HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.17 (156 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
A2M	109.5	99.9%	99.2%	No OMIM disease ID
ABCG5	147.1	100.0%	99.9%	Sitosterolemia 2, 618666
ABCG8	146.0	99.9%	98.9%	Sitosterolemia 1, 210250
ACBD5	144.7	99.8%	98.1%	No OMIM Disease ID
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTN1	140.2	100.0%	100.0%	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	125.9	100.0%	98.8%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	115.6	98.0%	96.0%	Thrombotic thrombocytopenic purpura, familial, 274150
ANKRD26	81.5	94.9%	89.1%	Thrombocytopenia 2, 188000
ANO6	132.9	99.8%	98.5%	Scott syndrome, 262890
AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	135.1	98.5%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	150.6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
BLOC1S3	79.6	100.0%	100.0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	103.0	99.3%	92.1%	?Hermansky-pudlak syndrome 9, 614171
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
C3	153.6	100.0%	99.7%	C3 deficiency, 613779
CALR	118.8	98.9%	92.5%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	131.1	97.4%	97.1%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785

CD36	115.0	99.9%	99.0%	Platelet glycoprotein IV deficiency, 608404
CD46	126.4	100.0%	99.1%	No OMIM disease ID
CDC42	89.8	98.1%	90.1%	Takenouchi-Kosaki syndrome, 616737
CFB	125.5	100.0%	100.0%	?Complement factor B deficiency, 615561
CFH	148.8	99.4%	97.4%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	156.0	94.1%	92.0%	No OMIM disease ID
CFHR3	89.5	93.2%	91.0%	No OMIM disease ID
CFI	137.6	99.2%	96.8%	Complement factor I deficiency, 610984
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	154.6	99.9%	99.1%	Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL3A1	99.6	99.4%	97.0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	146.8	100.0%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100.3	99.9%	99.3%	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	146.7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100
CYCS	60.4	99.5%	93.7%	Thrombocytopenia 4, 612004
DGKE	132.2	100.0%	98.3%	Nephrotic syndrome, type 7, 615008
DIAPH1	104.4	100.0%	99.8%	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, 124900
DNASE1	184.5	100.0%	100.0%	No OMIM disease ID
DTNBP1	118.8	99.8%	97.5%	Hermansky-Pudlak syndrome 7, 614076
ENG	137.2	100.0%	99.4%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	205.6	98.1%	98.1%	?Bleeding disorder, platelet-type, 22, 618462
ETV6	157.6	99.9%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
F10	191.3	99.9%	99.2%	Factor X deficiency, 227600
F11	129.2	100.0%	99.9%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	171.1	100.0%	99.7%	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	116.5	100.0%	99.8%	Factor XIIIA deficiency, 613225

F13B	100.3	98.3%	90.9%	Factor XIIIB deficiency, 613235
F2	137.0	99.8%	97.7%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F2RL3	155.1	100.0%	100.0%	No OMIM Disease ID
F5	148.2	99.4%	97.6%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055
F7	179.1	100.0%	100.0%	Factor VII deficiency, 227500
F8	105.6	99.4%	97.3%	Hemophilia A, 306700
F9	110.5	99.9%	97.9%	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
FBN1	138.3	100.0%	99.7%	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FCGR2A	173.8	100.0%	100.0%	No OMIM Disease ID
FCGR2B	131.3	99.7%	97.4%	No OMIM Disease ID
FCGR2C	161.6	99.4%	99.1%	No OMIM Disease ID
FERMT3	161.2	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FGA	142.0	99.4%	97.5%	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGB	138.9	99.9%	98.4%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGG	124.2	99.9%	98.3%	Hypofibrinogenemia, congenital, 202400 Hypodysfibrinogenemia, 616004 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FLI1	163.7	99.4%	98.0%	Bleeding disorder, platelet-type, 21, 617443
FLNA	156.4	100.0%	99.9%	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120

				Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FYB1	96.5	99.5%	96.0%	Thrombocytopenia 3, 273900
GATA1	102.0	99.9%	99.0%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	128.7	100.0%	99.7%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	180.2	100.0%	100.0%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GDF2	157.3	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	187.0	100.0%	99.1%	Bleeding disorder, platelet-type, 17, 187900
GGCX	105.2	100.0%	99.6%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GNE	115.7	100.0%	99.3%	Sialuria, 269921 Nonaka myopathy, 605820
GP1BA	144.7	99.1%	96.5%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	85.6	96.1%	86.6%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	135.4	100.0%	100.0%	Bleeding disorder, platelet-type, 11, 614201
GP9	160.5	100.0%	99.5%	Bernard-Soulier syndrome, type C, 231200
HABP2	111.6	100.0%	99.3%	No OMIM Disease ID
HOXA11	96.7	99.8%	97.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	125.7	100.0%	100.0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	133.9	99.9%	98.2%	Hermansky-Pudlak syndrome 3, 614072
HPS4	135.2	100.0%	100.0%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122.5	99.9%	98.9%	Hermansky-Pudlak syndrome 5, 614074

