

The following applies to DWPAN / Comprehensive Distal Weakness Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is was updated August 2024. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, or genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
AAAS	NM_015665.6	-	-
AARS1	NM_001605.2	-	-
ABCA1	NM_005502.4	c.4176-11T>G	-
ABCD1	NM_000033.4	c.-16_10del	-
ACTA1	NM_001100.4	-	-
AIFM1	NM_004208.4	c.697-44T>G	-
ALDH18A1	NM_002860.4	-	-
AMACR	NM_014324.6	-	-
ANO5	NM_213599.2	-	CNV in exon 2 may not be detected or reported
AP5Z1	NM_014855.3	-	-
APOA1	NM_000039.2	-	-
APTX	NM_175073.2	-	-
ARSA	NM_000487.6	-	-
ATL1	NM_015915.4	-	-
ATL3	NM_015459.5	-	CNV in exon 5 may not be detected or reported
ATM	NM_000051.3	c.4612-12A>G c.6573-12C>A	-
ATP1A1	NM_000701.8	-	-
ATP7A	NM_000052.7	-	-
B4GALNT1	NM_001478.5	-	-
BAG3	NM_004281.3	-	-
BICD2	NM_001003800.2	-	-
BIN1	NM_139343.3	-	-
BSCL2	NM_032667.6	-	-
C12orf65 (MTRFR)	NM_152269.5	-	-
C1orf194	NM_001122961.2	-	-
CAV3	NM_033337.3	-	-
CHCHD10	NM_213720.3	-	-
CLCF1	NM_013246.3	-	-
CLTCL1	NM_007098.4	-	-

# Targeted Genes and Methodology Details for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CNTNAP1</i>	NM_003632.3	-	-
<i>COQ4</i>	NM_016035.5	-	-
<i>COQ7</i>	NM_016138.5	-	-
<i>COX10</i>	NM_001303.4	-	CNV in exon 6 may not be detected or reported
<i>COX20</i>	NM_198076.6	-	-
<i>COX6A1</i>	NM_004373.4	-	-
<i>CPOX</i>	NM_000097.7	-	-
<i>CRLF1</i>	NM_004750.5	-	-
<i>CRYAB</i>	NM_001885.3	-	-
<i>CTDP1</i>	NM_004715.4	c.863+389C>T	-
<i>CYP27A1</i>	NM_000784.4	-	-
<i>CYP2U1</i>	NM_183075.3	-	-
<i>CYP7B1</i>	NM_004820.5	-	-
<i>DCTN1</i>	NM_004082.4	-	-
<i>DDHD1</i>	NM_001160147.2	-	-
<i>DES</i>	NM_001927.4	-	-
<i>DGAT2</i>	NM_032564.5	-	-
<i>DHH</i>	NM_021044.4	-	-
<i>DNAJB2</i>	NM_001039550.2	-	-
<i>DNAJB6</i>	NM_058246.4	-	-
<i>DNM2</i>	NM_001005360.2	-	-
<i>DNMT1</i>	NM_001130823.3	-	CNV in exon 5 may not be detected or reported
<i>DST</i>	NM_015548.5	-	-
<i>DST</i>	NM_001144769.3	-	-
<i>DYNC1H1</i>	NM_001376.5	-	-
<i>DYSF</i>	NM_003494.4	c.1054-43_1059delinsA	-
<i>EGR2</i>	NM_000399.5	-	-
<i>ELP1</i>	NM_003640.5	-	-
<i>ERCC8</i>	NM_000082.3	-	CNV in exon 5 may not be detected or reported
<i>FA2H</i>	NM_024306.5	-	-
<i>FAM126A</i>	NM_032581.4	-	-
<i>FBLN5</i>	NM_006329.3	-	-
<i>FBXO38</i>	NM_030793.5	-	-
<i>FDX2</i>	NM_001031734.4	-	-
<i>FGD4</i>	NM_139241.3	-	-
<i>FGF14</i>	NM_004115.3	-	-
<i>FHL1</i>	NM_001449.5	-	-

