HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.9 / DG 2.10 (132 genes)

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A2M	134.5	99%	99%	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
ABCG5	167.3	100%	99%	Sitosterolemia, 210250
ABCG8	166.5	99%	97%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ACTN1	164.8	100%	99%	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	138.6	100%	98%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	119.3	96%	93%	Thrombotic thrombocytopenic purpura, familial, 274150
ANKRD26	99.3	93%	85%	Thrombocytopenia 2, 188000
ANO6	155.8	98%	96%	Scott syndrome, 262890
AP3B1	128.1	99%	95%	Hermansky-Pudlak syndrome 2, 608233
BLOC1S3	56	99%	95%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	111.3	99%	96%	Hermansky-pudlak syndrome 9, 614171
BRAF	86.4	91%	82%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
C3	167.5	100%	99%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CALR	113.7	99%	95%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	146	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CD36	150.9	99%	98%	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia]

				{Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD46	157.5	99%	95%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CFB	23	84%	51%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	216.6	99%	97%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	202.7	92%	90%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	101.2	88%	83%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	198	98%	97%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
COL3A1	124.9	98%	93%	Ehlers-Danlos syndrome, type IV, 130050
CTLA4	212.7	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	89.9	99%	97%	Thrombocytopenia 4, 612004
DGKE	160	99%	97%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DIAPH1	131.4	99%	98%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	230.9	100%	100%	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	127.3	99%	97%	Hermansky-Pudlak syndrome 7, 614076
ENG	148.5	99%	96%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ETV6	152.2	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216

F10	187.1	99%	97%	Factor X deficiency, 227600
F11	170.5	100%	99%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	131.4	100%	99%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	182.8	100%	99%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	156.6	97%	92%	Factor XIIIB deficiency, 613235
F2	138.3	100%	99%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	111.6	100%	99%	No OMIM phenotype Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259)
F5	196.8	99%	98%	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	178.3	100%	99%	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	138.1	99%	98%	Hemophilia A, 306700
F9	156	99%	97%	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FCGR2A	237.8	100%	100%	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	157.9	99%	96%	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	208.9	99%	99%	Thrombocytopenic purpura, autoimmune, 188030

FERMT3	139.2	99%	98%	Leukocyte adhesion deficiency,type III,612840
FGA	178.9	99%	97%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	198.5	99%	97%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGG	178.7	99%	98%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FLI1	199.3	99%	97%	No OMIM phenotype Platelet dense granule secretion defect,excessive bleeding (Stockley (2013) Blood 122,4090)
FLNA	160.7	100%	99%	Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 FG syndrome 2, 300321 Frontometaphyseal dysplasia, 305620 Heterotopia, periventricular, 300049 Heterotopia, periventricular, ED variant, 300537 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
GATA1	100.8	99%	94%	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
GFI1B	204.5	100%	100%	Bleeding disorder, platelet-type, 17, 187900
GGCX	124.5	100%	99%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GP1BA	175.2	97%	95%	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	41.4	80%	69%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	140.1	100%	100%	Bleeding disorder, platelet-type, 11, 614201
GP9	100.8	98%	93%	Bernard-Soulier syndrome, type C, 231200
HABP2	148.6	100%	99%	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HOXA11	86.4	97%	87%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	136.6	100%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS3	175.7	100%	99%	Hermansky-Pudlak syndrome 3, 614072
HPS4	151.7	100%	99%	Hermansky-Pudlak syndrome 4, 614073
HPS5	160.7	99%	98%	Hermansky-Pudlak syndrome 5, 614074
HPS6	156.4	97%	90%	Hermansky-Pudlak syndrome 6, 614075
HRG	187.4	97%	95%	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
ITGA2	180.5	98%	96%	?Glycoprotein Ia deficiency, 614200
ITGA2B	136.6	99%	97%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related
ITGB3	151.9	99%	99%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446
JAK2	113.7	96%	94%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600800
KLKB1	194.2	99%	97%	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	202.7	100%	100%	[High molecular weight kininogen deficiency], 228960 [Kininogen deficiency], 228960
KRAS	89.6	99%	99%	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	156.5	99%	98%	Combined factor V and VIII deficiency, 227300
LYST	171	98%	96%	Chediak-Higashi syndrome, 214500
LZTR1	166.5	100%	99%	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MASTL	148.8	99%	99%	?Thrombocytopenia-2, 188000
MCFD2	114.3	100%	99%	Factor V and factor VIII, combined deficiency of, 613625
MECOM	179.9	100%	99%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	110.2	99%	96%	Griscelli syndrome, type 3, 609227
MPL	163.9	99%	97%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	161.7	100%	100%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}
MYH9	152.2	99%	98%	Deafness, autosomal dominant 17, 603622 Epstein syndrome, 153650 Fechtner syndrome, 153640 Macrothrombocytopenia and progressive sensorineural deafness, 600208 May-Hegglin anomaly, 155100 Sebastian syndrome, 605249
MYO5A	138.4	99%	98%	Griscelli syndrome, type 1, 214450
NBEA	150.4	91%	89%	No OMIM phenotype Autism, idiopathic (Castermans (2003) J Med Genet 40, 352) ?Schizophrenia (Fromer (2014) Nature 506, 179)

