HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ABCG5	121.1	100%	100%	Sitosterolemia,210250
ABCG8	137.9	99%	96%	Sitosterolemia,210250 Gallbladder disease 4,611465
ACTN1	132.9	100%	99%	Bleeding disorder,platelet-type,15,615193
ANKRD26	80.6	91%	80%	Thrombocytopenia 2,188000
ANO6	125.7	97%	94%	Scott syndrome,262890
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2,608233
BLOC1S3	38.5	99%	84%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	89.9	99%	84%	Hermansky-Pudlak syndrome 9,614171
CALR	96.4	99%	95%	Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
CD36	124.4	100%	99%	Platelet glycoprotein IV deficiency,608404 {Coronary heart disease,susceptibility to,7},610938 {Malaria,cerebral,reduced risk of},61162
COL3A1	100.5	89%	84%	Ehlers-Danlos syndrome, type IV,130050
CYCS	68.2	99%	98%	Thrombocytopenia 4,612004
DTNBP1	100.5	100%	95%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500
ETV6	114.9	100%	100%	Leukemia,acute myeloid,somatic,601626 Thrombocytopenia 5,616216
F10	153.9	98%	96%	Factor X deficiency,227600
F11	140.7	100%	97%	Factor XI deficiency,612416
F12	96.2	100%	98%	Factor XII deficiency,234000 Angioedema,hereditary,type III,610618
F13A1	164.5	100%	99%	Factor XIII deficiency,613225 {Myocardial infarction,protection against},608446 {Venous thrombosis,protection against},188050
F13B	141.4	95%	85%	Factor XIIIB deficiency,613235

F2	106.3	100%	97%	Dysprothrombinemia,613679 Hypoprothrombinemia,613679 Thrombophilia due to thrombin defect,188050 {Pregnancy loss,recurrent,susceptibility to,2},614390 {Stroke,ischemic,susceptibility to},601367
F5	176.1	99%	97%	Factor V deficiency,227400 Thrombophilia due to activated prtein C resistance,188055 {Budd-Chiari syndrome},600880 {Pregnancy loss,recurrent,susceptibility to,1},614389 {Stroke,ischemic,susceptibility to},601367
F7	138.6	100%	96%	Factor VII deficiency,227500 {Myocardial infarction,decreased susceptibility to},608446
F8	79.5	98%	96%	Hemophilia A,306700
F9	102.4	95%	86%	Hemophilia B,306900 Thrombophilia,X-linked,due to factor IX defect},300807 {Warfarin sensitivity},122700
FERMT3	103.7	100%	97%	Leukocyte adhesion deficiency,type III,612840
FGA	149.6	97%	95%	Afibrinogenemia,congenital,202400 Amyloidosis,familial visceral,105200 Dysfibrinogenemia,congenital,616004 Hypodysfibrinogenemia,congenital,616004
FGB	168	100%	98%	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004 Hypofibrinogenemia,congenital,202400
FGG	137.8	100%	98%	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004
FLI1	173.6	97%	97%	No OMIM disease ID Platelet dense granule secretion defect,excessive bleeding (Stockley (2013) Blood 122,4090)

FLNA	85.1	99%	97%	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	49.6	97%	90%	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GFI1B	141.8	100%	100%	Bleeding disorder, platelet-type, 17,187900
GGCX	104.8	99%	96%	Psuedoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors,combined deficiency of,1,277450
GP1BA	122.6	99%	95%	Bernard-Soulier syndrome, type A1 (recessive),231200 Bernard-Soulier syndrome, type A2 (dominant),153670 von Willebrand disease,platelet-type,177820 {Nonarteric anterior ischemic optic neuropathy,susceptibility to},258660
GP1BB	37	86%	65%	Bernard-Soulier syndrome,type B,231200 Giant platelet disorder,isolated,231200
GP6	101	100%	99%	Bleeding disorder,platelet-type,11,614201
GP9	66.9	99%	95%	Bernard-Soulier syndrome,type C,231200
HABP2	136.9	100%	99%	{?Thyroid cancer,nonmedullary,5},616535 {Venous thromboembolism,susceptibility to},188050
HOXA11	85.6	82%	79%	Radioulnar synostosis with amegakaryocytic thrombocytopenia,605432
HPS1	102.9	100%	98%	Hermansky-Pudlak syndrome 1,203300
HPS3	138	100%	95%	Hermansky-Pudlak syndrome 3,614072
HPS4	130.4	100%	98%	Hermansky-Pudlak syndrome 4,614073
HPS5	137.7	99%	98%	Hermansky-Pudlak syndrome 5,614074
HPS6	100.4	100%	92%	Hermansky-Pudlak syndrome 6,614075