# Targeted Genes and Methodology Details for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FIG4</i>	NM_014845.5	-	CNV in exon 17 may not be detected or reported
<i>FLNC</i>	NM_001458.4	-	-
<i>FLVCR1</i>	NM_014053.4	-	-
<i>FMR1</i>	NM_002024.5	-	CNV in exon 2 may not be detected or reported
<i>FXN</i>	NM_000144.5	-	-
<i>GALC</i>	NM_000153.4	c.-66G>C	CNV in exon 6 may not be detected or reported
<i>GAN</i>	NM_022041.3	-	CNV in exon 2 may not be detected or reported
<i>GARS1</i>	NM_002047.4	-	-
<i>GBA2</i>	NM_020944.3	-	-
<i>GBE1</i>	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTACATGACAGGT	-
<i>GBF1</i>	NM_004193.3	-	-
<i>GDAP1</i>	NM_018972.4	-	-
<i>GJB1</i>	NM_000166.6	c.-103C>T c.-17G>A c.-17+1G>T c.-17+2T>C	-
<i>GLA</i>	NM_000169.2	c.640-801C>T c.640-859G>A	-
<i>GM2A</i>	NM_000405.5	-	-
<i>GNB4</i>	NM_021629.4	-	-
<i>GNE</i>	NM_001128227.3	-	-
<i>GSN</i>	NM_000177.5	-	CNV in exon 16 may not be detected or reported
<i>HADHA</i>	NM_000182.5	-	CNV in exon 14 may not be detected or reported
<i>HADHB</i>	NM_000183.3	-	-
<i>HARS1</i>	NM_002109.6	-	-
<i>HEXA</i>	NM_000520.6	-	-
<i>HEXB</i>	NM_000521.4	c.1509-26G>A	CNV in exon 4 may not be detected or reported
<i>HINT1</i>	NM_005340.7	-	-
<i>HK1</i>	NM_000188.2	c.-40257G>C c.-40237G>C	-
<i>HMBS</i>	NM_000190.4	-	-
<i>HNRNPA1</i>	NM_031157.4	-	-
<i>HNRNPA2B1</i>	NM_031243.3	-	-
<i>HSPB1</i>	NM_001540.5	-	-
<i>HSPB8</i>	NM_014365.2	-	-
<i>HSPD1</i>	NM_002156.5	-	-

# Targeted Genes and Methodology Details for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>IARS2</i>	NM_018060.4	-	-
<i>IBA57</i>	NM_001010867.4	-	-
<i>IGHMBP2</i>	NM_002180.2	-	-
<i>INF2</i>	NM_022489.4	-	-
<i>KARS1</i>	NM_001130089.1	-	-
<i>KIF1A</i>	NM_004321.7	-	-
<i>KIF5A</i>	NM_004984.4	-	-
<i>LAMA2</i>	NM_000426.3	c.3556-13T>A	-
<i>LDB3</i>	NM_001080116.1	-	-
<i>LITAF</i>	NM_004862.3	-	-
<i>LMNA</i>	NM_170707.4	c.1698+13C>A	-
<i>LRSAM1</i>	NM_138361.5	-	-
<i>MARS1</i>	NM_004990.4	-	-
<i>MATR3</i>	NM_199189.2	-	Duplication analysis for CNV will not be performed
<i>MCM3AP</i>	NM_003906.5	-	-
<i>MFN2</i>	NM_014874.4	-	-
<i>MME</i>	NM_007289.3	-	-
<i>MORC2</i>	NM_001303256.3	-	-
<i>MPC1</i>	NM_016098.4	-	CNV in exon 2 may not be detected or reported
<i>MPV17</i>	NM_002437.5	-	-
<i>MPZ</i>	NM_000530.8	-	-
<i>MTMR2</i>	NM_016156.5	-	-
<i>MTTP</i>	NM_000253.3	-	-
<i>MYH14</i>	NM_024729.3	-	-
<i>MYH2</i>	NM_017534.6	-	-
<i>MYH7</i>	NM_000257.4	-	CNV in exon 27 may not be detected or reported
<i>MYOT</i>	NM_006790.3	-	-
<i>NAGLU</i>	NM_000263.4	-	-
<i>NDRG1</i>	NM_006096.4	-	-
<i>NEB</i>	NM_001271208.2	-	Sequence variants and CNV in exons 82–105 will not be detected or reported
<i>NEFH</i>	NM_021076.4	-	-
<i>NEFL</i>	NM_006158.4	-	-
<i>NF2</i>	NM_000268.3	-	-
<i>NGF</i>	NM_002506.3	-	-
<i>NIPA1</i>	NM_144599.5	-	-
<i>NTRK1</i>	NM_001012331.1	c.851-33T>A	-

