MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Primary Ciliary Dyskinesia Gene Panel

The following applies to PCDGG / Primary Ciliary Dyskinesia 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 test was updated 04/25/2024. 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 on this version or prior versions of this test, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ARMC4 (ODAD2)	NM_018076.5	-	Sensitivity for SNV and CNV detection may be reduced in exon 9 due to homology.
CCDC103	NM_213607.3	-	-
CCDC114 (ODAD1)	NM_144577.4	-	-
CCDC151 (ODAD3)	NM_145045.5	-	-
CCDC39	NM_181426.2	-	-
CCDC40	NM_017950.4	-	-
CCDC65	NM_033124.5	-	-
CCNO	NM_021147.5	-	-
CFAP298	NM_021254.4	-	CNV may not be detected in exon 3.
CFAP300	NM_032930.3	-	-
DNAAF1	NM_178452.6	-	-
DNAAF2	NM_018139.2	-	-
DNAAF3	NM_001256714.1	-	-
DNAAF4	NM_130810.4	-	-
DNAAF5	NM_017802.4	-	-
DNAH1	NM_015512.5	-	-
DNAH11	NM_001277115.2	-	CNV may not be detected in exon 55.
DNAH5	NM_001369.2	-	-
DNAH8	NM_001206927.2	-	-
DNAH9	NM_001372.4	-	-
DNAI1	NM_012144.4	-	-
DNAI2	NM_023036.6	-	-
DNAJB13	NM_153614.3	-	Sensitivity for SNV and CNV detection may be reduced in exon 7. Coverage 5–10x.
DNAL1	NM_031427.4	-	CNV may not be detected in exon 5.
DRC1	NM_145038.5	-	-
FOXJ1	NM_001454.4	-	-
GAS8	NM_001481.3	-	-
LRRC6 (DNAAF11)	NM_012472.6	-	-
MCIDAS	NM_001190787.3	-	-
OFD1	NM_003611.3	-	-

Targeted Genes and Methodology Details for Primary Ciliary Dyskinesia Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
PIH1D3 (DNAAF6)	NM_001169154.1	-	-
RPGR	NM_000328.3	-	-
RSPH1	NM_080860.4	-	-
RSPH3	NM_031924.6	-	-
RSPH4A	NM_001010892.3	-	-
RSPH9	NM_152732.5	-	-
SPAG1	NM_172218.2	-	CNV may not be detected in exon 5.
TTC25 (ODAD4)	NM_031421.5	-	Sequence and CNV analysis of exons 9-12 will not be performed.
ZMYND10	NM_015896.4	-	-

Effective Date	Version	Synopsis of Test Change
04/25/2024	V2	Added note regarding reduced sensitivity for *DNAJB13* exon 7 and *ODAD2* exon 9