HRG	153.1	97%	94%	Thrombophilia due to elevated HRG,613116 Thrombophilia due to HRG deficiency,613116
ITGA2	143.4	98%	97%	?glycoprotein Ia deficiency,614200
ITGA2B	99	98%	92%	Bleeding disorder,platelet-type,16,autosomal dominant,187800 Glanzmann thrombasthenia,273800 Thrombocytopenia,neonatal alloimmune,BAK antigen related
ITGB3	124.8	100%	97%	Bleeding disorder,platelet-type 16,autosomal dominant,187800 Glanzmann thrombasthenia,273800 Purpura,posttransfusion Thrombocytopenia,neonatal alloimmune {Myocardial infarction,susceptibility to},608446
KLKB1	152.3	97%	92%	Fletcher factor (prekallikrein) deficiency,612423
LMAN1	128.7	99%	87%	Combined factor V and VIII deficiency,227300
LYST	129.6	97%	92%	Chediak-Higashi syndrome,214500
MASTL	121.6	99%	98%	?Thrombocytopenia 2,188000
MCFD2	104.9	100%	100%	Factor V and factor VIII,combined deficiency of,613625
MLPH	83.1	97%	94%	Griscelli syndrome type 3,609227
MPL	127.5	98%	92%	Myelofibrosis with myeloid metaplasia,somatic,254450 Thrombocythemia 2,601977 Thrombocytopenia,congenital amegakaryocytic,604498
MTHFR	134.9	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	123.8	99%	95%	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	122.5	98%	95%	Griscelli syndrome type 1,214450
NBEAL2	147.3	99%	97%	Gray platelet syndrome,139090
P2RX1	114.5	100%	98%	Bleeding disorder due to P2RX1 defect,somatic,609821

P2RY12	168	100%	100%	Bleeding disorder, platelet-type 8,609821
PLAT	93.2	100%	100%	Hyperfibrinolysis,familial,due to increased release of PLAT, 612348 Thrombophilia,familial,due to decreased release of PLAT,612348
PLAU	104.4	100%	98%	Quebec platelet disorder,601709 {Alzheimer disease,late-onset,susceptibility to},104300
PLG	114	87%	87%	Dysplasminogenemia,217090 Plasminogen deficiency, type I,217090
PRKACG	218.2	100%	100%	?Bleeding disorder,platelet-type,19,616176
PROC	116.9	99%	95%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	77.7	93%	89%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive,614514
PROZ	120.7	100%	99%	[Protein Z deficiency], 614024
PTGS1	136.2	99%	99%	No OMIM disease ID
RAB27A	177.8	100%	100%	Griscelli syndrome,type 2,607624
RASGRP2	89.2	98%	95%	?Bleeding disorder,platelet-type,18,615888
RBM8A	96.4	98%	94%	Thrombocytopenia-absent radius syndrome,274000
RUNX1	106.5	96%	92%	Leukemia,acute myeloid,601626 Platelet disorder,familial,with associated myeloid malignancy,601399
SELP	124.9	100%	99%	{Atopy,susceptibility to},147050
SERPINC1	128.2	100%	100%	Thrombophilia due to antithrombin III deficiency,613118
SERPIND1	151.3	100%	100%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	118.8	100%	100%	Plasminogen activator inhibitor-1 deficiency,613329 {Transcription of plasminogen activator inhibitor,modulator of}
SERPINF2	115.5	100%	98%	Alpha-2-plasmin inhibitor deficiency,262850
SH2B3	79.5	84%	70%	Erythrocytosis,somatic,133100 Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
STIM1	125.1	99%	95%	Immunodeficiency 10,612783 Myopathy,tubular aggregate,1,160565 Stormorken syndrome,185070
STXBP2	107.6	99%	94%	Hemophagocytic lymphohistiocytosis,familial,5,613101

TBX1	70.9	73%	59%	Conotruncal anomaly face syndrome,217095 DiGeorge syndrome,188400 Tetralogy of Fallot,187500 Velocardiofacial syndrome,192430
TBXA2R	53.6	92%	86%	{Bleeding disorder,platelet-type,13,susceptibility to},614009
TBXAS1	133.8	100%	100%	?Thromboxane synthase deficiency,614158 Ghosal hematodiaphyseal syndrome,231095
THBD	85.5	100%	91%	Thrombophilia due to thrombomodulin defect,614486 {Hemolytic uremic syndrome,atypical,susceptibility to,6}
THPO	74.3	100%	100%	Thrombocythemia 1,187950
VIPAS39	135.4	100%	100%	Arthrogryposis,renal dysfunction and cholestasis 2,613404
VKORC1	132.8	100%	100%	Vitamin K-dependent clotting factors,combined deficiency of,2,607473 Warfarin resistance,122700
VPS33B	132.2	100%	99%	Arthrogryposis,renal dysfunction,and cholestasis 1,208085
VWF	107.4	99%	98%	von Willebrand disease, type 1,193400 von Willebrand disease,type 2A,2B,2M and 2N,613554 von Willebrand disease,type 3,277480
WAS	35.9	81%	62%	Neutropenia,severe congenital,X-linked,300299 Thrombocytopenia,X-linked,313900 Wiskott-Aldrich syndrome,301000
WIPF1	69.4	100%	97%	?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 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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