

# WES HEMOSTATIC/THROMBOTIC DISORDERS DG 3.4

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
A2M	168.8	100.0	100.0	614036
ABCG5	179.3	100.0	100.0	210250
ABCG8	179.4	100.0	100.0	210250
ACBD5	170.3	100.0	100.0	-
ACTB	292.9	100.0	100.0	243310
ACTN1	183.5	100.0	100.0	615193
ACVRL1	186.6	100.0	100.0	600376
ADAMTS13	204.5	100.0	100.0	274150
ANKRD26	137.0	97.2	97.2	188000
ANO6	158.0	100.0	100.0	262890
AP3B1	145.9	100.0	100.0	608233
AP3D1	180.5	100.0	100.0	617050
ARPC1B	180.2	100.0	100.0	617718
BLOC1S3	216.9	100.0	100.0	614077
BLOC1S5	163.8	100.0	100.0	619172
BLOC1S6	153.4	100.0	100.0	614171
BRAF	174.2	100.0	100.0	613706
C3	177.7	100.0	100.0	612925
CALR	202.9	100.0	100.0	187950
CBL	197.3	100.0	100.0	613563
CD36	145.3	100.0	100.0	608404
CD46	152.3	100.0	100.0	612922
CDC42	137.2	100.0	100.0	616737
CFB	235.1	100.0	100.0	612924
CFH	165.5	100.0	100.0	235400

CFI	163.9	100.0	100.0	612923
CHST14	217.4	100.0	100.0	601776
COL1A1	207.6	100.0	100.0	130000
COL3A1	167.4	100.0	100.0	130050
COL5A1	189.4	100.0	100.0	130000
COL5A2	153.3	100.0	100.0	130000
CTLA4	195.3	100.0	100.0	152700
CYCS	165.3	100.0	100.0	612004
DGKE	179.3	100.0	100.0	615008
DIAPH1	155.5	100.0	100.0	124900
DNASE1	192.2	100.0	100.0	152700
DTNBP1	154.9	100.0	100.0	614076
ENG	192.7	100.0	100.0	187300
EPHB2	206.0	100.0	99.9	618462
ETV6	216.4	100.0	100.0	616216
F10	201.4	100.0	100.0	227600
F11	179.8	100.0	100.0	612416
F12	223.0	100.0	100.0	234000;610618
F13A1	177.9	100.0	100.0	613225;188050
F13B	157.0	100.0	100.0	613235
F2	176.5	100.0	100.0	188050;613679
F2RL3	242.0	100.0	100.0	-
F5	179.4	100.0	100.0	188055;227400
F7	218.3	100.0	100.0	227500
F8	165.0	100.0	100.0	306700
F9	151.8	100.0	99.8	300807;306900
FBN1	164.0	100.0	100.0	154700
FERMT3	221.3	100.0	100.0	612840
FGA	184.0	100.0	100.0	202400;616004
FGB	171.2	100.0	100.0	202400;616004

FGG	174.1	100.0	100.0	202400;616004
FLI1	209.5	100.0	100.0	188025;147791
FLNA	208.6	100.0	100.0	300049;300048
FYB1	174.3	100.0	100.0	273900
GATA1	226.4	100.0	100.0	314050;300835;300367
GATA2	247.0	100.0	100.0	614038;614172
GBA	231.0	100.0	100.0	230900;231005;230800;231000
GDF2	224.3	100.0	100.0	615506
GFI1B	225.6	100.0	100.0	187900
GGCX	184.6	100.0	100.0	610842;277450
GNE	182.9	100.0	100.0	-
GP1BA	216.0	100.0	100.0	231200;153670;177820
GP1BB	165.8	100.0	100.0	231200
GP6	264.0	99.0	96.3	614201
GP9	231.6	100.0	100.0	231200
HABP2	174.9	100.0	100.0	188050
HOXA11	229.8	100.0	100.0	605432
HPS1	182.7	100.0	100.0	203300
HPS3	161.3	100.0	100.0	614072
HPS4	190.3	100.0	100.0	614073
HPS5	156.2	100.0	100.0	614074
HPS6	232.9	100.0	100.0	614075
HRG	193.0	100.0	100.0	613116
IKZF5	194.8	100.0	100.0	619130
ITGA2	156.8	100.0	100.0	614200
ITGA2B	194.8	100.0	100.0	187800;273800
ITGB3	174.8	100.0	100.0	187800;273800
JAK2	156.9	100.0	100.0	614521
KDSR	175.3	100.0	100.0	617526
KLKB1	169.0	100.0	100.0	612423

