

WES HEMOSTATIC/THROMBOTIC DISORDERS DG 3.00

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
A2M	128.1	99.9	99.6	614036
ABCG5	173.9	100.0	100.0	210250
ABCG8	150.2	99.1	97.3	210250
ACBD5	181.1	100.0	99.2	-
ACTB	74.3	99.7	96.1	243310
ACTN1	146.0	100.0	100.0	615193
ACVRL1	122.9	100.0	98.9	600376
ADAMTS13	98.0	97.1	93.8	274150
ANKRD26	94.5	95.0	89.3	188000
ANO6	169.0	99.9	98.7	262890
AP3B1	140.4	99.2	95.8	608233
AP3D1	125.3	99.8	98.6	617050
ARPC1B	131.7	100.0	100.0	617718
BLOC1S3	40.2	98.5	81.3	614077
BLOC1S6	122.0	99.9	97.8	614171
BRAF	80.6	91.0	81.1	613706
C3	153.5	99.9	99.2	612925
CALR	124.4	94.8	89.1	187950
CBL	152.5	97.3	97.1	613563
CD36	147.2	99.7	98.7	608404
CD46	151.5	99.9	99.4	612922
CDC42	121.8	97.9	90.9	616737
CFB	139.7	100.0	100.0	612924
CFH	194.1	99.9	99.0	235400
CFHR1	201.6	96.4	94.9	235400

CFHR3	111.8	94.0	92.2	235400
CFI	169.4	99.2	96.8	612923
CHST14	147.4	99.9	98.9	601776
COL1A1	154.6	99.9	98.6	130000
COL3A1	120.8	99.6	97.6	130050
COL5A1	159.9	98.8	98.0	130000
COL5A2	121.3	100.0	99.5	130000
CTLA4	162.1	100.0	100.0	152700
CYCS	78.6	99.1	94.9	612004
DGKE	130.4	99.8	98.1	615008
DIAPH1	119.5	99.8	99.0	-
DNASE1	167.2	100.0	99.9	152700
DTNBP1	126.7	99.8	98.7	614076
ENG	130.6	99.6	96.0	187300
EPHB2	200.8	98.1	98.1	-
ETV6	185.3	100.0	99.9	616216
F10	194.4	99.8	99.1	227600
F11	157.7	100.0	100.0	612416
F12	127.6	99.9	98.8	234000
F13A1	144.6	100.0	100.0	613225;188050
F13B	131.0	98.7	93.5	613235
F2	133.6	99.9	97.9	188050;613679
F2RL3	89.5	100.0	100.0	-
F5	172.8	99.4	98.5	188055;227400
F7	168.7	100.0	100.0	227500
F8	123.3	97.3	96.1	306700
F9	130.9	99.6	97.6	300807;306900
FBN1	169.7	100.0	99.9	154700
FCGR2A	195.0	100.0	100.0	152700
FCGR2B	135.8	99.5	95.4	152700

FCGR2C	170.9	98.9	98.6	188030
FERMT3	142.1	100.0	100.0	612840
FGA	169.3	99.1	97.2	202400;616004
FGB	165.0	99.8	99.1	202400;616004
FGG	175.8	99.7	98.2	202400;616004
FLI1	198.2	99.5	98.2	188025;147791
FLNA	138.5	100.0	99.9	300049;300048
FYB1	120.7	99.4	97.0	273900
GATA1	108.4	99.8	98.4	314050;300835;300367
GATA2	119.1	100.0	98.3	614038;614172
GBA	202.3	100.0	100.0	230900;231005;230800;231000
GDF2	149.2	100.0	100.0	615506
GFI1B	171.2	98.5	96.7	187900
GGCX	119.0	100.0	99.9	610842;277450
GNE	132.7	100.0	99.7	-
GP1BA	169.1	98.6	95.9	231200;153670;177820
GP1BB	32.0	72.9	59.6	231200
GP6	149.1	100.0	100.0	614201
GP9	122.0	96.5	89.3	231200
HABP2	134.0	100.0	99.9	188050
HOXA11	95.6	97.1	87.5	605432
HPS1	123.1	100.0	100.0	203300
HPS3	165.7	99.7	97.5	614072
HPS4	142.1	100.0	100.0	614073
HPS5	142.5	100.0	99.7	614074
HPS6	172.2	97.1	88.9	614075
HRG	152.5	95.0	94.2	613116
IKZF5	176.8	100.0	100.0	-
ITGA2	161.8	99.6	98.1	614200
ITGA2B	133.0	99.7	97.8	187800;273800

