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COMPLEMENT MEDIATED RENAL DISEASE PANEL DG-5.1.0 (25 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
C2	100%	100%	100%	100%	99.5%	C2 deficiency, 217000;{Macular degeneration, age-related, 14, reduced risk of}, 615489
C3	97%	97%	100%	99.9%	99.2%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378
C5	100%	100%	100%	100%	99.8%	C5 deficiency, 609536;[Eculizumab, poor response to], 615749
CD46	100%	100%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CD55	96.6%	93.7%	100%	100%	99.5%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CD59	89.3%	89.3%	100%	100%	100%	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300

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CFB	100%	100%	100%	100%	99.4%	Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400
CFHR1	98.3%	96.4%	91.7%	87.3%	76.3%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR2	76.4%	76.4%	100%	99.8%	99.5%	
CFHR3	98%	96.9%	93.2%	89.3%	78.5%	{Macular degeneration, age-related, reduced risk of}, 603075;{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFHR4	100%	100%	100%	99.6%	98.7%	
CFHR5	100%	100%	100%	100%	99.8%	Nephropathy due to CFHR5 deficiency, 614809

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CFI	98.6%	95.9%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CFP	100%	100%	98.8%	88.3%	70.4%	Properdin deficiency, X-linked, 312060
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
EXOSC3	100%	100%	100%	100%	99.7%	Pontocerebellar hypoplasia, type 1B, 614678
EXOSC5	100%	100%	100%	100%	98.6%	Cerebellar ataxia, brain abnormalities, and cardiac conduction defects, 619576
HSD11B2	100%	100%	100%	100%	99.4%	Apparent mineralocorticoid excess, 218030
IQGAP1	100%	100%	100%	100%	99.4%	
MMACHC	100%	100%	100%	100%	99.5%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
PLG	89.3%	89.3%	100%	100%	99.8%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
THBD	100%	100%	100%	99.8%	98.7%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926

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VTN	100%	100%	100%	100%	99.6%	

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors