

## Muskuläre Erkrankungen

**Amyotrophe Lateralsklerose (ALS) [GP122XL]**

1. SOD1, C9orf72, 2. ALS2, ANG, CHMP2B, DCTN1, ERBB4, FIG4, FUS, MATR3, NEFH, OPTN, PFN1, PRPH, SETX, SQSTM1, TARDBP, UBQLN2, VAPB, VCP, VEGFA

**Emery-Dreyfuss-Muskeldystrophie [GP123XL]**

EMD, FHL1, LMNA, SYNE1, SYNE2

**Gliedergürtelmuskeldystrophie (LGMD) [GP124] (~16 kb)**

1. SGCA, SGCB, SGCG, SGCD, 2. ANO5, CAPN3, DYSF, FKRP

**Muskeldystrophie [GP125XL]**

ANO5, B3GNT1 (=B4GAT1), CAPN3, CHKB, COL6A1, COL6A2, COL6A3, DMD, DYSF, EMD, FHL1, FKRP, FKTN, ISPD, LAMA2, LARGE, LMNA, PABPN1, POMGNT1, POMT1, POMT2, SEPN1, SGCA, SGCB, SGCD, SGCG, SYNE1, SYNE2, TCAP, TTN

**Walker-Warburg-Syndrom [GP133] (~16kb)**

B3GNT1 (=B4GAT1), FKRP, FKTN, ISPD, LARGE, POMGNT1, POMT1, POMT2, TCAP, TRIM32

**Myasthenie-Syndrome, kongenitale [GP126] (~20 kb)**

1. CHRNE, 2. COLQ, DOK7, RAPSN, CHAT, GFPT1, 3. AGRN, CHRNA1, CHRNB1, CHRND

**umfassende Diagnostik [GP126XL]**

AGRN, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, FBN2, GFPT1, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, TPM2

**Myopathie, myofibrilläre (MFM) [GP128] (~16 kb)**

DES, CRYAB, MYOT, LDB3, FLNC, BAG3, FHL1, DNAJB6

**Myopathie, nemaline (NM) [GP129XL]**

1. NEB (25 kb), ACTA1, 2. TPM3, TPM2, TNNT1, CFL2

**Myopathie, umfassende Diagnostik [GP130XL]**

ACTA1, ACVR1, ANO5, BAG3, BIN1, C10orf2, CAV3, CFL2, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, ISCU, KBTBD13, KLHL9, LAMP2, LDB3, MAMLD1, MATR3, MEGF10, MSTN, MTM1, MTMR14, MYF6, MYH14, MYH2, MYH7, MYOT, NEB, OPA1, ORAI1, PABPN1, PLEC, POLG, POLG2, PUS1, RRM2B, RYR1, SEPN1, SIL1, STIM1, SUCLA2, TK2, TNNT1, TPM2, TPM3, TRIM32, TTN, VCP, VMA21, YARS2