WES HEMOSTATIC/THROMBOTIC DISORDERS DG 3.5

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
A2M	144.4	100.0	100.0	614036
ABCG5	144.4	100.0	100.0	210250
ABCG8	119.8	100.0	100.0	210250
ACBD5	138.7	100.0	100.0	-
ACTB	238.4	100.0	100.0	243310
ACTN1	131.0	100.0	100.0	615193
ACVRL1	127.1	100.0	100.0	600376
ADAMTS13	137.4	100.0	100.0	274150
ANKRD26	146.6	97.2	97.2	188000
ANO6	146.5	100.0	100.0	262890
AP3B1	148.8	100.0	100.0	608233
AP3D1	125.0	100.0	100.0	617050
ARPC1B	117.2	100.0	100.0	617718
BLOC1S3	184.0	100.0	100.0	614077
BLOC1S5	142.0	100.0	100.0	619172
BLOC1S6	151.7	100.0	100.0	614171
BRAF	139.7	100.0	100.0	613706
C3	121.6	100.0	100.0	612925
CALR	127.2	100.0	100.0	187950
CBL	127.1	100.0	100.0	613563
CD36	157.2	100.0	99.7	608404
CD46	159.2	100.0	100.0	612922
CDC42	156.6	100.0	100.0	616737
CFB	129.9	100.0	100.0	612924
CFH	165.1	100.0	100.0	235400

CFI	155.7	100.0	100.0	610984, 612923, 615439
CHST14	124.8	100.0	100.0	601776
COL1A1	120.1	100.0	100.0	130000
COL3A1	146.7	100.0	100.0	130050
COL5A1	121.9	100.0	100.0	130000
COL5A2	145.7	100.0	100.0	130000
CTLA4	131.1	100.0	100.0	152700
CYCS	184.4	100.0	100.0	612004
DGKE	153.0	100.0	100.0	615008
DIAPH1	135.6	100.0	100.0	124900
DNASE1	141.2	100.0	100.0	152700
DTNBP1	125.3	100.0	100.0	614076
ENG	119.1	100.0	100.0	187300
EPHB2	137.1	100.0	99.9	618462
ETV6	126.9	100.0	100.0	616216
F10	128.2	100.0	100.0	227600
F11	148.4	100.0	100.0	612416
F12	159.0	100.0	100.0	234000;610618
F13A1	136.3	100.0	100.0	613225;188050
F13B	159.6	99.8	98.6	613235
F2	125.1	100.0	100.0	188050;613679
F2RL3	131.8	100.0	100.0	-
F5	143.4	100.0	100.0	188055;227400
F7	140.5	100.0	100.0	227500
F8	108.2	100.0	99.9	306700
F9	115.9	100.0	100.0	300807;306900
FBN1	149.7	100.0	100.0	154700
FERMT3	122.6	100.0	100.0	612840
FGA	137.0	100.0	100.0	202400;616004
FGB	140.7	100.0	100.0	202400;616004

FGG	158.4	100.0	100.0	202400;616004
FLI1	133.3	100.0	100.0	188025;147791
FLNA	105.8	100.0	99.9	300049;300048
FYB1	141.4	100.0	100.0	273900
GATA1	94.0	100.0	100.0	314050;300835;300367
GATA2	129.9	100.0	100.0	614038;614172
GBA	136.2	100.0	100.0	230900;231005;230800;231000
GDF2	120.2	100.0	100.0	615506
GFI1B	117.9	100.0	100.0	187900
GGCX	139.6	100.0	100.0	610842;277450
GNE	126.6	100.0	100.0	-
GP1BA	115.6	100.0	100.0	231200;153670;177820
GP1BB	160.3	100.0	100.0	231200
GP6	116.9	99.1	96.2	614201
GP9	135.7	100.0	100.0	231200
HABP2	130.9	100.0	100.0	188050
HOXA11	139.8	100.0	100.0	605432
HPS1	136.7	100.0	100.0	203300
HPS3	147.0	100.0	100.0	614072
HPS4	133.6	100.0	100.0	614073
HPS5	138.5	100.0	100.0	614074
HPS6	126.4	100.0	100.0	614075
HRG	134.3	100.0	100.0	613116
IKZF5	133.6	100.0	100.0	619130
ITGA2	151.8	100.0	99.9	614200
ITGA2B	124.2	100.0	100.0	187800;273800
ITGB3	128.3	100.0	100.0	187800;273800
JAK2	148.4	100.0	100.0	614521
KDSR	148.6	100.0	100.0	617526
KLKB1	154.0	100.0	100.0	612423