				?Obesity, extreme (Mariman (2015) Physiol Genomics 47,225) ?Tetralogy of Fallot (Silversides (2012) PloS Genet 8)
NBEAL2	189.5	99%	99%	Gray platelet syndrome, 139090
NRAS	203.3	100%	100%	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
P2RX1	134.3	100%	99%	Bleeding disorder due to P2RX1 defect, somatic, 609821
P2RY12	240.6	100%	100%	Bleeding disorder, platelet-type, 8, 609821
PLA2G4A	170.7	100%	99%	Phospholipase A2, group IV A, deficiency of
PLA2G7	158.1	100%	99%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAT	110.2	100%	99%	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	123.7	99%	99%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	138.6	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	280.8	100%	100%	?Bleeding disorder, platelet-type, 19, 616176
PROC	142.6	99%	97%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	104.9	97%	93%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	152.4	100%	99%	[Protein Z deficiency], 614024
PTGS1	170	99%	99%	No OMIM phenotype
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950

PTPN22	162.4	99%	94%	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
RAB27A	171.6	100%	100%	Griscelli syndrome, type 2, 607624
RAF1	144.9	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	109.7	99%	98%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	109.4	99%	97%	Thrombocytopenia-absent radius syndrome, 274000
RIT1	190.3	100%	100%	Noonan syndrome 8, 615355
RUNX1	111.4	97%	92%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SELP	149.4	100%	99%	{Atopy, susceptibility to}, 147050
SERPINC1	152.9	100%	100%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	157.3	100%	100%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	171	100%	99%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}
SERPINF2	166.3	100%	99%	Alpha-2-plasmin inhibitor deficiency,262850
SH2B3	113.1	95%	84%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SLFN14	230.7	100%	100%	Bleeding disorder,platelet-type,20,616913
SOS1	120.1	98%	95%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	115.8	99%	97%	Noonan syndrome 9, 616559
SRC	124.7	99%	98%	Colon cancer, advanced, somatic ?Thrombocytopenia 6,616937
STIM1	144	99%	97%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
STXBP2	144.3	99%	98%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TALDO1	153.4	100%	99%	Transaldolase deficiency, 606003
TBX1	90.6	79%	70%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400

				Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBXA2R	76.3	96%	91%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	170.1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
THBD	129.5	99%	98%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THPO	95.6	100%	100%	Thrombocythemia 1, 187950
TREX1	302.9	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	191	100%	100%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	156.9	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	161.5	100%	99%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	142.9	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	129.6	100%	99%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAS	72	89%	80%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	88.4	100%	99%	?Wiskott-Aldrich syndrome 2, 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 : April 14th 2017
This list is accurate for panel version DG 2.9 and DG 2.10

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