MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Ataxia Gene Panel

The following applies to ATAXP / Inherited Ataxia 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 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
AAAS	NM_015665.6	-	-
ABCB7	NM_004299.6	-	-
ABHD12	NM_001042472.3	-	-
ACO2	NM_001098.3	-	-
ADPRHL2 (ADPRS)	NM_017825.3	-	-
AFG3L2	NM_006796.3	-	-
ALDH5A1	NM_001080.3	-	-
ALG6	NM_013339.4	-	-
ALS2	NM_020919.4	-	-
ANO10	NM_018075.5	-	-
APOPT1 (COA8)	NM_032374.4	-	-
APTX	NM_175073.2	-	-
ARSA	NM_000487.6	-	-
ATCAY	NM_033064.5	-	-
ATM	NM_000051.3	c.4612-12A>G c.6573-12C>A	-
ATP1A3	NM_152296.5	-	-
ATP8A2	NM_016529.6	-	CNV in exon 6 may not be detected or reported
AUH	NM_001698.2	-	-
C12orf65 (MTRFR)	NM_152269.5	-	-
C19orf12	NM_001031726.3	-	CNV in exon 1 may not be detected or reported
CA8	NM_004056.6	-	-
CACNA1A	NM_001127221.1	-	-
CACNA1G	NM_018896.5	-	-
CAMTA1	NM_015215.4	-	-
CAPN1	NM_001198868.2	-	-
CC2D2A	NM_001080522.2	-	-
CCDC88C	NM_001080414.4	-	-

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Targeted Genes and Methodology Details for Inherited Ataxia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CHCHD10	NM_213720.3	-	-
CLCN2	NM_004366.6	-	-
CLN5	NM_006493.4	-	-
CLPP	NM_006012.4	-	-
COQ2	NM_015697.8	-	-
COQ8A	NM_020247.5	-	-
COX20	NM_198076.6	-	-
CP	NM_000096.4	-	CNV in exon 19 may not be detected or reported
CTBP1	NM_001328.3	-	CNV in exon 1 may not be detected or reported
CWF19L1	NM_018294.6	-	-
CYP27A1	NM_000784.4	-	-
DARS2	NM_018122.5	c.228-33 to c.228-11 c.1345-17_1345-5del	-
DLD	NM_000108.5	-	-
DNAJC19	NM_145261.4	-	-
DNAJC3	NM_006260.5	-	-
DNMT1	NM_001130823.3	-	CNV in exon 5 may not be detected or reported
EBF3	NM_001005463.3	-	-
EIF2B1	NM_001414.4	-	-
EIF2B2	NM_014239.4	-	-
EIF2B3	NM_020365.5	-	-
EIF2B4	NM_015636.3	-	-
EIF2B5	NM_003907.3	-	-
ELOVL4	NM_022726.4	-	-
ELOVL5	NM_021814.5	-	-
EPM2A	NM_005670.4	-	-
FA2H	NM_024306.5	-	-
FBXL4	NM_012160.4	-	-
FGF14	NM_004115.3	-	-
FLVCR1	NM_014053.4	-	-
FMR1	NM_002024.5	-	CNV in exon 2 may not be detected or reported
FOLR1	NM_016725.3	-	-
FXN	NM_000144.5	-	-
GAMT	NM_000156.6	-	-
GBA2	NM_020944.3	-	-
GCDH	NM_000159.4	-	-

Targeted Genes and Methodology Details for Inherited Ataxia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
GDAP2	NM_017686.4	-	CNV in exon 12 may not be detected or reported
GFAP	NM_002055.5	c.1171+475_1171+482delinsATC c.1171+472G>A	-
GJC2	NM_020435.4	-	-
GOSR2	NM_004287.4	-	CNV in exon 2 may not be detected or reported
GRID2	NM_001510.4	-	-
GRM1	NM_001278064.2	-	-
HEPACAM	NM_152722.5	-	-
HEXA	NM_000520.6	-	-
HIBCH	NM_014362.4	-	-
ITM2B	NM_021999.5	-	-
ITPR1	NM_002222.6	-	-
KCNA1	NM_000217.3	-	-
KCNA2	NM_004974.4	-	-
KCNC3	NM_004977.3	-	CNV in exon 4 may not be detected or reported
KCND3	NM_004980.4	-	-
KCNJ10	NM_002241.5	-	-
KCTD7	NM_153033.4	-	-
KIF1C	NM_006612.6	-	-
KIF5A	NM_004984.4	-	-
L2HGDH	NM_024884.3	-	CNV in exon 6 may not be detected or reported
LAMA1	NM_005559.4	-	-
LYRM7	NM_181705.4	-	CNV in exon 3 may not be detected or reported
MARS2	NM_138395.4	-	-
MECR	NM_016011.5	-	-
MLC1	NM_015166.3	-	-
MPV17	NM_002437.5	-	-
MRE11	NM_005591.3	-	-
MSTO1	NM_018116.3	-	Sequence variants and CNV in exons 1–7, 13 and 14 will not be detected or reported
MTFMT	NM_139242.4	-	-
MTO1	NM_012123.4	-	-
MTTP	NM_000253.3	-	-
NDUFAF2	NM_174889.5	-	-
NDUFAF6	NM_152416.4	c.420+784C>T	-
NDUFS1	NM_005006.7	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
NDUFS7	NM_024407.5	-	-
NDUFV1	NM_007103.4	-	-
NEU1	NM_000434.4	-	-
NKX6-2	NM_177400.3	-	-
NOTCH3	NM_000435.3	-	Sequence variants and CNV in exons 82–105 will not be detected or reported
NUBPL	NM_025152.3	c.815-27T>C	-
OPA1	NM_015560.2	c.625-5459G>A	-
OPA3	NM_025136.4	-	-
OTC	NM_000531.6	-	-
PANK2	NM_153638.3	-	-
PCNA	NM_002592.2	-	-
PDHA1	NM_000284.4	c.419-17_419-14del c.511-30G>A c.759+26G>A	-
PDSS2	NM_020381.4	-	CNV in exon 7 may not be detected or reported
PDYN	NM_024411.5	-	-
PEX10	NM_153818.1	-	-
PEX7	NM_000288.4	c.-45C>T	-
PHYH	NM_006214.4	-	-
PLA2G6	NM_003560.4	-	-
PLD3	NM_012268.4	-	-
PLP1	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	-
PMM2	NM_000303.3	c.179-25A>G c.640-15479C>T c.640-23A>G	-
PMPCA	NM_015160.3	-	-
PNKP	NM_007254.4	c.1386+49_1387-33del	-
PNPLA6	NM_006702.5	-	-
POLG	NM_002693.2	-	-
POLR1C	NM_203290.4	-	-
POLR3A	NM_007055.4	c.1909+18G>A c.1909+22G>A c.3337-11T>C c.*18C>T	-
POLR3B	NM_018082.6	-	-
PRKCG	NM_002739.5	-	-
PRNP	NM_000311.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PRRT2	NM_145239.3	c.880-34G>A	-
PSAP	NM_002778.4	c.777+1915C>A	-
PTRH2	NM_016077.4	-	-
PUM1	NM_001020658.2	-	-
RARS1	NM_002887.4	-	-
RNASEH1	NM_002936.5	-	-
RNF170	NM_001160223.1	-	-
RNF216	NM_207111.4	-	-
RPGRIP1L	NM_015272.5	-	CNV in exon 23 may not be detected or reported
RRM2B	NM_015713.5	-	-
RUBCN	NM_001145642.4	-	-
SACS	NM_014363.6	-	-
SAMD9L	NM_152703.5	-	-
SCN2A	NM_021007.3	-	-
SCN8A	NM_014191.4	-	-
SCN8A	NM_001330260.2	-	-
SCYL1	NM_020680.4	-	-
SDHA	NM_004168.4	-	-
SDHAF1	NM_001042631.2	-	-
SETX	NM_015046.7	-	-
SIL1	NM_022464.5	-	-
SLC16A2	NM_006517.5	-	-
SLC17A5	NM_012434.5	-	-
SLC19A3	NM_025243.4	c.980-14A>G	-
SLC1A3	NM_004172.5	-	-
SLC25A3	NM_005888.3	-	-
SLC25A46	NM_138773.4	-	-
SLC2A1	NM_006516.3	c.680-11G>A c.679+25_680-43del	
SLC44A1	NM_080546.5	-	-
SLC52A2	NM_024531.5	-	-
SLC52A3	NM_033409.4	-	-
SLC6A19	NM_001003841.3	-	-
SLC9A1	NM_003047.5	-	-
SLC9A6	NM_006359.3	-	-
SNX14	NM_153816.6	-	-
SPAST	NM_014946.3	-	-
SPG11	NM_025137.4	-	-

Targeted Genes and Methodology Details for Inherited Ataxia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SPG7	NM_003119.4	-	-
SPR	NM_003124.5	-	-
SPTBN2	NM_006946.3	-	-
SQSTM1	NM_003900.5	-	-
SRD5A3	NM_024592.5	-	-
STUB1	NM_005861.4	-	-
SUMF1	NM_182760.4	-	-
SURF1	NM_003172.4	-	-
SYNE1	NM_033071.3	c.15705-12A>G	-
TACO1	NM_016360.4	-	-
TBP	NM_003194.5	-	Variants provided only upon request
TDP1	NM_018319.4	-	-
TDP2	NM_016614.3	-	-
TGM6	NM_198994.3	-	-
THG1L	NM_017872.5	-	-
TIMM8A	NM_004085.4	-	-
TMEM216	NM_001173990.3	-	-
TMEM240	NM_001114748.2	-	-
TMEM67	NM_153704.6	-	-
TMEM70	NM_017866.6	-	-
TPK1	NM_022445.4	-	-
TPP1	NM_000391.4	c.887-18A>G	-
TSFM	NM_001172696.2	-	CNV in exon 5 may not be detected or reported
TTBK2	NM_173500.4	-	-
TTC19	NM_017775.4	-	-
TTPA	NM_000370.3	-	-
TUBB4A	NM_006087.4	-	-
TWNK	NM_021830.5	-	-
UBA5	NM_024818.4	-	CNV in exon 5 may not be detected or reported
VAMP1	NM_014231.5	-	-
VARS2	NM_001167734.1	-	-
VLDLR	NM_003383.5	-	-
VPS13D	NM_015378.4	-	-
WDR73	NM_032856.4	-	-
WDR81	NM_001163809.1	-	-
WFS1	NM_006005.3	-	-