

## Panels used for analysis of Neuromuscular gene panel (Neuro v6)

| Ataxia  |         |         |         |
|---------|---------|---------|---------|
| ABCB7   | DOCK3   | MTPAP   | SLC1A3  |
| ABHD12  | EBF3    | MYO19   | SLC9A1  |
| ADPRS   | EEF2    | NGLY1   | SNX14   |
| AFG3L2  | ELOVL4  | NKX6-2  | SPG7    |
| ANO10   | ELOVL5  | NOP56   | SPTBN2  |
| APTX    | EP300   | NOVA2   | STUB1   |
| ATCAY   | FAT1    | NUBPL   | SVBP    |
| ATM     | FAT2    | OPA1    | SYNE1   |
| ATP1A3  | FGF14   | OTUD4   | SYT14   |
| ATP2B3  | FLVCR1  | PDHA1   | TBCE    |
| ATP8A2  | FXN     | PDYN    | TBP     |
| BEAN1   | GALC    | PIGS    | TDP1    |
| BRAT1   | GDAP2   | PIK3R5  | TDP2    |
| CA8     | GOSR2   | PLD3    | TGM6    |
| CACNA1A | GRID2   | PMPCA   | THG1L   |
| CACNA1G | GRM1    | PNKP    | TMEM240 |
| CACNB4  | GRN     | POLG    | TOMM70  |
| CAMTA1  | HEXA    | POLR3A  | TPP1    |
| CCDC88C | HEXB    | PPP2R2B | TSFM    |
| CHP1    | IRF2BPL | PRKCG   | TTBK2   |
| CLCN2   | ITPR1   | PUM1    | TTPA    |
| CLN6    | KCNA1   | RNF170  | VAMP1   |
| CLPP    | KCNC3   | RNF216  | VLDR    |
| COA7    | KCND3   | RUBCN   | VPS13D  |
| COQ5    | KIF1C   | SACS    | WDR81   |
| COQ8A   | KIF26B  | SCYL1   | WVOX    |
| CWF19L1 | MME     | SETX    | XRCC1   |
| CYP27A1 | MMP8    | SIL1    |         |
| DNMT1   | MRE11A  | SLC17A5 |         |

| CMT      |         |         |         |          |
|----------|---------|---------|---------|----------|
| AARS1    | DST     | HSPB1   | NEFL    | SLC25A46 |
| ABHD12   | DYNC1H1 | HSPB3   | NEMF    | SLC5A7   |
| ADPRS    | EGR2    | HSPB8   | NGF     | SORD     |
| AIFM1    | ELP1    | IGHMBP2 | NTRK1   | SOX10    |
| ARHGEF10 | FBLN5   | INF2    | PKD3    | SPG11    |
| ATL1     | FBXO38  | KARS1   | PDXK    | SPTAN1   |
| ATL3     | FOX2    | KIF1A   | PLEKHG5 | SPTBN4   |
| ATP1A1   | FGD4    | KIF1B   | PMP22   | SPTLC1   |
| BSCL2    | FIG4    | KIF5A   | PMP22   | SPTLC2   |
| C10RF194 | FLVCR1  | LITAF   | PRDM12  | SPTLC3   |
| CAPRN1   | GALC    | LMNA    | PRPS1   | SURF1    |
| CCT5     | GAN     | LRSAM1  | PRX     | SYT2     |
| CD59     | GARS1   | MARS1   | RAB7A   | TFG      |
| COA7     | GBE1    | MCM3AP  | REEP1   | TRIM2    |
| COX6A1   | GBF1    | MED25   | RETREG1 | TRPA1    |
| CTDP1    | GDAP1   | MFN2    | SBF1    | TRPV4    |
| DCAF8    | GJB1    | MME     | SBF2    | TTR      |
| DCTN1    | GJB3    | MORC2   | SCN10A  | TUBB3    |
| DGAT2    | GLA     | MPZ     | SCN11A  | UBA5     |
| DHTKD1   | GNB4    | MTMR2   | SCN9A   | VCP      |
| DNAJB2   | GSN     | MYH14   | SCO2    | WARS1    |
| DNAJB5   | HARS1   | MYH7    | SEPTIN9 | WNK1     |
| DNM2     | HINT1   | NAGLU   | SGPL1   | YARS1    |
| DNMT1    | HK1     | NDRG1   | SH3TC2  |          |
| DRP2     | HOXD10  | NEFH    | SLC12A6 |          |

