

# WES HEMOSTATIC/THROMBOTIC DISORDERS DG 2.16

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
A2M	107.3	100.0	99.3	614036
ABCG5	139.5	100.0	99.9	210250
ABCG8	133.9	99.7	98.2	210250
ACBD5	145.1	99.6	98.0	-
ACTN1	131.5	100.0	100.0	615193
ACVRL1	113.6	100.0	98.4	600376
ADAMTS13	103.8	98.1	95.2	274150
ANKRD26	83.3	95.3	90.1	188000
ANO6	133.3	99.8	98.0	262890
AP3B1	112.1	99.5	96.5	608233
AP3D1	125.2	98.4	97.9	617050
ARPC1B	139.6	100.0	100.0	617718
BLOC1S3	67.4	100.0	99.9	614077
BLOC1S6	101.1	99.2	95.1	614171
BRAF	72.5	92.4	80.2	613706
C3	141.6	100.0	99.4	612925
CALR	111.8	98.1	91.7	187950
CBL	126.0	97.3	97.0	613563
CD36	119.8	99.7	99.1	608404
CD46	125.5	99.7	98.7	612922
CFB	119.4	100.0	99.9	612924
CFH	155.4	99.4	97.9	235400
CFHR1	158.4	93.6	90.8	235400
CFHR3	98.4	93.8	91.6	235400
CFI	139.0	99.5	97.0	612923

CHST14	160.6	99.9	98.9	601776
COL1A1	141.0	99.8	98.4	130000
COL3A1	99.2	99.3	96.8	130050
COL5A1	136.4	99.9	98.9	130000
COL5A2	100.2	99.9	99.4	130000
CTLA4	141.0	100.0	100.0	152700
CYCS	61.0	99.1	93.1	612004
DGKE	127.8	99.8	98.3	615008
DIAPH1	101.7	99.9	99.6	-
DNASE1	168.5	100.0	100.0	152700
DTNBP1	113.4	99.8	97.9	614076
ENG	128.4	99.9	98.7	187300
EPHB2	190.2	98.1	98.1	-
ETV6	148.3	100.0	99.4	616216
F10	173.6	99.8	99.1	227600
F11	126.7	100.0	99.9	612416
F12	151.3	99.9	99.5	234000
F13A1	112.9	100.0	99.6	188050;613225
F13B	105.8	98.6	92.9	613235
F2	128.1	99.8	97.1	613679;188050
F2RL3	130.4	100.0	100.0	-
F5	145.5	99.5	97.7	227400;188055
F7	162.0	100.0	100.0	227500
F8	106.0	99.4	97.2	306700
F9	109.5	99.9	98.4	306900;300807
FBN1	137.1	100.0	99.8	154700
FCGR2A	164.6	100.0	100.0	152700
FCGR2B	119.7	99.9	96.9	152700
FCGR2C	148.2	98.2	97.8	188030
FERMT3	144.9	100.0	99.9	612840

FGA	137.0	99.3	97.3	616004;202400
FGB	136.7	99.7	98.2	616004;202400
FGG	126.4	99.8	98.0	616004;202400
FLI1	153.8	99.3	97.9	188025;147791
FLNA	142.7	100.0	99.9	300049;300048
FYB1	96.3	99.5	96.8	273900
GATA1	92.9	99.9	98.2	300367;300835;314050
GATA2	115.0	100.0	99.0	614172;614038
GBA	169.8	100.0	100.0	231005;231000;230900;230800
GDF2	142.4	100.0	100.0	615506
GFI1B	170.9	99.9	98.2	187900
GGCX	101.2	100.0	99.4	277450;610842
GNE	113.8	100.0	99.3	-
GP1BA	136.8	98.7	95.7	153670;231200;177820
GP1BB	68.6	94.5	83.1	231200
GP6	123.2	100.0	99.7	614201
GP9	134.3	99.9	98.3	231200
HABP2	109.3	100.0	99.4	188050
HOXA11	88.3	100.0	98.0	605432
HPS1	115.8	100.0	99.9	203300
HPS3	132.7	99.9	98.8	614072
HPS4	128.1	100.0	99.9	614073
HPS5	122.8	99.9	98.7	614074
HPS6	164.6	99.9	97.8	614075
HRG	126.1	95.0	94.2	613116
ITGA2	134.1	99.7	97.9	614200
ITGA2B	124.4	99.9	98.9	187800;273800
ITGB3	112.8	100.0	99.8	187800;273800
JAK2	103.5	97.6	95.0	614521
KLKB1	132.3	100.0	99.5	612423

