

## NEUROMUSKULÁRNÍ ONEMOCNĚNÍ – seznam analyzovaných genů

1. Svalové dystrofie: *ANO5, BVES, CAPN3, CAV3, CAVIN1, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, HNRNPDL, ISPD, KBTBD13, LAMA2, LIMS2, LMNA, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMT1, POMT2, PYROXD1, SGCA, SGCB, SGCD, SGCG, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VCP*
2. Kongenitální svalové dystrofie: *ACTA1, ALG13, B3GALNT2, B4GAT1, COL6A1, COL6A2, COL6A3, COL12A1, DAG1, DNM2, DPM1, DPM2, FKRP, FKTN, GMPPB, GOLGA2, GOSR2, CHKB, INPP5K, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MSTO1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, RYR1, SELENON, TCAP, TRAPPC11, TRIP4*
3. Kongenitální myopatie: *ACTA1, ACTN2, BIN1, CACNA1H, CACNA1S, CCDC78, CFL2, CNTN1, DNM2, FXR1, HACD1, HNRNPA1, HRAS, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MTM1, MYBPC3, MYH2, MYH7, MYL1, MYMK, MYO18B, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SELENON, SPEG, SPTBN4, STAC3, TNNT1, TNPO3, TPM2, TPM3, TRIM32, TTN, ZAK*
4. Distální myopatie: *ACTN2, ADSSL1, ANO5, CAV3, DNAJB6, DNM2, DYSF, FLNC, GNE, HSPB8, KLHL9, LDB3, LRP12, MATR3, MYH7, MYOT, NEB, SQSTM1, TIA1, TTN, VCP*
5. Další myopatie: *ACVR1, BAG3, CASQ1, CAV3, CLN3, CRYAB, DES, FHL1, FLNC, HNRNPA2B1, ISCU, KY, LAMP2, LDB3, MB, MSTN, MYOT, ORAI1, PABPN1, PLEC, PYROXD1, RYR1, SELENON, STIM1, TRIM54, TRIM63, TTN, VCP, VMA21*
6. Myotonické syndromy: *ATP2A1, CAV3, CLCN1, HSPG2*
7. Svalová onemocnění spojená s iontovými kanály: *CACNA1A, CACNA1S, CLCN1, KCNA1, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ18, KCNJ2, KCNQ1, SCN4A, SCN5A*
8. Maligní hypertermie: *CACNA1S, RYR1, BCHE*
9. Kongenitální myastenické syndromy: *AGRN, ALG14, ALG2, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, LAMA5, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, RPH3A, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, UNC13A, VAMP1*
10. Onemocnění motorického neuronu: *AARS, ALS2, ANG, ANXA11, AR, ASAH1, ASCC1, ATP7A, ATXN2, BICD2, BSCL2, C9orf72, DCTN1, DNAJB2, DYNC1H1, ERBB3, ERBB4, EXOSC3, EXOSC8, FBXO38, FIG4, FUS, GARS, GLE1, HEXB, HNRNPA1, HSPB1, HSPB3, HSPB8, CHCHD10, CHMP2B, IGHMBP2, KIF26B, KIF5A, MAPT, MATR3, MYH14, NEFH, NEK1, OPTN, PFN1, PIP5K1C, PLEKHG5, PRPH, PRUNE1, RBM7, REEP1, SETX, SIGMAR1, SLC52A2, SLC52A3, SLC5A7, SOD1, SPG11, SQSTM1, SYT2, TARDBP, TBK1, TRIP4, TRPV4, TUBA4A, UBA1, UBQLN2, VAPB, VCP, VRK1*
11. Další neuromuskulární onemocnění: *COL25A1, DGUOK, ELP1, FDX2, CHCHD10, KIF21A, MGME1, MYH3, MYH8, OPA1, PHOX2A, POLG, POLG2, PUS1, RNASEH1, RRM2B, SGCE, SLC25A4, SUCLA2, SYNE1, TK2, TNNI2, TNNT3, TOR1A, TPM2, TTR, TUBB3, TWNK, YARS2*
12. Dědičné kardiomyopatie: *AARS2, ABCC9, ACTC1, ACTN2, AKAP9, ALPK3, ANK2, ANKRD1, BAG3, CACNA1C, CACNB2, CALM1, CALM2, CALR3, CASQ2, CAV3, COX15, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, EYA4, FKTN, FLNA, FLNC, GAA, GATAD1, GJA5, GPD1L, HCN4, ILK, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, LAMA4, LDB3, LMNA, MIB1, MRPL3, MRPL44, MTO1, MURC, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, NDUFAF1, NEXN, NPPA, NUP155, PKP2, PLN, PRDM16, PRKAG2, PSEN2, RAF1, RBM20, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SGCD, SNTA1, SYNE1, TAZ, TCAP, TECRL, TGFB3, TMEM43, TMPO,*

*TNNC1, TNNI3, TNNT2, TPM1, TRDN, TSFM, TTN, VCL*; geny mimo [www.musclegenetable.fr](http://www.musclegenetable.fr): *CALM3, CTNNA3, DNAJC19, EMD, FHL1, CHD7, LAMP2, PDLIM3, RANGRF, TRPM4, ZIC3*

13. Další geny (mimo [www.musclegenetable.fr](http://www.musclegenetable.fr)): svalové dystrofie a myopatie: *AMPD1, DNMT3B, GFER, GLA, CHD7, MICU1, MTMR14, MYF6, POPDC2, POPDC3, RBCK1, SIL1, SUN1, SUN2, TNNC2, UNC45B*; myastenické syndromy: *AK9, NGLY1*; onemocnění motorického neuronu: *CCNF, DNAH10, EXOSC9, SPG7*; Ehlers-Danlosův syndrom (ve spojení s myopatií): *COL1A1, COL3A1, COL5A1, COL5A2, DSE, FKBP14, PLOD1, TNXB*
14. Artrogrypóza: *ADCY6, ADGRG6, ALG3, CACNA1E, CNTNAP1, DHCR24, DNM2, ECEL1, ERBB3, ERCC1, ERCC5, ERCC6, ERGIC1, FBN1, FBN2, FBN3, FKBP10, GLE1, GLDN, CHST14, KAT6B, KIAA1109, KIF26B, KIF5C, LGI4, MAGEL2, MUSK, MYBPC1, MYBPC2, MYH3, MYH8, MYOD1, NALCN, NEK9, NUP88, PI4KA, PIEZO2, PIP5K1C, PSD3, SLC35A3, SMPD4, SYNE1, TNNI2, TNNT3, TPM2, TUBB2B, UNC50, VIPAS39, VPS33B, VPS8, ZBTB42, ZC4H2*