# Targeted Genes and Methodology Details for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>OPA1</i>	NM_015560.2	c.625-5459G>A	-
<i>PDK3</i>	NM_001142386.3	-	-
<i>PDYN</i>	NM_024411.5	-	-
<i>PEX7</i>	NM_000288.4	c.-45C>T	-
<i>PHYH</i>	NM_006214.4	-	-
<i>PLA2G6</i>	NM_003560.4	-	-
<i>PLEKHG5</i>	NM_020631.5	-	-
<i>PLP1</i>	NM_000533.5	c.453+28_453+46del c.453+159G>A c.453+164G>A c.454-322G>A c.454-314T>A/G c.454-312C>G	
<i>PMP2</i>	NM_002677.5	-	-
<i>PMP22</i>	NM_000304.4	-	-
<i>PNKP</i>	NM_007254.4	c.1386+49_1387-33del	-
<i>PNPLA6</i>	NM_006702.5	-	-
<i>POLG</i>	NM_002693.2	-	-
<i>PPOX</i>	NM_000309.5	-	-
<i>PRDM12</i>	NM_021619.3	-	-
<i>PRKCG</i>	NM_002739.5	-	-
<i>PRNP</i>	NM_000311.5	-	-
<i>PRPS1</i>	NM_002764.4	-	-
<i>PRX</i>	NM_181882.3	-	-
<i>PTRH2</i>	NM_016077.4	-	-
<i>RAB7A</i>	NM_004637.6	-	-
<i>REEP1</i>	NM_022912.3	-	-
<i>RETREG1</i>	NM_001034850.2	-	-
<i>RNASEH1</i>	NM_002936.5	-	-
<i>RRM2B</i>	NM_015713.5	-	-
<i>RTN2</i>	NM_005619.5	-	-
<i>SACS</i>	NM_014363.6	-	-
<i>SBF1</i>	NM_002972.4	-	-
<i>SBF2</i>	NM_030962.3	-	CNV in exon 10 may not be detected or reported
<i>SCN10A</i>	NM_006514.3	-	-
<i>SCN11A</i>	NM_014139.2	-	-
<i>SCN9A</i>	NM_002977.3	-	-
<i>SCO2</i>	NM_005138.2	-	-
<i>SELENON</i>	NM_020451.3	c.-71 to c.-11	CNV in exon 3 may not be detected or reported

# Targeted Genes and Methodology Details for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SEPTIN9</i>	NM_006640.4	c.-134G>C	-
<i>SETX</i>	NM_015046.7	-	-
<i>SH3TC2</i>	NM_024577.4	-	-
<i>SIGMAR1</i>	NM_005866.4	-	-
<i>SLC12A6</i>	NM_133647.1	-	CNV in exon 3 may not be detected or reported
<i>SLC25A19</i>	NM_021734.4	-	-
<i>SLC25A46</i>	NM_138773.4	-	-
<i>SLC52A2</i>	NM_024531.5	-	-
<i>SLC52A3</i>	NM_033409.4	-	-
<i>SLC5A7</i>	NM_021815.5	-	-
<i>SMN1</i>	NM_022874.2	g.27134T>G (NG_008691.1) Provided only upon request	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed
<i>SMN2</i>	NM_022876.2	-	Analyzed for the presence of exon 7 CNV only; analysis for other sequence variants and CNV will not be performed
<i>SNAP29</i>	NM_004782.4	-	-
<i>SOD1</i>	NM_000454.4	c.358-11A>G	-
<i>SORD</i>	NM_003104.6	-	Sequence variants in exons 3 and 9 will not be detected or reported, CNV in exons 3 and 8–9 will not be detected or reported
<i>SOX10</i>	NM_006941.4	-	-
<i>SPAST</i>	NM_014946.3	-	-
<i>SPG11</i>	NM_025137.4	-	-
<i>SPG21</i>	NM_016630.7	-	-
<i>SPG7</i>	NM_003119.4	-	-
<i>SPTAN1</i>	NM_001130438.3	-	-
<i>SPTLC1</i>	NM_006415.4	-	-
<i>SPTLC2</i>	NM_004863.3	-	-
<i>SQSTM1</i>	NM_003900.5	-	-
<i>SUCLA2</i>	NM_003850.2	-	-
<i>SURF1</i>	NM_003172.4	-	-
<i>TDP1</i>	NM_018319.4	-	-
<i>TFG</i>	NM_006070.6	-	-
<i>TIA1</i>	NM_022173.4	-	-
<i>TRIM2</i>	NM_001130067.2	-	-
<i>TRPV4</i>	NM_021625.5	-	-

# Targeted Genes and Methodology Details

## for Comprehensive Distal Weakness Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TSFM</i>	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
<i>TTN</i>	NM_133378.4	-	Sequence variants and CNV in exons 153–155 will not be detected or reported
<i>TTPA</i>	NM_000370.3	-	-
<i>TTR</i>	NM_000371.3	-	-
<i>TUBB3</i>	NM_006086.4	-	-
<i>TWNK</i>	NM_021830.5	-	-
<i>TYMP</i>	NM_001953.5	-	-
<i>UBA1</i>	NM_003334.4	-	-
<i>VCP</i>	NM_007126.5	-	-
<i>VPS13D</i>	NM_015378.4	-	-
<i>VRK1</i>	NM_003384.3	-	-
<i>VWA1</i>	NM_022834.5	-	-
<i>WARS1</i>	NM_004184.4	-	-
<i>WASHC5</i>	NM_014846.4	-	-
<i>WNK1</i>	NM_213655.4	-	-
<i>YARS1</i>	NM_003680.3	-	-
<i>ZFYVE26</i>	NM_015346.4	-	-

Effective Date	Version	Synopsis of Change
August 6, 2024	V2	Added gene <i>SEPTIN9</i>