HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 2.4.x

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ABCG5	120.9	99.60%	92.70%	Sitosterolemia,210250
ABCG8	88.9	100.00%	93.70%	Sitosterolemia,210250 Gallbladder disease 4,611465
ACTN1	88.1	100.00%	98.80%	Bleeding disorder,platelet-type,15,615193
ANKRD26	109.5	97.40%	95.70%	Thrombocytopenia 2,188000
ANO6	93.8	98.40%	95.60%	Scott syndrome,262890
AP3B1	109.5	100.00%	100.00%	Hermansky-Pudlak syndrome 2,608233
BLOC1S3	30	76.60%	68.30%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	122.1	92.00%	80.70%	Hermansky-Pudlak syndrome 9,614171
CALR	136	96.40%	91.80%	Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
CD36	119	100.00%	100.00%	Platelet glycoprotein IV deficiency,608404 {Coronary heart disease,susceptibility to,7},610938 {Malaria,cerebral,reduced risk of},61162
COL3A1	68	97.60%	94.70%	Ehlers-Danlos syndrome, type IV,130050
CYCS	45.4	98.80%	83.60%	Thrombocytopenia 4,612004
DTNBP1	113.9	100.00%	99.80%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500
ETV6	106.9	100.00%	100.00%	Leukemia,acute myeloid,somatic,601626 Thrombocytopenia 5,616216
F10	88.2	100.00%	100.00%	Factor X deficiency,227600
F11	103.1	100.00%	96.30%	Factor XI deficiency,612416
F12	91.8	100.00%	99.90%	Factor XII deficiency,234000 Angioedema,hereditary,type III,610618
F13A1	90.8	97.90%	95.00%	Factor XIII deficiency,613225 {Myocardial infarction,protection against},608446 {Venous thrombosis,protection against},188050
F13B	91.1	99.20%	98.10%	Factor XIIIB deficiency,613235

F2	84	98.80%	87.80%	Dysprothrombinemia,613679 Hypoprothrombinemia,613679 Thrombophilia due to thrombin defect,188050 {Pregnancy loss,recurrent,susceptibility to,2},614390 {Stroke,ischemic,susceptibility to},601367
F5	127.8	100.00%	98.50%	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	91.8	100.00%	97.50%	Factor VII deficiency,227500 {Myocardial infarction,decreased susceptibility to},608446
F8	63.8	98.70%	94.40%	Hemophilia A,306700
F9	74.7	100.00%	100.00%	Hemophilia B,306900 Thrombophilia,X-linked,due to factor IX defect},300807 {Warfarin sensitivity},122700
FERMT3	95.5	99.00%	94.40%	Leukocyte adhesion deficiency,type III,612840
FGA	166.9	100.00%	98.90%	Afibrinogenemia,congenital,202400 Amyloidosis,familial visceral,105200 Dysfibrinogenemia,congenital,616004 Hypodysfibrinogenemia,congenital,616004
FGB	116.9	100.00%	99.20%	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004 Hypofibrinogenemia,congenital,202400
FGG	123	100.00%	99.90%	Afibrinogenemia,congenital,202400 Dysfibrinogenemia,congenital,616004
FLI1	104.3	96.40%	91.90%	No OMIM disease ID
FLNA	60	98.30%	92.10%	Cardiac valvular dysplasia,X-linked,314400 Congenital short bowel syndrome,300048 FG syndrome 2,300321 Frontometaphyseal dysplasia,305620 Heteropia,periventricular,300049 Heteropia,periventricular,ED variant,300537 Intestinal pseudoobstruction,neuro

GATA1	54.1	97.50%	84.10%	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 dysery
GFI1B	105.4	100.00%	100.00%	Bleeding disorder, platelet-type, 17,187900
GGCX	86.3	100.00%	95.40%	Psuedoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors,combined deficiency of,1,277450
GP1BA	118.2	97.80%	96.20%	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	22.7	88.00%	52.30%	Bernard-Soulier syndrome,type B,231200 Giant platelet disorder,isolated,231200
GP6	107.2	100.00%	97.70%	Bleeding disorder,platelet-type,11,614201
GP9	45.5	99.90%	83.30%	Bernard-Soulier syndrome,type C,231200
HABP2	74.3	100.00%	99.50%	{?Thyroid cancer,nonmedullary,5},616535 {Venous thromboembolism,susceptibility to},188050
HOXA11	109.6	100.00%	100.00%	Radioulnar synostosis with amegakaryocytic thrombocytopenia,605432
HPS1	73.8	98.20%	93.40%	Hermansky-Pudlak syndrome 1,203300
HPS3	111.9	100.00%	99.00%	Hermansky-Pudlak syndrome 3,614072
HPS4	107.4	100.00%	97.50%	Hermansky-Pudlak syndrome 4,614073
HPS5	95.3	96.30%	96.30%	Hermansky-Pudlak syndrome 5,614074
HPS6	75.3	99.40%	84.90%	Hermansky-Pudlak syndrome 6,614075
HRG	132.6	94.40%	93.90%	Thrombophilia due to elevated HRG,613116 Thrombophilia due to HRG deficiency,613116
ITGA2	115.7	100.00%	100.00%	?glycoprotein Ia deficiency,614200
ITGA2B	67.2	96.80%	88.80%	Bleeding disorder,platelet-type,16,autosomal dominant,187800 Glanzmann thrombasthenia,273800 Thrombocytopenia,neonatal alloimmune,BAK antigen related
ITGB3	88.6	100.00%	100.00%	Bleeding disorder,platelet-type 16,autosomal dominant,187800 Glanzmann thrombasthenia,273800 Purpura,posttransfusion Thrombocytopenia,neonatal alloimmune {Myocardial infarction,susceptibility to},608446