ITGB3	122.8	100.0	99.4	187800;273800
JAK2	127.4	98.1	95.8	614521
KDSR	182.9	100.0	99.5	617526
KLKB1	157.5	100.0	99.5	612423
KNG1	167.1	100.0	100.0	228960
KRAS	84.3	99.5	96.9	609942
LMAN1	166.8	99.8	99.2	227300
LYST	169.3	99.6	98.3	214500
LZTR1	136.8	100.0	99.9	616564
MASTL	170.0	100.0	100.0	188000
MCFD2	85.7	99.5	96.9	613625
MECOM	157.3	100.0	99.9	616738
MLPH	106.3	100.0	98.8	609227
MPIG6B	144.5	100.0	100.0	617441
MPL	140.2	100.0	99.5	601977;604498
MTHFR	117.2	97.3	96.0	188050
MYH9	134.9	100.0	99.3	603622;155100
MYO5A	136.2	99.8	98.9	214450
NBEA	146.7	92.0	90.6	-
NBEAL2	166.4	99.4	99.4	139090
NRAS	185.6	100.0	100.0	613224
ORAI1	183.1	99.1	96.4	-
P2RY12	243.0	100.0	100.0	609821
PIGA	85.7	93.8	86.7	300818
PLA2G4A	147.3	99.9	99.4	618372
PLA2G7	151.3	99.9	99.0	614278
PLAT	98.0	100.0	99.1	612348
PLAU	114.2	100.0	99.8	601709
PLG	117.3	87.8	87.5	217090
PRKACG	191.9	100.0	99.9	616176

PROC	143.2	100.0	100.0	612304;176860
PROS1	112.8	96.7	92.1	612336;614514
PROZ	147.7	100.0	99.8	614024
PTGS1	151.6	100.0	99.8	605735
PTPN11	100.3	99.1	93.7	163950
PTPN22	148.3	99.5	97.1	152700
PTPRJ	168.3	97.7	97.2	-
RAB27A	163.4	100.0	100.0	607624
RAF1	125.8	100.0	100.0	611553
RASGRP2	105.0	99.7	97.3	615888
RBM8A	109.3	99.8	97.9	274000
RIT1	178.3	100.0	100.0	615355
RNU4ATAC				616651
RUNX1	85.5	99.3	94.9	601399
SERPINC1	142.2	100.0	100.0	613118
SERPIND1	161.9	100.0	100.0	612356
SERPINE1	151.7	100.0	100.0	613329
SERPINF2	174.0	100.0	99.8	262850
SH2B3	106.6	99.4	95.1	187950
SLFN14	222.4	100.0	100.0	616913
SOS1	123.6	99.8	98.4	610733
SOS2	123.5	100.0	99.2	616559
SRC	121.9	100.0	99.8	616937
STIM1	126.2	99.8	98.0	185070
STXBP2	104.1	82.1	79.7	613101
TALDO1	151.8	100.0	97.9	606003
TBX1	89.7	87.0	77.5	192430
TBXA2R	102.6	97.6	93.8	614009
TBXAS1	151.1	100.0	100.0	614158;231095
THBD	139.3	100.0	99.7	614486

THPO	83.3	81.4	81.0	187950
TNXB	103.2	99.1	93.7	606408
TPM4	55.2	82.9	70.4	-
TREX1	248.4	100.0	100.0	152700
TUBB1	163.4	100.0	100.0	613112
VIPAS39	133.8	100.0	100.0	613404
VKORC1	147.5	100.0	100.0	607473
VPS33B	128.1	100.0	100.0	208085
VWF	107.9	99.8	98.6	193400;613554;277480
WAS	81.1	95.9	85.3	301000;313900
WIPF1	111.8	100.0	99.9	614493

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.

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