KNG1	167.4	100.0	100.0	228960;228960
KRAS	158.7	100.0	100.0	609942
LMAN1	170.9	100.0	100.0	227300
LYST	160.3	100.0	100.0	214500
LZTR1	182.8	100.0	100.0	616564
MASTL	154.5	100.0	100.0	188000
MCFD2	171.1	100.0	100.0	613625
MECOM	197.1	100.0	100.0	616738
MLPH	166.7	100.0	100.0	609227
MPIG6B	297.3	100.0	100.0	617441
MPL	219.1	100.0	100.0	601977;604498
MTHFR	178.1	100.0	100.0	188050
MYH9	198.1	100.0	100.0	603622;155100
MYO5A	153.6	100.0	100.0	214450
NBEA	157.1	100.0	100.0	-
NBEAL2	197.4	100.0	100.0	139090
NFE2	219.2	100.0	100.0	No OMIM phenotype
NRAS	184.4	100.0	100.0	613224
OCRL	146.6	100.0	100.0	300555;309000
ORAI1	216.6	100.0	100.0	-
P2RY12	167.0	100.0	100.0	609821
PIGA	168.2	100.0	100.0	300818
PLA2G4A	160.0	100.0	100.0	618372
PLA2G7	146.4	100.0	100.0	614278
PLAT	180.7	100.0	100.0	612348
PLAU	185.8	100.0	100.0	601709
PLG	170.5	100.0	100.0	217090
PRKACG	220.3	100.0	100.0	616176
PROC	191.3	100.0	100.0	612304;176860
PROS1	143.7	98.4	98.4	612336;614514

PROZ	200.8	100.0	100.0	614024
PTGS1	186.9	100.0	100.0	605735
PTPN11	185.6	100.0	100.0	163950
PTPN22	159.6	100.0	100.0	152700
PTPRJ	175.8	100.0	100.0	-
RAB27A	126.3	100.0	100.0	607624
RAF1	181.9	100.0	100.0	611553
RASGRP2	194.7	100.0	100.0	615888
RBM8A	174.7	100.0	100.0	274000
RIT1	176.5	100.0	100.0	615355
RNU4ATAC				616651
RUNX1	251.5	100.0	100.0	601399
SERPINC1	195.3	100.0	100.0	613118
SERPIND1	210.5	100.0	100.0	612356
SERPINE1	187.5	100.0	100.0	613329
SERPINF2	278.4	100.0	100.0	262850
SH2B3	204.4	100.0	100.0	187950
SLFN14	169.7	100.0	100.0	616913
SOS1	144.0	100.0	100.0	610733
SOS2	153.7	100.0	100.0	616559
SRC	217.3	100.0	100.0	616937
STAB2	161.2	100.0	100.0	No OMIM phenotype
STIM1	182.8	100.0	100.0	185070
STXBP2	248.1	99.8	98.7	613101
TALDO1	172.4	100.0	100.0	606003
TBX1	158.0	98.1	95.9	192430
TBXA2R	224.9	100.0	99.7	614009
TBXAS1	168.9	100.0	100.0	614158;231095
THBD	268.2	100.0	100.0	614486
THPO	181.5	100.0	100.0	187950

TNXB	273.2	100.0	100.0	606408
TPM4	158.1	100.0	100.0	-
TREX1	263.2	100.0	100.0	152700
TUBB1	215.6	100.0	100.0	613112
VIPAS39	140.0	100.0	100.0	613404
VKORC1	172.1	93.1	93.0	607473
VPS33B	151.1	100.0	100.0	208085
VWF	184.1	100.0	100.0	193400;613554;277480
WAS	174.2	100.0	100.0	301000;313900
WIPF1	194.6	100.0	100.0	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.*

*TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.*

*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 no value for coverage are non protein coding genes.*

*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: September 1st, 2021.*