MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Atypical Hemolytic Uremic Syndrome / Thrombotic Microangiopathy / Complement 3 Glomerulopathy 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 may 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 (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria. To verify if a specific region/exon/variant is covered by this assay or to confirm transcript version used, contact a laboratory genetic counselor at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ADAMTS13	NM_139025.4	-	-
C3	NM_000064.4	-	-
C5	NM_001735.3	Eculizumab resistance-associated variants: c.2654G>A (p.Arg885His) c.2653C>T (p.Arg885Cys)	Analyses for sequence variants and CNV is not performed for the gene except for the specified variants.
CD46	NM_002389.4	*CD46* (MCPggaac) risk haplotype	-
CFB	NM_001710.6	-	-
CFH	NM_000186.4	*CFH*-H3 gene risk haplotype	Exon 20 CNV and *CFH–CFHR* hybrid alleles may not be detected.
CFHR1	NM_002113.2	-	CNV analysis in exon 1, 4 is not performed
CFHR2	NM_005666.4	-	CNV analysis in exon 1 is not performed
CFHR3	NM_021023.5	-	CNV analysis in exon 5 is not performed
CFHR4	NM_001201551.2	-	CNV analysis in exon 9 is not performed
CFHR5	NM_030787.4	-	-
CFI	NM_000204.4	-	CNV analysis in exon 7 is not performed
DGKE	NM_003647.3	chr17: g.54925466A>G (c.888+40A>G)	-
MMACHC	NM_015506.3	-	-
THBD	NM_000361.3	-	-