KNG1	143.8	100.0	100.0	228960;228960
KRAS	161.6	100.0	100.0	609942
LMAN1	153.8	100.0	100.0	227300
LYST	142.7	100.0	99.8	214500
LZTR1	134.2	100.0	100.0	616564
MASTL	144.6	100.0	100.0	188000
MCFD2	134.7	100.0	100.0	613625
MECOM	145.4	100.0	100.0	616738
MLPH	123.3	100.0	100.0	609227
MPIG6B	128.7	100.0	100.0	617441
MPL	123.2	100.0	100.0	601977;604498
MTHFR	123.4	100.0	100.0	188050
MYH9	127.0	100.0	100.0	603622;155100
MYO5A	136.3	100.0	100.0	214450
NBEA	147.0	99.7	99.2	-
NBEAL2	123.6	100.0	100.0	139090
NFE2	124.3	100.0	100.0	No OMIM phenotype
NRAS	139.4	100.0	100.0	613224
OCRL	112.9	100.0	100.0	300555;309000
ORAI1	123.0	100.0	100.0	-
P2RY12	151.2	100.0	100.0	609821
PIGA	108.6	100.0	100.0	300818
PLA2G4A	151.7	100.0	100.0	618372
PLA2G7	159.4	100.0	100.0	614278
PLAT	124.1	100.0	100.0	612348
PLAU	127.8	100.0	100.0	601709
PLG	143.0	100.0	100.0	217090
PRKACG	146.3	100.0	100.0	616176
PROC	131.3	100.0	100.0	612304;176860
PROS1	149.8	100.0	100.0	612336;614514

PROZ	138.4	100.0	100.0	614024
PTGS1	124.3	100.0	100.0	605735
PTPN11	144.8	100.0	100.0	163950
PTPN22	149.6	100.0	100.0	152700
PTPRJ	125.3	100.0	100.0	-
RAB27A	137.5	100.0	100.0	607624
RAF1	133.3	100.0	100.0	611553
RASGRP2	130.8	100.0	100.0	615888
RBM8A	141.6	100.0	100.0	274000
RIT1	136.9	100.0	100.0	615355
RNU4ATAC				616651
RUNX1	151.6	100.0	100.0	601399
SERPINC1	123.1	100.0	100.0	613118
SERPIND1	137.2	100.0	100.0	612356
SERPINE1	118.8	100.0	100.0	613329
SERPINF2	113.9	100.0	100.0	262850
SH2B3	148.6	100.0	100.0	187950
SLFN14	136.9	100.0	100.0	616913
SOS1	145.6	100.0	100.0	610733
SOS2	141.1	100.0	100.0	616559
SRC	133.9	100.0	100.0	616937
STAB2	131.5	100.0	100.0	No OMIM phenotype
STIM1	132.3	100.0	100.0	185070
STXBP2	165.7	100.0	99.9	613101
TALDO1	122.3	100.0	100.0	606003
TBX1	117.8	97.7	95.5	192430
TBXA2R	136.4	99.9	99.0	614009
TBXAS1	134.3	100.0	100.0	614158;231095
THBD	151.2	100.0	100.0	614486
THPO	120.4	100.0	100.0	187950

TNXB	166.8	100.0	100.0	606408
TPM4	112.6	100.0	100.0	-
TREX1	142.6	100.0	100.0	152700
TUBB1	146.9	100.0	100.0	613112
VIPAS39	129.6	100.0	100.0	613404
VKORC1	149.2	97.8	92.7	607473
VPS33B	128.9	100.0	100.0	208085
VWF	120.1	100.0	100.0	193400;613554;277480
WAS	85.1	100.0	98.8	301000;313900
WIPF1	131.4	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.

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