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AGE-RELATED MACULAR DEGENERATION PANEL DG-5.1.0 (16 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
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
C9	99.3%	99.3%	100%	99.9%	99.6%	C9 deficiency, 613825;{Macular degeneration, age-related, 15, susceptibility to}, 615591
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

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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
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
CTNNA1	94.8%	94.8%	100%	100%	99.8%	Macular dystrophy, patterned, 2, 608970
PRDM13	100%	100%	100%	99.9%	98.7%	Pontocerebellar hypoplasia, type 17, 619909;Cerebellar dysfunction, impaired intellectual development, and hypogonadotropic hypogonadism, 619761

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PRPH2	100%	100%	100%	100%	99.4%	Macular dystrophy, patterned, 1, 169150;Choroidal dystrophy, central areolar 2, 613105;Retinitis punctata albescens, 136880;Leber congenital amaurosis 18, 608133;Macular dystrophy, vitelliform, 3, 608161;Retinitis pigmentosa 7 and digenic form, 608133
TIMP3	84%	84%	100%	100%	99.4%	Sorsby fundus dystrophy, 136900
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