

# HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.11

## (133 genes)

<i>Gene</i>	<i>Median</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
A2M	119.2	99	98	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
ABCG5	145.4	100	99	Sitosterolemia, 210250
ABCG8	148.7	99	96	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ACTN1	143.9	100	99	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	123	99	98	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	100	96	91	Thrombotic thrombocytopenic purpura, familial, 274150
ANKRD26	79.7	89	77	Thrombocytopenia 2, 188000
ANO6	137.2	98	92	Scott syndrome, 262890
AP3B1	95	97	90	Hermansky-Pudlak syndrome 2, 608233
BLOC1S3	28.7	88	64	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	97.3	98	91	Hermansky-pudlak syndrome 9, 614171
BRAF	74.3	87	77	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	145.8	100	99	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CALR	113.6	99	97	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	133.2	99	98	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785

CD36	123.2	99	95	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	115.2	97	93	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CFB	33.4	23	23	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	183.1	98	95	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	234.8	95	94	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	103	92	87	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	152.5	98	96	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
COL3A1	104.2	97	92	Ehlers-Danlos syndrome, type IV, 130050
CTLA4	194.1	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	94	Thrombocytopenia 4, 612004
DGKE	142.3	99	95	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DIAPH1	120.5	99	97	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	201.3	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	113.9	99	95	Hermansky-Pudlak syndrome 7, 614076
ENG	128.9	97	93	Telangiectasia, hereditary hemorrhagic, type 1, 187300

ETV6	140.3	100	99	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
F10	185.4	99	98	Factor X deficiency, 227600
F11	155	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	111.6	100	99	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	147.4	100	99	Factor XIII A deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	113.4	96	87	Factor XIII B deficiency, 613235
F2	124.4	99	98	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	103	100	99	No OMIM phenotype Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259)
F5	173.6	99	97	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	164.4	100	98	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	118.8	99	98	Hemophilia A, 306700
F9	145.5	99	95	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FCGR2A	245.1	100	100	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700

FCGR2B	166.1	99	97	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	208	98	97	Thrombocytopenic purpura, autoimmune, 188030
FERMT3	122.6	100	98	Leukocyte adhesion deficiency, type III, 612840
FGA	157.2	99	96	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	190.7	99	97	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGG	136.9	99	96	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FLI1	189.8	99	97	No OMIM phenotype Platelet dense granule secretion defect, excessive bleeding (Stockley (2013) Blood 122,4090)
FLNA	139.2	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	84.2	99	95	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	189.6	100	100	Bleeding disorder, platelet-type, 17, 187900
GGCX	115.4	100	99	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450

GP1BA	153.1	97	94	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	64	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	136.8	100	100	Bleeding disorder, platelet-type, 11, 614201
GP9	123.1	96	89	Bernard-Soulier syndrome, type C, 231200
HABP2	136.3	100	99	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050
HOXA11	86.4	87	78	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	118	100	99	Hermansky-Pudlak syndrome 1, 203300
HPS3	135.2	99	96	Hermansky-Pudlak syndrome 3, 614072
HPS4	144.5	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	133	99	98	Hermansky-Pudlak syndrome 5, 614074
HPS6	139.2	91	84	Hermansky-Pudlak syndrome 6, 614075
HRG	178.5	95	94	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
ITGA2	136.3	97	95	?Glycoprotein Ia deficiency, 614200
ITGA2B	119.9	99	97	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related
ITGB3	138.9	99	97	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion Thrombocytopenia, neonatal alloimmune {Myocardial infarction, susceptibility to}, 608446
JAK2	90.5	95	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	143.5	99	96	Fletcher factor (prekallikrein) deficiency, 612423

KNG1	193.7	100	100	[High molecular weight kininogen deficiency], 228960 [Kininogen deficiency], 228960
KRAS	64.6	99	98	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	129.9	99	94	Combined factor V and VIII deficiency, 227300
LYST	134.6	97	93	Chediak-Higashi syndrome, 214500
LZTR1	134.3	100	99	Noonan syndrome 10, 616564 {Schwannomatosis-2, susceptibility to}, 615670
MASTL	130.5	100	99	?Thrombocytopenia-2, 188000
MCFD2	101.5	99	98	Factor V and factor VIII, combined deficiency of, 613625
MECOM	143.3	100	99	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	95.2	99	95	Griscelli syndrome, type 3, 609227
MPL	135.2	99	97	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	131.3	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	130.8	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	124.8	99	97	Griscelli syndrome, type 1, 214450

NBEA	128.3	90	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	173	99	99	Gray platelet syndrome, 139090
NRAS	188.2	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	115.3	100	99	Bleeding disorder due to P2RX1 defect, somatic, 609821
P2RY12	185.8	100	100	Bleeding disorder, platelet-type, 8, 609821
PLA2G4A	134.2	99	98	Phospholipase A2, group IV A, deficiency of
PLA2G7	124.4	99	97	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAT	98	100	99	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	111.5	99	98	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	115.4	87	86	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	218.1	100	99	?Bleeding disorder, platelet-type, 19, 616176
PROC	140.4	99	97	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	101.1	96	91	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	131.4	99	98	[Protein Z deficiency], 614024
PTGS1	153.1	99	98	No OMIM phenotype

PTPN11	103.4	97	92	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	134.4	98	91	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
RAB27A	144.1	100	99	Griscelli syndrome, type 2, 607624
RAF1	127.4	100	99	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	97.5	99	98	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	106.6	100	99	Thrombocytopenia-absent radius syndrome, 274000
RIT1	165.5	100	100	Noonan syndrome 8, 615355
RUNX1	91.6	94	86	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SELP	125.8	100	99	{Atopy, susceptibility to}, 147050
SERPINC1	143.2	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	181.9	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	155.2	100	100	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}
SERPINF2	144.1	99	99	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	97.5	90	79	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SLFN14	193	100	100	Bleeding disorder, platelet-type, 20, 616913
SOS1	95.5	96	90	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	97	98	92	Noonan syndrome 9, 616559
SRC	105.5	99	97	Colon cancer, advanced, somatic ?Thrombocytopenia 6, 616937
STIM1	127.2	100	99	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070



STXBP2	125.3	98	97	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TALDO1	130.6	100	99	Transaldolase deficiency, 606003
TBX1	75.5	77	67	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBXA2R	83.9	97	93	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	140.3	100	100	Ghosal hematodiaphyseal syndrome, 231095 ?Thromboxane synthase deficiency, 614158
THBD	108.4	99	97	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THPO	88.1	100	100	Thrombocythemia 1, 187950
TREX1	242.7	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	144.7	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	162.4	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	138.4	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	121.2	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	66.6	88	78	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	77.5	100	99	?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 : April 14th, 2017.*

*This list is accurate for panel version DG 2.11*

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

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