| Dystonia / Parkinsons |         |          |
|-----------------------|---------|----------|
| ACTB                  | HPCA    | SLC2A1   |
| ADCY5                 | HTRA2   | SLC30A10 |
| ANO3                  | IRF2BPL | SLC39A14 |
| ATAD1                 | KCNMA1  | SLC6A3   |
| ATP13A2               | KCNN2   | SLC6A5   |
| ATP1A3                | KCTD17  | SNCA     |
| BCAP31                | KMT2B   | SPR      |
| CACNA1B               | LRRK2   | SYNJ1    |
| CHCHD2                | MECR    | TAF1     |
| DNAJC6                | MICU1   | TBC1D24  |
| DRD2                  | NKX2-1  | TH       |
| ECHS1                 | NUBPL   | THAP1    |
| EIF4G1                | PARK7   | TOMM70   |
| FBXO7                 | PDE2A   | TOR1A    |
| GBA                   | PINK1   | TOR1AIP1 |
| GCH1                  | PLA2G6  | TUBB4A   |
| GIGYF2                | PNKD    | UCHL1    |
| GLRA1                 | PRKN    | VPS13A   |
| GLRB                  | PRKRA   | VPS13C   |
| GNAL                  | PRRT2   | VPS16    |
| GNAO1                 | SGCE    | VPS35    |
| HEXA                  | SLC20A2 | VPS41    |
| HEXB                  |         |          |

| FHM    |         |       |
|--------|---------|-------|
| ATP1A2 | CACNA1A | SCN1A |

| HSP      |           |          |         |
|----------|-----------|----------|---------|
| ABCD1    | DDHD2     | KLC2     | SLC16A2 |
| ADAR     | DNM2      | KLC4     | SLC1A4  |
| ADD3     | DNMT1     | L1CAM    | SLC33A1 |
| AFG3L2   | DSTYK     | LYST     | SOX10   |
| ALDH18A1 | ENTPD1    | MAG      | SPART   |
| ALDH3A2  | ERLIN1    | MAPK8IP3 | SPAST   |
| ALS2     | ERLIN2    | MARS1    | SPG11   |
| AMPD2    | EXOSC3    | MARS2    | SPG21   |
| AP4B1    | FA2H      | MTPAP    | SPG7    |
| AP4E1    | FARS2     | NEFL     | SPTAN1  |
| AP4M1    | FLRT1     | NIPA1    | STUB1   |
| AP4S1    | GAD1      | NKX6-2   | SVBP    |
| AP5Z1    | GALC      | NOVA2    | TBCE    |
| ARL6IP1  | GBA2      | NT5C2    | TECPR2  |
| ARSI     | GJC2      | PCYT2    | TFG     |
| ATL1     | GLRX5     | PEX10    | TPP1    |
| ATP13A2  | GPT2      | PGAP1    | TREX1   |
| ATP1A1   | GRN       | PLA2G6   | TUBB4A  |
| ATP2B4   | HACE1     | PLP1     | UBAP1   |
| ATP6AP2  | HEXA      | PNPLA6   | UCHL1   |
| B4GALNT1 | HEXB      | POLR3A   | UNC79   |
| BSCL2    | HSPD1     | RAB3GAP2 | UNC80   |
| MTRFR    | IBA57     | REEP1    | USP8    |
| C19orf12 | IFIH1     | REEP2    | VAMP1   |
| CACNA1E  | IRF2BPL   | RNASEH2B | VPS37A  |
| CAPN1    | KANK1     | RTN2     | WASHC5  |
| CCT5     | KIDINS220 | SACS     | WDR48   |
| CPT1C    | KIF1A     | SAMHD1   | ZFR     |
| CYP2U1   | KIF1C     | SARS2    | ZFYVE26 |
| CYP7B1   | KIF5A     | SELENOI  | ZFYVE27 |
| DDHD1    |           |          |         |

