MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Angioedema and Complement Disorders Gene Panel

The following applies to AECDP / Angioedema and Complement Disorders 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
ANGPT1	NM_001146.5	-	-
C1QA	NM_015991.4	-	-
C1QB	NM_000491.5	-	-
C1QC	NM_172369.5	-	-
C1R	NM_001733.7	Whole gene deletion and duplication performed	CNV analysis in individual exons 1–7 is not performed
C1S	NM_201442.4	-	-
C2	NM_000063.6	chr6:g.31902068_31902095del (c.841_849+19del); chr6:g.31911326_31911347del (c.1567+22_1567+43del)	-
C3	NM_000064.4	-	-
C5	NM_001735.3	-	-
C6	NM_000065.4	-	-
C7	NM_000587.4	chr5:g.40931143T>A (c.63-23T>A)	CNV analysis in exon 14 is not performed
C8A	NM_000562.3	chr1:g.57351588G>A (c.856-12G>A)	-
C8B	NM_000066.4	-	-
C9	NM_001737.5	-	-
CD46	NM_002389.4	-	CNV analysis in exon 2 is not performed
CD55	NM_000574.5	-	-
CD59	NM_203330.2	-	-
CFB	NM_001710.6	-	-
CFD	NM_001928.4	-	-
CFH	NM_000186.4	-	CNV analysis in exon 20 is not performed
CFI	NM_000204.4	-	CNV analysis in exon 7 is not performed
CFP	NM_002621.2	-	-
F12	NM_000505.3	-	-

Targeted Genes and Methodology Details for Angioedema and Complement Disorders Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
KNG1	NM_001102416.3	-	-
MASP2	NM_006610.4	-	-
PLG	NM_000301.3	-	-
SERPING1	NM_000062.3	chr11:g.57365567G>T (c.-22-155G>T); chr11:g.57365720A>C (c.-22-2A>C); chr11:g.57365720A>G (c.-22-2A>G); chr11:g.57365721G>A (c.-22-1G>A); chr11:g.57373471A>G (c.686-12A>G)	-
THBD	NM_000361.3	-	-