Gen-Panels Neuromuskuläre Erkrankungen | LI

Hinweise:

- Gen-Panels mit mehr als 10 Genen dürfen gemäss Analysenliste des BAG nur durch Ärzte mit einem FMH-Titel Medizinische Genetik verordnet werden.
- Die Gen-Panels sind nicht endgültig und können je nach Fragestellung auf Wunsch angepasst werden.
- Die Gen-Panels können abhängig von der aktuellen Datenlage zum Zeitpunkt der Analysen von denjenigen auf dieser Liste abweichen.

Neuromuskuläre Erkrankungen, umfassendes Panel v13 (493 Gene)

AARS1, ABCA1, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACOX1, ACTA1, ACTN2, ACVR1, ADCY6, ADGRG6, ADSS1, AGL, AGRN, AGTPBP1, AGXT, AHCY, AIFM1, ALDOA, ALG13, ALG14, ALG2, ALG3, ALS2, AMPD1, ANO5, AP1S1, APOA1, APOO, APTX, AR, ARHGEF10, ARSA, ASAH1, ASCC1, ASCC3, ATL1, ATL3, ATM, ATP1A1, ATP2A1, ATP7A, B3GALNT2, B3GNT2, B4GALNT1, B4GAT1, BAG3, BCKDHB, BET1, BICD2, BIN1, BSCL2, BVES, CACNA1S, CADM3, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCT5, CD59, CFAP276, CFL2, CHAT, CHCHD10, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, CNTNAP1, COA7, COL12A1, COL13A1, COL4A1, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ8A, COX20, COX6A1, CPOX, CPT2, CRPPA, CRYAB, CTDP1, CYP27A1, DAG1, DARS2, DCAF8, DCTN1, DCTN2, DEGS1, DES, DGAT2, DGUOK, DHTKD1, DHX16, DMD, DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC3, DNM2, DNMT1, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DTNA, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENDOG, ENO3, EPG5, ERBB3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAH, FAM126A, FBLN5, FBN2, FBXO38, FDX2, FGD4, FHL1, FHL2, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNC, FLVCR1, FMR1, FXR1, G6PC1, GAA, GALC, GAN, GARS1, GBA2, GBE1, GBF1, GDAP1, GFER, GFPT1, GGPS1, GIPC1, GJB1, GJB3, GJC2, GLA, GLDN, GLE1, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GSN, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HNRNPA1, HNRNPA2B1, HNRNPDL, HOXD10, HSPB1, HSPB3, HSPB8, HSPG2, IARS2, IGHMBP2, INF2, INPP5K, ISCU, ITGA7, ITPR3, JAG1, JAG2, JPH1, KARS1, KBTBD13, KCNA1, KCNA2, KCNE3, KCNJ2, KCNJ5, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRP12, LRP4, LRSAM1, LYST, MAG, MAP3K20, MARS1, MATR3, MCM3AP, MCOLN1, MED25, MEGF10, MFN2, MICU1, MLIP, MMACHC, MME, MORC2, MPV17, MPZ, MSTN, MSTO1, MT-ATP6, MTM1, MTMR14, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, MUSK, MYBPC1, MYBPC3, MYF5, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYO9B, MYOD1, MYOT, MYPN, NAGA, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK9, NEMF, NGF, NOP56, NTRK1, NUDT2, OPA1, OPA3, ORAI1, PABPN1, PAX7, PDHA1, PDK3, PDYN, PEX10, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PHYH, PIEZO2, PIGB, PIP5K1C, PLEC, PLEKHG5, PLIN4, PLOD2, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, PNPLA8, POGLUT1, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PPOX, PPP2R2B, PRDM12, PREPL, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PSAT1, PTEN, PTPN11, PTRH2, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RILPL1, RRM2B, RXYLT1, RYR1, RYR3, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SCYL1, SELENON, SEMA3A, SEPTIN9, SETX, SGCA, SGCB, SGCD, SGCG, SGPL1, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A19, SLC25A20, SLC25A26, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SLC9A3R1, SMCHD1, SMN1, SMPX, SNAP25, SORD, SOX10, SPAST, SPEG, SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SQSTM1, STAC3, STIM1, SUCLA2, SURF1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TANGO2, TCAP, TDP1, TECPR2, TFG, TIA1, TK2, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM2, TRIM32, TRIM63, TRIP4, TRPA1, TRPV4, TSEN54, TSFM, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, UNC13A, UNC45B, VAMP1, VAPB, VCP, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, YARS2, ZC4H2, ZFYVE26

Dystrophien v10 (82 Gene)

ACTA1, ALG13, ANO5, B3GALNT2, B4GAT1, BET1, BVES, CAPN3, CAV3, CAVIN1, CHKB, COL12A1, COL4A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DMD, DNAJB6, DNM2, DNMT3B, DOLK, DPM1, DPM2, DPM3, DTNA, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GGPS1, GMPPB, GNE, GOLGA2, GOSR2, HNRNPA2B1, HNRNPDL, INPP5K, ITGA7, JAG2, LAMA2, LARGE1, LIMS2, LMNA, LRIF1, MICU1, MSTO1, MYMK, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PYROXD1, RXYLT1, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TRIP4, TTN, VCP

Maligne Hyperthermie v1 (3 Gene)

CACNA1S, RYR1, STAC3

Metabolische Myopathien v3 (75 Gene)

ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, AGL, AHCY, ALDOA, AMPD1, ANO5, CACNA1S, CAV3, COQ2, COQ8A, CPT2, DMD, DYSF, ENO3, ETFA, ETFB, ETFDH, FDX2, FKRP, FKTN, FLAD1, G6PC1, GAA, GBE1, GMPPB, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, LAMP2, LDHA, LPIN1, MYH3, OPA1, OPA3, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PNPLA2, PNPLA8, POLG, POLG2, PRKAG2, PUS1, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SIL1, SLC16A1, SLC22A5, SLC25A20, SUCLA2, TAFAZZIN, TANGO2, TK2, TSEN54, TSFM, TWNK, TYMP, YARS2

Muskelatrophien v7 (44 Gene)

AARS1, ABHD12, AIFM1, ASAH1, ASCC1, ATL1, ATP7A, BICD2, BSCL2, CCT5, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, KIF1A, KIF1B, LAS1L, LITAF, MPZ, MTRFR, NEFL, PLEKHG5, PMP22, PRX, RAB7A, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, TRIP4, TRPV4, UBA1, VAPB, VRK1, YARS1

Myofibrilläre Myopathie v2 (13 Gene)

BAG3, CRYAB, DES, DNAJB6, FHL1, FLNC, KY, LDB3, MYOT, PYROXD1, SVIL, TTN, UNC45B

Myopathien v13 (277 Gene)

AARS1, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ACVR1, ADSS1, AGL, AHCY, AIFM1, ALDOA, ALG13, AMPD1, ANO5, APOO, ASAH1, ASCC1, ASCC3, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BSCL2, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CCT5, CFL2, CHCHD10, CHKB, CLCN1, CNTN1, COL12A1, COL4A1, COL6A1, COL6A2, COL6A3, COQ2, COQ8A, CPT2, CRPPA, CRYAB, DAG1, DCTN1, DES, DHX16, DMD, DNA2, DNAJB2, DNAJB4, DNAJB6, DNM2, DNMT3B, DOK7, DOLK, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EMD, ENDOG, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBXO38, FDX2, FHL1, FHL2, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, G6PC1, GAA, GARS1, GBE1, GFER, GGPS1, GIPC1, GMPPB, GNE, GOLGA2, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KCNA1, KIF1A, KIF1B, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRP12, MAP3K20, MATR3, MCOLN1, MEGF10, MICU1, MLIP, MPZ, MSTN, MSTO1, MTM1, MTMR14, MTRFR, MYBPC1, MYBPC3, MYF5, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYOD1, MYOT, MYPN, NEB, NEFL, OPA1, OPA3, ORAI1, PABPN1, PAX7, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PIEZO2, PLEC, PLEKHG5, PLIN4, PMP22, PNPLA2, PNPLA6, PNPLA8, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PRKAG2, PRX, PUS1, PYGM, PYROXD1, RAB7A, RBCK1, REEP1, RILPL1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SELENON, SETX, SGCA, SGCB, SGCD, SGCG, SIGMAR1, SIL1, SLC16A1, SLC22A5, SLC25A20, SLC25A26, SLC25A4, SLC25A42, SLC5A7, SMCHD1, SMN1, SMPX, SPEG, SQSTM1, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYNE2, TAFAZZIN, TANGO2, TCAP, TIA1, TK2, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIM63, TRIP4, TRPV4, TSEN54, TSFM, TTN, TWNK, TYMP, UBA1, UNC45B, VAPB, VCP, VMA21, VRK1, YARS1, YARS2

Myotonie v6 (6 Gene)

ATP2A1, CAV3, CLCN1, HINT1, HSPG2, KCNA1, SCN4A

Myasthene Syndrome/Myasthenie v7 (30 Gene)

AGRN, ALG14, ALG2, CHAT, CHD8, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, TOR1AIP1, VAMP1

Nemalin-Myopathie v1 (12 Gene)

ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYO18B, MYPN, NEB, TNNT1, TPM2, TPM3

Neuropathien v11 (216 Gene)

AARS1, ABCA1, ABHD12, ACOX1, AGTPBP1, AGXT, AIFM1, AP1S1, APOA1, APTX, AR, ARHGEF10, ARSA, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CADM3, CCT5, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX20, COX6A1, CPOX, CTDP1, CYP27A1, DARS2, DCAF8, DCTN1, DEGS1, DGUOK, DHTKD1, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, ETFDH, FAH, FAM126A, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FMR1, GALC, GAN, GARS1, GBA2, GBF1, GDAP1, GJB1, GJC2, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IARS2, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KCNA2, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, LYST, MAG, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MTMR2, MTRFR, MT-RNR1, MT-TL1, MTTP, MYH14, MYO9B, NAGA, NAGLU, NDRG1, NEFH, NEFL, NEMF, NGF, NOP56, NTRK1, NUDT2, OPA1, OPA3, PDHA1, PDK3, PDYN, PEX10, PEX7, PHYH, PIGB, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PPOX, PPP2R2B, PRDM12, PRKCG, PRNP, PRPS1, PRX, PSAT1, PTEN, PTPN11, PTRH2, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SCYL1, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SMN1, SORD, SOX10, SPAST, SPG11, SPG7, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TDP1, TECPR2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VAPB, VCP, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, XRCC1, YARS1, ZFYVE26

Periodische Paralyse v2 (3 Gene)

CACNA1S, KCNJ2, SCN4A

Small-Fibre-Neuropathie v1 (5 Gene)

GLA, SCN4A, SCN9A, SCN10A, SCN11A