| IBGC  |       |         |
|-------|-------|---------|
| JAM2  | NRROS | PDGFRB  |
| JAM3  | OCLN  | SLC20A2 |
| MYORG | PDGFB | XPR1    |

| Leukodystrophy |         |          |
|----------------|---------|----------|
| AARS2          | EIF2B1  | PANK2    |
| ABCD1          | EIF2B2  | PDCD10   |
| ACTA2          | EIF2B3  | PLA2G6   |
| AIMP1          | EIF2B4  | PLP1     |
| AIMP2          | EIF2B5  | POLR1C   |
| APP            | EPRS1   | POLR3A   |
| ARSA           | FAM126A | POLR3B   |
| ASPA           | FTL     | PSAP     |
| C19orf12       | GALC    | PYCR2    |
| CCM2           | GFAP    | RARS1    |
| CHD4           | GJC2    | REPS1    |
| CLCN2          | GLA     | RNASET2  |
| CNOT3          | GUCY1A1 | RNF213   |
| COASY          | HEPACAM | RPIA     |
| COL4A1         | HIKESHI | SETD5    |
| COL4A2         | HSPD1   | TMEM106B |
| CRAT           | HTRA1   | TMEM63A  |
| CSF1R          | KARS1   | TREX1    |
| CST3           | KRIT1   | TUBB4A   |
| DARS2          | LMNB1   | UBTF     |
| DEGS1          | MLC1    | UFM1     |
| EIF2AK1        | NAXD    | VPS11    |
| EIF2AK2        | NOTCH3  | WDR45    |

| MND / SMA |         |
|-----------|---------|
| AIFM1     | MATR3   |
| ALS2      | NEK1    |
| ANG       | OPTN    |
| ANXA11    | PFN1    |
| ASAH1     | PLEKHG5 |
| ASCC1     | PRPH    |
| ATP7A     | SETX    |
| BICD2     | SIGMAR1 |
| CHCHD10   | SLC52A1 |
| CHMP2B    | SLC52A2 |
| DCTN1     | SLC52A3 |
| DNAJB2    | SOD1    |
| DYNC1H1   | SPG11   |
| ERBB4     | SQSTM1  |
| EXOSC8    | TARDBP  |
| EXOSC9    | TBCE    |
| FIG4      | TBK1    |
| FUS       | TRIP4   |
| GLT8D1    | TRPV4   |
| HEXA      | TUBA4A  |
| HEXB      | UBA1    |
| HNRNPA1   | UBQLN2  |
| HNRNPA2B1 | VAPB    |
| IGHMBP2   | VCP     |
| LAS1L     | VRK1    |

| Neurodegeneration (AD/FTD/CAA/NP) |        |
|-----------------------------------|--------|
| APP                               | NPC2   |
| CHCHD10                           | PRNP   |
| CHMP2B                            | PSEN1  |
| CST3                              | PSEN2  |
| CYLD                              | SMPD1  |
| FUS                               | SQSTM1 |
| GBE1                              | TARDBP |
| GRN                               | TBK1   |
| ITM2B                             | TUBA4A |
| MAPT                              | VCP    |
| NPC1                              |        |

| Rett syndrome |  |
|---------------|--|
| CDKL5         |  |
| FOXG1         |  |
| GABBR2        |  |
| MECP2         |  |
| SMC1A         |  |