**پنل بیماری های عصبی عضلانی**

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| **ردیف** | **نام بیماری** | **تعداد ژن** | **ژن های مورد بررسی** |
| 1 | Spinal Muscular Atrophy | 31 | SMN1 deletion analysis can be deselected if not required.AR-repeat analysis optional.AARS, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, RBM7, REEP1, SCO2, SETX, SIGMAR1, SLC5A7, TRPV4, UBA1, VAPB, VRK1 |
| 2 | Hereditary Neuropathies | 101 | PMP22 deletion analysis can be deselected if not requiredAARS, ABHD12, AIFM1, ARHGEF10, ATL1, ATL3, BSCL2, C10ORF2, C12ORF65, CCT5, COX6A1, CTDP1, DCAF8, DCTN2, DGAT2, DHTKD1, DNAJB2, DNAJB5, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GJB3, GNB4, HADHA, HADHB, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PLEKHG5, PMP2, PMP22, POLG, PRDM12, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SEPT9, SH3TC2, SLC12A6, SOX10, SPG11, SPTLC1, SPTLC2, SURF1, TECPR2, TFG, TRIM2, TRPV4, TTR, TYMP, VCP, WNK1, YARS, ZNF106 |
| 3 | Congenital and Distal Myopathies | 82 | ACTA1, ACVR1, ADSSL1, ANO5, BAG3, BIN1, C10ORF2, CACNA1S, CASQ1, CAV3, CCDC78, CFL2, CHCHD10, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNA2, DNAJB5, DNM2, DYSF, FHL1, FHL2, FKBP14, FLNC, GNE, HACD1 (PTPLA), HNRNPA1, HNRNPA2B1, ISCU, KBTBD13, KLHL40, KLHL41, KLHL9, KY, LAMP2, LDB3, LMOD3, MATR3, MEGF10, MICU1, MSTN, MTM1, MTMR14, MYF6, MYH14, MYH2, MYH7, MYOT, NEB, OPA1, ORAI1, PABPN1, PLEC, POLG, POLG2, PUS1, PYROXD1, RRM2B, RYR1, SEPN1, SIL1, SPEG, STAC3, STIM1, SUCLA2, TIA1, TK2, TNNT1, TPM2, TPM3, TRIM32, TRIM54, TRIM63, TTN, VCP, VMA21, YARS2, ZAK |
| 4 | Limb- Girdle Muscular Dystrophies | 39 | ANO5, BVES, CAPN3, CAV3, DAG1, DES, DMD, DNAJB6, DPM3, DYSF, FKRP, FKTN, FLNC, GAA, GMPPB, GNE, HNRNPDL, ISPD, LAMA2, LIMS2, LMNA, MYOT, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN |
| 5 | Muscular Dystrophies | 42 | DMD deletion analysis can be deselected if not requiredANO5, B3GALNT2, B4GAT1 (B3GNT1), CHKB, COL12A1, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, GOLGA2, ISPD, ITGA7, LAMA2, LARGE, LMNA, PABPN1, POMGNT1, POMGNT2, POMT1, POMT2, PTRF, SEPN1, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TMEM5, TOR1AIP1, TRAPPC11, TRIP4, TTN |
| 6 | Congenital Myasthenic Syndromes and Arthrogryposis | 58 | ACTA1, ADCY6, AGRN, ALG14, ALG2, ALG3, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CNTNAP1, COL13A1, COLQ, DNM2, DOK7, DPAGT1, ECEL1, ERBB3, FBN2, FBN3, FKBP10, GFPT1, GLDN, GLE1, GMPPB, GPR126, LAMB2, LRP4, MUSK, MYBPC1, MYH3, MYH8, MYO9A, NALCN, NEK9, PIEZO2, PIP5K1C, PLEC, PLOD2, PREPL, RAPSN, SCN4A, SEMA3A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYNE1, SYT2, TNNI2, TNNT3, TPM2, VIPAS39, VPS33B, ZC4H2 |
| 7 | Myotonia | 5 | ATP2A1, CAV3, CLCN1, HINT1, SCN4A |
