



The following applies to MNDP / Inherited Motor Neuron Disease 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. A polymerase chain reaction-based assay is performed to test for the presence of C9orf72 GGGGCC hexanucleotide repeat expansions. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from December 2022 to the present. 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
<i>ALS2</i>	NM_020919.4	-	-
<i>ANG</i>	NM_001145.4	-	-
<i>ANXA11</i>	NM_001157.3	-	-
<i>ASAH1</i>	NM_177924.5	-	CNV in exon 4 may not be detected or reported
<i>C9orf72</i>	NM_001256054.3	GGGGCC hexanucleotide repeat expansion	Analyses for sequence variants and CNV will not be performed
<i>CCNF</i>	NM_001761.3	-	-
<i>CHCHD10</i>	NM_213720.3	-	-
<i>CHMP2B</i>	NM_014043.4	-	-
<i>DCTN1</i>	NM_004082.4	-	-
<i>ERBB4</i>	NM_005235.3	-	-
<i>FIG4</i>	NM_014845.5	-	CNV in exon 17 may not be detected or reported
<i>FUS</i>	NM_004960.3	c.*59G>A	-
<i>HEXB</i>	NM_000521.4	c.1509-26G>A	CNV in exon 4 may not be detected or reported
<i>HNRNPA1</i>	NM_031157.4	-	-
<i>HNRNPA2B1</i>	NM_031243.3	-	-
<i>KIF5A</i>	NM_004984.4	-	-
<i>MATR3</i>	NM_199189.2	-	Duplication analysis for CNV will not be performed
<i>NEFH</i>	NM_021076.4	-	-
<i>OPTN</i>	NM_021980.4	-	-
<i>PFN1</i>	NM_005022.4	-	-
<i>SETX</i>	NM_015046.7	-	-
<i>SIGMAR1</i>	NM_005866.4	-	-
<i>SOD1</i>	NM_000454.4	c.358-11A>G	-
<i>SPG11</i>	NM_025137.4	-	-
<i>SQSTM1</i>	NM_003900.5	-	-
<i>TAF15</i>	NM_139215.3	-	-
<i>TARDBP</i>	NM_007375.3	-	-
<i>TBK1</i>	NM_013254.4	-	CNV in exon 16 may not be detected or reported
<i>TIA1</i>	NM_022173.4	-	-
<i>TUBA4A</i>	NM_006000.3	-	-
<i>UBQLN2</i>	NM_013444.3	-	-
<i>VAPB</i>	NM_004738.5	-	-
<i>VCP</i>	NM_007126.5	-	-
<i>VRK1</i>	NM_003384.3	-	-