MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Phagocytic Disorders and Chronic Granulomatous Disease Gene Panel

The following applies to PHCGD / Phagocytic Disorders and Chronic Granulomatous 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 noncoding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) 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 July 2025 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, 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
CEBPE	NM_001805.3	-	-
CFTR	NM_000492.4	Poly T tract; TG repeat region for 5T alleles only; deletion/duplication analysis; chr7:g.117179041_117179044del (c.870-1113_870-1110del); chr7:g.117199500G>A (c.1393-18G>A); chr7:g.117227784T>A (c.1585-9T>A); chr7:g.117227785G>A (c.1585-8G>A); chr7:g.117218381A>G (c.1585-9412A>G); chr7:g.117229521A>G (c.1680-886A>G); chr7:g.117229524A>G (c.1680-883A>G); chr7:g.117229530G>T (c.1680-877G>T); chr7:g.117246713T>G (c.2909-15T>G); chr7:g.117250260A>T (c.2989-313A>T); chr7:g.117251609A>G (c.3140-26A>G); chr7:g.117251619T>A (c.3140-16T>A); chr7:g.117266272C>G (c.3469-1304C>G); chr7:g.117267864A>G (c.3717+40A>G); chr7:g.117280015C>T (c.3718-2477C>T); chr7:g.117288374A>G (c.3874-4522A>G)	CNV analysis in exon 13 is not performed
CSF2RA	NM_006140.5	-	-
CSF2RB	NM_000395.3	-	-
CTSC	NM_001814.6	chr11:g.88070895G>T (c.-55C>A)	-
CYBA	NM_000101.4	chr16:g.88712620G>C (c.288-15C>G)	-
CYBB	NM_000397.4	chrX:g.37639262 to chrX:g.37639267 (c.-69 to c.-64); chrX:g.37641327_37641330delinsGAA (c.46-14_46-11delinsGAA); chrX:g.37642715_37642731del (c.142-28_142-12del); chrX:g.37642731delinsACCTCTTCTAG (c.142-12delinsACCTCTTCTAG)	-
CYBC1	NM_001033046.4	-	-
FERMT3	NM_031471.6	-	-
G6PD	NM_001042351.3	-	-

Targeted Genes and Methodology Details for Phagocytic Disorders and Chronic Granulomatous Disease Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ITGB2	NM_000211.5	chr21:g.46321660A>C (c.500-12T>G); chr21:g.46320404G>T (c.742-14C>A)	-
MRTFA	NM_020831.6	-	CNV analysis in exon 3 is not performed
NCF1	NM_000265.6	chr7:g.74191615_74191616del (c.75_76del)	CNV analysis is not performed
NCF2	NM_000433.3	-	-
NCF4	NM_000631.5; NM_013416.3	-	-
RAC2	NM_002872.5	-	-
SLC35C1	NM_018389.5	-	-
SLC7A7	NM_001126106.2	-	-
SMARCD2	NM_001098426.2	-	-
WDR1	NM_017491.5	-	-