

HEMOSTATIC/THROMBOTIC DISORDERS DG 2.15 (145 genes)

Releasedate: 31-01-2019

<i>Gene</i>	<i>Median Coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
A2M	119.2	99.5	98.5	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
ABCG5	145.2	100	99.2	Sitosterolemia, 210250
ABCG8	148.4	99.2	96.6	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ACBD5	145.3	97.8	96	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACTN1	143.6	100	99.9	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	122.7	99.9	98	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	99.9	96.3	91.7	Thrombotic thrombocytopenic purpura, familial, 274150
ANKRD26	81	88.7	76.8	Thrombocytopenia 2, 188000
ANO6	137.4	98	92.9	Scott syndrome, 262890
AP3B1	95	97.8	90.2	Hermansky-Pudlak syndrome 2, 608233
AP3D1	121	98.1	97.8	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	126.4	100	99.9	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
BLOC1S3	28.7	88.7	65.3	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.2	98.7	91.3	?Hermansky-pudlak syndrome 9, 614171
BRAF	74.4	87.6	77.2	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706

C3	145.5	100	99.7	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CALR	113.5	99.9	97.4	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	129.8	96.9	95.7	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CD36	123.3	99.2	95.7	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD46	115.1	97.8	93.2	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CFB	147.1	100	100	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	183.2	98.7	95.3	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	236.5	95.8	94.2	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101	90.6	85.6	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	145.5	96.6	92.8	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CHST14	165.6	95.7	93.3	Ehlers-Danlos syndrome, musculocontractural type 1, 601776

COL1A1	134.9	98.1	96.3	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL3A1	104.3	97.8	92.3	Ehlers-Danlos syndrome, vascular type, 130050
COL5A1	114.3	97.7	95	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	89.1	99.5	97.4	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	193.9	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CYCS	72.1	99.6	95	Thrombocytopenia 4, 612004
DGKE	142.3	99.5	95.2	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DIAPH1	120.7	99.3	97.8	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	198.8	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	115.2	99.3	95.1	Hermansky-Pudlak syndrome 7, 614076
ENG	128.8	97.4	93.6	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ETV6	140.1	100	99.9	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
F10	185.1	99	98.3	Factor X deficiency, 227600
F11	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	111.4	100	99.5	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	147.3	100	99.4	Factor XIII A deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	113.5	96.6	87.6	Factor XIII B deficiency, 613235

F2	124.2	99.8	98.1	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	102.6	100	99.9	No OMIM phenotype Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259)
F5	173.5	99	97.3	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	166.6	100	98.5	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	118	99.5	98.1	Hemophilia A, 306700
F9	144.5	99.2	95.9	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FCGR2A	244.9	100	100	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	176.9	99.9	97.2	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	206.8	98.1	97.6	Thrombocytopenic purpura, autoimmune, 188030
FERMT3	122.4	100	98.9	Leukocyte adhesion deficiency, type III, 612840
FGA	157	99	96.6	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	190.8	99.7	97.9	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400

FGG	137	99.3	96.5	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FLI1	189.6	99.4	97.7	Bleeding disorder, platelet-type, 21, 617443
FLNA	138.1	100	99.5	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620 Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FYB1	92.3	97.7	91.9	Thrombocytopenia 3, 273900
GATA1	83.5	99.6	95.7	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GDF2	163.2	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	170.2	98.2	95.7	Bleeding disorder, platelet-type, 17, 187900
GGCX	115.3	100	99.7	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GNE	153.7	100	99.8	Nonaka myopathy, 605820 Sialuria, 269921
GP1BA	153	97	94.3	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	34.5	74.2	64.3	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	136.5	100	100	Bleeding disorder, platelet-type, 11, 614201