| 8 | Metabolic Myopathies | 43 | ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, AGL, AMPD1, CPT2, ENO3, ETFA, ETFB, ETFDH, G6PC, GAA, GBE1, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, LDHA, LPIN1, PDHA1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PNPLA2, POLG2, PUS1, PYGM, RBCK1, RRM2B, SLC16A1, SLC22A5, SLC25A20, TAZ, YARS2 |
| 9 | Walker- Warburg Syndrome | 15 | B3GALNT2, B4GAT1 (B3GNT1), COL4A1, DAG1, FKRP, FKTN, GMPPB, ISPD, LARGE, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5 |
| 10 | Periodic Paralyses | 5 | CACNA1S, SCN4A, KCNJ2, KCNJ5, KCNE3 |
| 11 | Complete Panel- Neuromuscular Disease | 331 | AARS, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACVR1, ADCY6, ADSSL1, AGL, AGRN, AIFM1, ALG14, ALG2, ALG3, AMPD1, ANO5, ARHGEF10, ASAH1, ASCC1, ATL1, ATL3, ATP2A1, ATP7A, B3GALNT2, B3GNT1, BAG3, BICD2, BIN1, BSCL2, BVES, C10ORF2, C12ORF65, CACNA1S, CAPN3, CASQ1, CAV3, CCDC78, CCT5, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, CNTNAP1, COL12A1, COL13A1, COL4A1, COL6A1, COL6A2, COL6A3, COLQ, COX6A1, CPT2, CRYAB, CTDP1, DAG1, DCAF8, DCTN1, DCTN2, DES, DGAT2, DHTKD1, DMD, DNA2, DNAJB2, DNAJB5, DNAJB6, DNM2, DNMT1, DOK7, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, ERBB3, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM134B, FBLN5, FBN2, FBN3, FBXO38, FGD4, FHL1, FHL2, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNC, G6PC, GAA, GAN, GARS, GBE1, GDAP1, GFPT1, GJB1, GJB3, GLDN, GLE1, GMPPB, GNB4, GNE, GOLGA2, GPR126, GYG1, GYS1, HADH, HADHA, HADHB, HARS, HEXA, HINT1, HK1, HNRNPA1, HNRNPA2B1, HNRNPDL, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, ISCU, ISPD, ITGA7, KARS, KBTBD13, KCNE3, KCNJ2, KCNJ5, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMB2, LAMP2, LARGE, LAS1L, LDB3, LDHA, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRP4, LRSAM1, MARS, MATR3, MED25, MEGF10, MFN2, MICU1, MME, MORC2, MPV17, MPZ, MSTN, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYO9A, MYOT, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK9, NGF, NTRK1, OPA1, ORAI1, PABPN1, PDHA1, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG2, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD2, PMP2, PMP22, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM12, PREPL, PRPS1, PRX, PTPLA, PTRF, PUS1, PYGM, PYROXD1, RAB7A, RAPSN, RBCK1, RBM7, REEP1, RRM2B, RYR1, SBF1, SBF2, SCN10A, SCN11A, SCN4A, SCN9A, SCO2, SEMA3A, SEPN1, SEPT9, SETX, SGCA, SGCB, SGCD, SGCG, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC5A7, SMCHD1, SNAP25, SOX10, SPEG, SPG11, SPTLC1, SPTLC2, STAC3, STIM1, SUCLA2, SURF1, SYNE1, SYNE2, SYT2, TAZ, TCAP, TECPR2, TFG, TIA1, TK2, TMEM43, TMEM5, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIM54, TRIM63, TRIP4, TRPV4, TTN, TTR, TYMP, UBA1, VAPB, VCP, VIPAS39, VMA21, VPS33B, VRK1, WNK1, YARS, YARS2, ZAK, ZC4H2, ZNF106 |