MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inherited Parkinson Disease Gene Panel

The following applies to PARDP / Inherited Parkinson 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. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from March 2023 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
ADORA1	NM_000674.3	-	-
AHSA1	NM_012111.3	-	Variants provided only upon request
ANG	NM_001145.4	-	-
ANO3	NM_031418.4	-	-
APP	NM_000484.4	-	-
ATP13A2	NM_022089.4	-	-
ATP1A3	NM_152296.5	-	-
ATP6AP2	NM_005765.3	c.301-11_301-10del	-
ATP7B	NM_000053.4	c.-676A>G c.-460G>A c.-447C>T c.-442G>A c.-436_-422del15 c.-388C>T c.-210A>T c.-133A>C c.-128A>C c.-123C>A c.-128_-124delAGCCG c.1947-19T>A c.3061-12T>A	-
C19orf12	NM_001031726.3	-	CNV in exon 1 may not be detected or reported
CHCHD2	NM_016139.4	-	-
CHMP2B	NM_014043.4	-	-
CLN3	NM_001042432.1	-	-
CP	NM_000096.4	-	CNV in exon 19 may not be detected or reported
CSF1R	NM_005211.3	c.1969+115_1969+116del c.1859-119G>A	-
CYP27A1	NM_000784.4	-	-
DCAF17	NM_025000.4	-	-
DCTN1	NM_004082.4	-	-
DDC	NM_000790.4	-	-
DNAJB2	NM_001039550.2	-	-
DNAJC12	NM_021800.3	-	-
DNAJC13	NM_015268.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
DNAJC6	NM_001256864.2	-	-
DNM1L	NM_012062.5	c.251-1532dup	-
EIF4G1	NM_198241.3	-	-
FBXO7	NM_012179.4	-	-
FTL	NM_000146.4	5'UTR c.-200 to c.-143	-
FUS	NM_004960.3	c.*59G>A	-
GBA	NM_001005741.3	-	-
GCH1	NM_000161.3	-	CNV in exon 4 may not be detected or reported
GIGYF2	NM_001103146.2	-	CNV in exons 8 and 18 may not be detected or reported
GRN	NM_002087.3	c.-256 to c.-8+10	-
HTRA2	NM_013247.4	-	-
KIF5A	NM_004984.4	-	-
LRP10	NM_014045.5	-	-
LRRK2	NM_198578.4	-	-
LYST	NM_000081.4	-	-
MAPT	NM_005910.5	c.823-15T>C c.915+11 to c.915+24	-
OPTN	NM_021980.4	-	-
PANK2	NM_153638.3	-	-
PARK7	NM_007262.5	-	CNV in exon 4 may not be detected or reported
PDE10A	NM_001130690.2	-	CNV in exons 1 and 10 may not be detected or reported
PDE8B	NM_003719.4	-	-
PDGFB	NM_002608.4	-	-
PDGFRB	NM_002609.4	-	-
PEX1	NM_000466.3	-	-
PINK1	NM_032409.3	-	-
PLA2G6	NM_003560.4	-	-
PLD3	NM_012268.4	-	-
PODXL	NM_005397.4	-	-
POLG	NM_002693.2	-	-
POLG2	NM_007215.4	-	-
PRKAR1B	NM_001164761.1	-	-
PRKN	NM_004562.3	-	-
PRKRA	NM_003690.5	-	CNV analysis will not be performed
PRRT2	NM_145239.3	c.880-34G>A	-
PSEN1	NM_000021.4	-	-
PSEN2	NM_000447.3	-	-
PTRHD1	NM_001013663.1	-	-
RAB29	NM_003929.3	-	-
RAB39B	NM_171998.4	-	-
RIC3	NM_001206671.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
SIGMAR1	NM_005866.4	-	-
SLC18A2	NM_003054.6	-	-
SLC20A2	NM_006749.5	-	-
SLC30A10	NM_018713.2	-	-
SLC39A14	NM_015359.6	-	-
SLC6A3	NM_001044.5	-	-
SNCA	NM_000345.4	-	-
SNCB	NM_001001502.3	-	-
SOD1	NM_000454.4	c.358-11A>G	-
SPG11	NM_025137.4	-	-
SPR	NM_003124.5	-	-
SQSTM1	NM_003900.5	-	-
SYNJ1	NM_003895.3	-	-
TAF1	NM_004606.4	-	-
TAF15	NM_139215.3	-	-
TARDBP	NM_007375.3	-	-
TENM4	NM_001098816.3	-	-
TH	NM_199292.3	c.-71C>T c.-70G>A c.1198-24T>A	
THAP1	NM_018105.3	-	-
TMEM230	NM_001009923.2	-	CNV in exon 2 may not be detected or reported
TOR1A	NM_000113.3	-	-
TUBA4A	NM_006000.3	-	-
TWNK	NM_021830.5	-	-
UBQLN2	NM_013444.3	-	-
UCHL1	NM_004181.5	-	-
UNC13A	NM_001080421.2	-	-
VCP	NM_007126.5	-	-
VPS13A	NM_033305.3	-	-
VPS13C	NM_020821.3	-	CNV in exons 1–2 and 12 may not be detected or reported
VPS35	NM_018206.6	-	-
WDR45	NM_007075.3	-	-
XPR1	NM_004736.4	-	-

Effective Date	Version	Synopsis of Test Change
3/10/2023	V2	Updated *ATP7B* additional variants detected