HPS6	183.5	100.0%	99.2%	Hermansky-Pudlak syndrome 6, 614075
HRG	130.2	94.7%	94.2%	Thrombophilia due to HRG deficiency, 613116
IKZF5	171.8	100.0%	100.0%	No OMIM Disease ID
ITGA2	131.4	99.7%	97.7%	No OMIM Disease ID
ITGA2B	137.3	100.0%	99.4%	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGB3	119.9	100.0%	100.0%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
JAK2	100.6	97.1%	95.0%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
KDSR	165.5	100.0%	99.5%	Erythrokeratodermia variabilis et progressiva 4, 617526
KLKB1	132.4	99.9%	99.2%	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	153.1	100.0%	100.0%	No OMIM Disease ID
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LMAN1	147.0	99.9%	98.5%	Combined factor V and VIII deficiency, 227300
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
LZTR1	157.2	100.0%	99.9%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MASTL	135.3	100.0%	100.0%	No OMIM Disease ID
MCFD2	97.5	97.7%	91.3%	Factor V and factor VIII, combined deficiency of, 613625
MECOM	133.4	100.0%	99.8%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738

MLPH	104.1	99.8%	97.9%	Griscelli syndrome, type 3, 609227
MPIG6B	130.3	100.0%	100.0%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	134.4	100.0%	99.9%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MTHFR	124.2	98.5%	96.7%	Homocystinuria due to MTHFR deficiency, 236250
MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	109.9	99.7%	98.5%	Griscelli syndrome, type 1, 214450
NBEA	121.3	91.9%	90.4%	No OMIM Disease ID
NBEAL2	182.9	100.0%	99.7%	Gray platelet syndrome, 139090
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
ORAI1	226.3	100.0%	99.0%	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
P2RY12	187.4	100.0%	100.0%	Bleeding disorder, platelet-type, 8, 609821
PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PLA2G4A	121.8	100.0%	99.4%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	117.7	100.0%	99.1%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAT	97.8	100.0%	99.8%	No OMIM disease ID
PLAU	105.2	100.0%	99.3%	Quebec platelet disorder, 601709
PLG	95.7	87.8%	86.7%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	226.7	100.0%	100.0%	?Bleeding disorder, platelet-type, 19, 616176
PROC	158.3	100.0%	100.0%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	92.6	97.3%	92.4%	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PROZ	140.7	99.8%	97.7%	No OMIM disease ID

PTGS1	149.6	100.0%	99.6%	No OMIM Disease ID
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN22	124.5	99.5%	95.6%	No OMIM disease ID
PTPRJ	139.8	98.2%	96.5%	Colon cancer, somatic, 114500
RAB27A	123.3	100.0%	99.6%	Griscelli syndrome, type 2, 607624
RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RASGRP2	111.5	100.0%	99.8%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	89.0	100.0%	98.2%	Thrombocytopenia-absent radius syndrome, 274000
RIT1	142.5	100.0%	100.0%	Noonan syndrome 8, 615355
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
RUNX1	92.9	99.9%	97.4%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SERPINC1	122.9	100.0%	100.0%	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	139.0	100.0%	100.0%	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	142.1	100.0%	99.9%	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF2	167.5	100.0%	100.0%	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	122.5	99.8%	98.8%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SLFN14	174.8	100.0%	100.0%	Bleeding disorder, platelet-type, 20, 616913
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	98.0	99.7%	97.7%	Noonan syndrome 9, 616559
SRC	135.9	100.0%	99.8%	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STXBP2	110.2	84.1%	80.8%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TALDO1	158.9	100.0%	99.8%	Transaldolase deficiency, 606003

TBX1	114.2	93.7%	88.3%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBXA2R	138.4	98.2%	94.7%	No OMIM disease ID
TBXAS1	135.5	100.0%	100.0%	Ghosal hematodiaphyseal syndrome, 231095
THBD	208.2	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486
THPO	102.6	100.0%	99.9%	Thrombocythemia 1, 187950
TNXB	119.2	99.8%	97.6%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	64.9	97.4%	88.2%	No OMIM Disease ID
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TUBB1	164.6	100.0%	100.0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	114.4	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	161.2	100.0%	100.0%	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VPS33B	111.7	100.0%	100.0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	105.1	100.0%	99.3%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAS	75.4	95.3%	84.4%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WIPF1	95.7	100.0%	99.3%	?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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors