MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Spastic Paraplegia Gene Panel

The following applies to ISPP / Inherited Spastic Paraplegia 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
ABCD1	NM_000033.4	c.-16_10del	-
ACO2	NM_001098.3	-	-
AFG3L2	NM_006796.3	-	-
ALDH18A1	NM_002860.4	-	-
AMPD2	NM_001257360.1	-	-
AP4B1	NM_006594.5	-	-
AP4E1	NM_007347.5	-	-
AP4M1	NM_004722.4	-	-
AP4S1	NM_007077.4	-	-
AP5Z1	NM_014855.3	-	-
APOPT1 (COA8)	NM_032374.4	-	-
ARG1	NM_000045.4	-	-
ARL6IP1	NM_015161.3	-	-
ASNS	NM_133436.3	-	-
ATL1	NM_015915.4	-	-
AUH	NM_001698.2	-	-
B4GALNT1	NM_001478.5	-	-
BICD2	NM_001003800.2	-	-
BOLA3	NM_212552.3	-	-
BSCL2	NM_032667.6	-	-
C12orf65 (MTRFR)	NM_152269.5	-	-
COQ7	NM_016138.5	-	-
CPT1C	NM_001136052.2	-	CNV in exons 6–7 may not be detected or reported
CTC1	NM_025099.6	-	-
CTSD	NM_001909.5	-	-
CYP27A1	NM_000784.4	-	-
CYP2U1	NM_183075.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
CYP7B1	NM_004820.5	-	-
DARS1	NM_001349.4	-	CNV in exons 2–3, 5–10, 12–14 will not be detected or reported
DARS2	NM_018122.5	c.228-33 to c.228-11 c.1345-17_1345-5del	CNV analysis in exons 1, 26 is not performed
DDHD1	NM_001160147.2	-	-
DDHD2	NM_015214.3	-	-
DLD	NM_000108.5	-	-
EARS2	NM_001083614.2	-	-
ENTPD1	NM_001776.6	-	-
ERLIN1	NM_006459.4	-	-
ERLIN2	NM_007175.8	-	-
EXOSC3	NM_016042.4	-	-
FA2H	NM_024306.5	-	-
FAR1	NM_032228.6	-	-
FARS2	NM_006567.5	-	-
FUCA1	NM_000147.4	-	-
FXN	NM_000144.5	-	-
GALC	NM_000153.4	c.-66G>C	CNV in exon 6 may not be detected or reported
GBA2	NM_020944.3	-	-
GBE1	NM_000158.4	c.2053-3358_2053-3350delinsTGTTTTTTACATGACAGGT	-
GJC2	NM_020435.4	-	-
GLB1	NM_000404.4	-	-
GM2A	NM_000405.5	-	-
GPT2	NM_133443.4	-	-
HACE1	NM_020771.4	-	CNV in exons 5 and 7 may not be detected or reported
HEXA	NM_000520.6	-	-
HIBCH	NM_014362.4	-	-
HSPD1	NM_002156.5	-	-
HTRA1	NM_002775.5	-	-
IBA57	NM_001010867.4	-	-
IFIH1	NM_022168.4	-	-
IRF2BPL	NM_024496.4	-	-
ISCA2	NM_194279.4	-	-
KDM5C	NM_004187.4	-	-
KIDINS220	NM_020738.4	-	-
KIF1A	NM_004321.7	-	-
KIF5A	NM_004984.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
L1CAM	NM_000425.5	c.2432-19A>C	-
L2HGDH	NM_024884.3	-	CNV in exon 6 may not be detected or reported
LYRM7	NM_181705.4	-	CNV in exon 3 may not be detected or reported
MAG	NM_002361.4	-	-
MARS2	NM_138395.4	-	-
MED17	NM_004268.5	-	-
MTFMT	NM_139242.4	-	-
NIPA1	NM_144599.5	-	-
NT5C2	NM_012229.4	-	-
NUBPL	NM_025152.3	c.815-27T>C	-
OPA3	NM_025136.4	-	-
PANK2	NM_153638.3	-	-
PDHX	NM_003477.3	c.816+11C>G	-
PEX16	NM_004813.3	-	-
PGAP1	NM_024989.4	-	CNV in exons 9 and 17 may not be detected or reported
PLA2G6	NM_003560.4	-	-
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	
PNP	NM_000270.3	-	-
PNPLA6	NM_006702.5	-	-
PNPLA8	NM_015723.5	-	-
PRF1	NM_001083116.3	-	-
PRUNE1	NM_021222.3	-	-
PSAP	NM_002778.4	c.777+1915C>A	-
PYCR2	NM_013328.4	-	-
RAB18	NM_021252.5	-	-
RAB3GAP1	NM_012233.3	-	-
RAB3GAP2	NM_012414.4	-	CNV in exon 17 may not be detected or reported
RARS1	NM_002887.4	-	-
REEP1	NM_022912.3	-	-
REEP2	NM_001271803.2	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
RNASEH2A	NM_006397.2	-	-
RNASEH2B	NM_024570.4	-	CNV in exon 8 may not be detected or reported
RNASEH2C	NM_032193.4	-	-
RTN2	NM_005619.5	-	-
SACS	NM_014363.6	-	-
SAMHD1	NM_015474.3	-	-
SDHAF1	NM_001042631.2	-	-
SERAC1	NM_032861.4	-	CNV in exon 3 may not be detected or reported
SLC12A6	NM_133647.1	-	CNV in exon 3 may not be detected or reported
SLC16A2	NM_006517.5	-	-
SLC19A3	NM_025243.4	c.980-14A>G	-
SLC33A1	NM_004733.4	-	-
SLC6A8	NM_005629.4	-	-
SOX2	NM_003106.4	-	-
SPART	NM_015087.5	-	-
SPAST	NM_014946.3	-	-
SPG11	NM_025137.4	-	-
SPG21	NM_016630.7	-	-
SPG7	NM_003119.4	-	-
SPTAN1	NM_001130438.3	-	-
SUMF1	NM_182760.4	-	-
TACO1	NM_016360.4	-	-
TBC1D20	NM_144628.4	-	-
TECPR2	NM_014844.5	-	-
TFG	NM_006070.6	-	-
TREX1	NM_033629.6	-	-
TTC19	NM_017775.4	-	-
TYROBP	NM_003332.4	-	-
UBAP1	NM_001171201.1	-	-
UBQLN2	NM_013444.3	-	-
VAMP1	NM_014231.5	-	-
VPS13D	NM_015378.4	-	-
WASHC5	NM_014846.4	-	-
ZFYVE26	NM_015346.4	-	-
ZFYVE27	NM_001002261.3	-	-