KNG1	150.7	100.0	100.0	228960
KRAS	67.2	99.4	97.3	609942
LMAN1	144.3	99.8	99.4	227300
LYST	136.3	99.4	97.8	214500
LZTR1	143.6	100.0	99.7	616564
MASTL	134.9	100.0	100.0	188000
MCFD2	91.4	97.6	91.0	613625
MECOM	131.2	100.0	99.6	616738
MLPH	97.4	99.7	97.2	609227
MPIG6B	119.0	100.0	99.8	617441
MPL	125.8	100.0	99.8	601977;604498
MTHFR	114.9	98.2	96.4	188050
MYH9	128.5	99.6	98.5	155100;603622
MYO5A	109.0	99.7	98.6	214450
NBEA	125.1	91.9	90.2	-
NBEAL2	166.0	100.0	99.5	139090
NRAS	145.5	100.0	100.0	613224
ORAI1	198.9	99.8	98.2	-
P2RY12	186.2	100.0	100.0	609821
PLA2G4A	124.5	99.8	99.3	-
PLA2G7	120.9	99.8	98.6	614278
PLAT	92.0	100.0	99.2	612348
PLAU	99.6	100.0	99.0	601709
PLG	93.4	87.8	86.8	217090
PRKACG	204.5	100.0	100.0	616176
PROC	142.6	100.0	100.0	176860;612304
PROS1	95.8	97.7	92.7	612336;614514
PROZ	130.0	99.8	98.8	614024
PTGS1	137.6	100.0	99.4	605735
PTPN11	78.3	98.6	90.7	163950

PTPN22	127.8	99.6	95.9	152700
PTPRJ	137.4	97.7	96.3	-
RAB27A	126.1	100.0	99.8	607624
RAF1	108.3	100.0	99.9	611553
RASGRP2	102.5	100.0	99.7	615888
RBM8A	87.4	99.8	97.4	274000
RIT1	139.2	100.0	100.0	615355
RUNX1	84.6	99.6	96.3	601399
SERPINC1	117.9	100.0	100.0	613118
SERPIND1	134.8	100.0	99.9	612356
SERPINE1	134.0	100.0	100.0	613329
SERPINF2	151.6	100.0	99.9	262850
SH2B3	108.4	99.9	97.6	187950
SLFN14	172.0	100.0	100.0	616913
SOS1	102.0	99.6	97.4	610733
SOS2	99.7	99.7	97.9	616559
SRC	124.1	100.0	99.5	616937
STIM1	120.7	99.8	96.8	185070
STXBP2	100.2	83.7	80.4	613101
TALDO1	148.2	100.0	99.6	606003
TBX1	101.2	93.0	86.9	192430
TBXA2R	119.6	97.2	93.9	614009
TBXAS1	128.8	100.0	100.0	614158;231095
THBD	181.1	100.0	100.0	614486
THPO	97.3	100.0	99.7	187950
TNXB	105.6	99.5	95.8	606408
TPM4	59.5	96.9	86.3	-
TREX1	233.4	100.0	100.0	152700
TUBB1	150.0	100.0	100.0	613112
VIPAS39	114.7	100.0	99.9	613404

VKORC1	146.5	100.0	99.9	607473
VPS33B	107.2	100.0	99.9	208085
VWF	98.2	99.9	99.1	193400;277480;613554
WAS	70.4	94.2	83.6	313900;301000
WIPF1	89.1	100.0	99.1	614493

*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.*

*Median Coverage describes the average number of reads seen across 50 exomes.*

*% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.*

*% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.*

*Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*