COMPLEMENT MEDIATED RENAL DISEASE PANEL DG-5.2.0 (25 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ADAMTS13	100% | 100%	96% | 96%	98% | 98%	Thrombotic thrombocytopenic purpura, hereditary, 274150
C2	100% | 100%	97% | 97%	99% | 99%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	97% | 97%	95% | 95%	98% | 98%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C5	100% | 100%	97% | 97%	99% | 99%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
CD46	100% | 100%	98% | 98%	100% | 100%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	93% | 93%	96% | 96%	99% | 99%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	89% | 89%	98% | 98%	99% | 99%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300
CFB	99% | 99%	97% | 97%	99% | 99%	Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	98% | 98%	98% | 98%	100% | 100%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	91% | 89%	93% | 90%	96% | 94%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR2	73% | 71%	96% | 95%	99% | 99%	
CFHR3	93% | 93%	95% | 94%	96% | 96%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100% | 100%	97% | 97%	99% | 99%	
CFHR5	100% | 100%	98% | 98%	99% | 99%	Nephropathy due to CFHR5 deficiency, 614809

CFI	95% | 95%	98% | 98%	99% | 99%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CFP	98% | 98%	98% | 98%	100% | 100%	Properdin deficiency, X-linked, 312060
DGKE	100% | 100%	98% | 98%	99% | 99%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
EXOSC3	100% | 100%	97% | 97%	99% | 99%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100% | 100%	96% | 96%	99% | 99%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
HSD11B2	100% | 100%	95% | 95%	98% | 98%	Apparent mineralocorticoid excess, 218030
IQGAP1	100% | 100%	97% | 97%	100% | 100%	
MMACHC	100% | 100%	96% | 96%	99% | 99%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
PLG	89% | 89%	97% | 97%	99% | 99%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
THBD	100% | 100%	90% | 90%	99% | 99%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
VTN	100% | 100%	96% | 96%	99% | 99%	