2	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2M/ CHARCOT-MARIE-TOOTH DOMINANT
AARS	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2N
YARS	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE C
INF2	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE E
KARS	CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE, B
PMP22	CHARCOT-MARIE-TOOTH DISEASE, TYPE 1A/ NEUROPATHY, RECURRENT, WITH PRESSURE
LITAF	CHARCOT-MARIE-TOOTH DISEASE, TYPE 1C
KIF1B	CHARCOT-MARIE-TOOTH DISEASE, TYPE 2A1
MFN2	CHARCOT-MARIE-TOOTH DISEASE, TYPE 2A2
RAB7	CHARCOT-MARIE-TOOTH DISEASE, TYPE 2B
MED25	CHARCOT-MARIE-TOOTH DISEASE, TYPE 2B2
NEFL	CHARCOT-MARIE-TOOTH DISEASE, TYPE 2E/ CHARCOT-MARIE-TOOTH NEUROPATHY, TYPE
MTMR2	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B1
SBF2	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2
SH3TC2	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4C
NDRG1	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4D
PRX	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4F
FGD4	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4H
FIG4	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4J
PRPS1	CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 5
GJB1	CHARCOT-MARIE-TOOTH NEUROPATHY, X-LINKED DOMINANT, 1
LRSAM1	CHARCOT-MARIE-TOOTHE DISEASE, AXONAL, TYPE 2P
TFG	HEREDITARY MOTOR AND SENSORY NEUROPATHY, PROXIMAL TYPE
TRPV4	HEREDITARY MOTOR AND SENSORY NEUROPATHY, TYPE IIC/CHARCOT-MARIE-TOOTH
IGHMBP2	NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE VI
EGR2	NEUROPATHY, CONGENITAL HYPOMYELINATING/ CHARCOT-MARIE-TOOTH DISEASE, TYPE
MPZ	NEUROPATHY, CONGENITAL HYPOMYELINATING/ CHARCOT-MARIE-TOOTH DISEASE, TYPE
HSPB8	NEUROPATHY, DISTAL HEREDITARY MOTOR, TYPE IIA/ CHARCOT-MARIE-TOOTH DISEASE,
BSCL2	NEUROPATHY, DISTAL HEREDITARY MOTOR, TYPE V/ CHARCOT-MARIE-TOOTH DISEASE
GARS	NEUROPATHY, DISTAL HEREDITARY MOTOR, TYPE V/CHARCOT-MARIE-TOOTH DISEASE,
VCP	ATYPICAL AMYOTROPHIC LATERAL SCLEROSIS.