GP9	123.3	96.6	89.3	Bernard-Soulier syndrome, type C, 231200
HABP2	136.9	100	99.6	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HOXA11	86.3	88	78.5	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	117.8	100	99.3	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99.6	96.4	Hermansky-Pudlak syndrome 3, 614072
HPS4	141.9	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99.9	98.7	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.1	91	84.3	Hermansky-Pudlak syndrome 6, 614075
HRG	178.3	95	94.3	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
ITGA2	136.5	97.7	95.4	?Glycoprotein Ia deficiency, 614200
ITGA2B	107.2	99.6	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGB3	142.2	99.3	97.4	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
JAK2	90.6	95.9	94.1	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
KLKB1	143.4	99.6	96.7	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	193.3	100	100	[High molecular weight kininogen deficiency], 228960 [Kininogen deficiency], 228960

KRAS	64.7	99.9	98.7	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LMAN1	129.8	99.3	94.4	Combined factor V and VIII deficiency, 227300
LYST	134.6	97.8	93.9	Chediak-Higashi syndrome, 214500
LZTR1	134	100	99.4	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MASTL	130.5	100	99.3	?Thrombocytopenia-2, 188000
MCFD2	101.7	99.9	98.8	Factor V and factor VIII, combined deficiency of, 613625
MECOM	143.4	100	99.6	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	99.1	99.6	95.9	Griscelli syndrome, type 3, 609227
MPIG6B	85.4	100	99.3	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	126.1	98.4	97.2	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	125.3	99.5	97.4	Griscelli syndrome, type 1, 214450

NBEA	128.4	90.8	89.4	No OMIM phenotype Lentaigne et al., Blood 2016 127:2814-2823 Inherited platelet disorders: toward DNA-based diagnosis
NBEAL2	172.7	99.5	99.3	Gray platelet syndrome, 139090
NRAS	188.4	100	100	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
ORAI1	237.3	93.8	89.8	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
P2RY12	186.2	100	100	Bleeding disorder, platelet-type, 8, 609821
PLA2G4A	134.3	99.9	98.8	Phospholipase A2, group IV A, deficiency of, 0
PLA2G7	125.6	99.9	97.2	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAT	97.8	100	99.8	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	111.4	99.8	98.4	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	115.4	87.8	87	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	217.6	100	99.9	?Bleeding disorder, platelet-type, 19, 616176
PROC	138.5	99.7	97.2	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	101.4	96.8	91.4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	131.3	99.9	98.2	[Protein Z deficiency], 614024
PTGS1	148	99.6	99	No OMIM phenotype

PTPN11	103.1	97.9	92.5	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	134.5	98	91.9	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
RAB27A	143.9	100	99.9	Griscelli syndrome, type 2, 607624
RAF1	127.3	100	99.7	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	97.5	99.9	98.6	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RIT1	165.6	100	100	Noonan syndrome 8, 615355
RUNX1	92	97.2	89.7	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SERPINC1	143.1	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	181.8	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	155	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF2	143.8	99.9	99.3	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	97.5	90.7	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SLFN14	193.1	100	100	Bleeding disorder, platelet-type, 20, 616913
SOS1	94.3	96.7	90.3	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	97.1	98.5	92.8	Noonan syndrome 9, 616559
SRC	105.2	99.8	97.3	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
STIM1	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STXBP2	102.3	88.9	83.8	Hemophagocytic lymphohistiocytosis, familial, 5, 613101

TALDO1	130.5	100	99.9	Transaldolase deficiency, 606003
TBX1	75.3	77.1	67.4	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBXA2R	83.7	97.6	92.9	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
THBD	108.2	99.8	97.8	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THPO	88.2	100	100	Thrombocythemia 1, 187950
TNXB	96.4	98.4	91.4	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	51.9	92.1	79.5	No OMIM phenotype Pleines et al.,2017. Mutations in tropomyosin 4 underlie a rare form of human macrothrombocytopenia. J Clin Invest. 2017 127:814-829.
TREX1	242.4	100	100	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TUBB1	186.5	100	100	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	144.6	100	100	Arthrogyrosis, renal dysfunction, and cholestasis 2, 613404
VKORC1	162.1	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	138.3	100	100	Arthrogyrosis, renal dysfunction, and cholestasis 1, 208085
VWF	120.9	100	99.6	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	77.5	100	99.2	?Wiskott-Aldrich syndrome 2, 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: December 31st, 2018.

This list is accurate for panel version DG 2.15

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