MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis Gene Panel

The following applies to AFTDP / Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis 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 test 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
ALS2	NM_020919.4	-	-
ANG	NM_001145.4	-	-
ANXA11	NM_001157.3	-	-
APP	NM_000484.4	-	-
ASAH1	NM_177924.5	-	CNV in exon 4 may not be detected or reported
C9orf72	NM_001256054.3	GGGGCC hexanucleotide repeat expansion	Analyses for sequence variants and CNV will not be performed
CCNF	NM_001761.3	-	-
CHCHD10	NM_213720.3	-	-
CHMP2B	NM_014043.4	-	-
CSF1R	NM_005211.3	c.1969+115_1969+116del c.1859-119G>A	-
DCTN1	NM_004082.4	-	-
ERBB4	NM_005235.3	-	-
FIG4	NM_014845.5	-	CNV in exon 17 may not be detected or reported
FUS	NM_004960.3	c.*59G>A	-
GRN	NM_002087.3	c.-256 to c.-8+10	-
HEXB	NM_000521.4	c.1509-26G>A	CNV in exon 4 may not be detected or reported
HNRNPA1	NM_031157.4	-	-
HNRNPA2B1	NM_031243.3	-	-
ITM2B	NM_021999.5	-	-
KIF5A	NM_004984.4	-	-
MAPT	NM_005910.5	c.823-15T>C c.915+11 to c.915+24	-
MATR3	NM_199189.2	-	Duplication analysis for CNV will not be performed
NEFH	NM_021076.4	-	-
NOTCH3	NM_000435.3	c.341-26_341-24del	-

Targeted Genes and Methodology Details for Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
NPC1	NM_000271.5	c.882-28A>T/G c.1554-1009G>A c.3591+105A>T c.3591+121C>T	-
NPC2	NM_006432.4	-	-
OPTN	NM_021980.4	-	-
PANK2	NM_153638.3	-	-
PFN1	NM_005022.4	-	-
PRNP	NM_000311.5	-	-
PSEN1	NM_000021.4	-	-
PSEN2	NM_000447.3	-	-
SETX	NM_015046.7	-	-
SIGMAR1	NM_005866.4	-	-
SNCA	NM_000345.4	-	-
SOD1	NM_000454.4	c.358-11A>G	-
SPG11	NM_025137.4	-	-
SPTLC1	NM_006415.4	-	-
SQSTM1	NM_003900.5	-	-
TAF15	NM_139215.3	-	-
TARDBP	NM_007375.3	-	-
TBK1	NM_013254.4	-	CNV in exon 16 may not be detected or reported
TBP	NM_003194.5	-	Variants provided only upon request
TIA1	NM_022173.4	-	-
TIMM8A	NM_004085.4	-	-
TREM2	NM_018965.4	-	-
TUBA4A	NM_006000.3	-	-
TYROBP	NM_003332.4	-	-
UBQLN2	NM_013444.3	-	-
VAPB	NM_004738.5	-	-
VCP	NM_007126.5	-	-
VRK1	NM_003384.3	-	-

Effective Date	Version	Synopsis of Test Change
August 6, 2024	V2	Added gene *SPTLC1*