KLKB1	132.9	100.00%	99.60%	Fletcher factor (prekallikrein) deficiency,612423
LMAN1	117.9	100.00%	100.00%	Combined factor V and VIII deficiency,227300
LYST	117.1	99.50%	97.20%	Chediak-Higashi syndrome,214500
MASTL	118.9	100.00%	100.00%	?Thrombocytopenia 2,188000
MCFD2	50.4	99.50%	88.10%	Factor V and factor VIII,combined deficiency of,613625
MLPH	73.8	92.20%	89.00%	Griscelli syndrome type 3,609227
MPL	106.9	100.00%	93.70%	Myelofibrosis with myeloid metaplasia,somatic,254450 Thrombocythemia 2,601977 Thrombocytopenia,congenital amegakaryocytic,604498
MTHFR	92	99.20%	97.10%	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	85.7	99.20%	97.50%	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	89.9	99.00%	95.90%	Griscelli syndrome type 1,214450
NBEAL2	106	99.50%	97.60%	Gray platelet syndrome,139090
P2RX1	73.2	97.80%	95.40%	Bleeding disorder due to P2RX1 defect,somatic,609821
P2RY12	152.8	100.00%	100.00%	Bleeding disorder, platelet-type 8,609821
PLAT	91.4	99.50%	93.20%	Hyperfibrinolysis,familial,due to increased release of PLAT, 612348
PLAU	97.4	99.30%	96.70%	Quebec platelet disorder,601709 {Alzheimer disease,late-onset,susceptibility to},104300
PLG	62	73.80%	69.00%	Dysplasminogenemia,217090 Plasminogen deficiency, type I,217090
PRKACG	181.5	100.00%	100.00%	?Bleeding disorder,platelet-type,19,616176
PROC	78.3	100.00%	97.30%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	53.6	79.10%	68.40%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive,614514

PROZ	87.3	100.00%	98.90%	[Protein Z deficiency], 614024
PTGS1	98.1	100.00%	97.70%	No OMIM disease ID
RAB27A	110	100.00%	100.00%	Griscelli syndrome,type 2,607624
RASGRP2	70.4	100.00%	94.40%	?Bleeding disorder,platelet-type,18,615888
RBM8A	81.3	100.00%	100.00%	Thrombocytopenia-absent radius syndrome,274000
RUNX1	64.3	94.80%	84.20%	Leukemia,acute myeloid,601626 Platelet disorder,familial,with associated myeloid malignancy,601399
SELP	111.4	99.40%	97.50%	{Atopy,susceptibility to},147050
SERPINC1	121.5	100.00%	100.00%	Thrombophilia due to antithrombin III deficiency,613118
SERPIND1	110.1	100.00%	100.00%	Thrombophilia due to heparin cofactor II deficiency,612356
SERPINE1	94.6	97.40%	91.70%	Plasminogen activator inhibitor-1 deficiency,613329 {Transcription of plasminogen activator inhibitor,modulator of}
SERPINF2	125.1	100.00%	95.50%	Alpha-2-plasmin inhibitor deficiency,262850
SH2B3	95.2	95.80%	92.90%	Erythrocytosis,somatic,133100 Myelofibrosis,somatic,254450 Thrombocythemia,somatic,187950
STIM1	84.4	97.50%	95.10%	Immunodeficiency 10,612783 Myopathy,tubular aggregate,1,160565 Stormorken syndrome,185070
STXBP2	80.1	100.00%	96.00%	Hemophagocytic lymphohistiocytosis,familial,5,613101
TBX1	65.9	84.60%	70.80%	Conotruncal anomaly face syndrome,217095 DiGeorge syndrome,188400 Tetralogy of Fallot,187500 Velocardiofacial syndrome,192430
TBXA2R	77.6	98.60%	90.40%	{Bleeding disorder,platelet-type,13,susceptibility to},614009
TBXAS1	100.4	100.00%	98.10%	?Thromboxane synthase deficiency,614158 Ghosal hematodiaphyseal syndrome,231095
THBD	76.1	100.00%	100.00%	Thrombophilia due to thrombomodulin defect,614486 {Hemolytic uremic syndrome,atypical,susceptibility to,6}
THPO	110.5	95.80%	90.00%	Thrombocythemia 1,187950
VIPAS39	109.8	100.00%	99.50%	Arthrogryposis,renal dysfunction and cholestasis 2,613404
VKORC1	146.2	100.00%	100.00%	Vitamin K-dependent clotting factors,combined deficiency of,2,607473 Warfarin resistance,122700
VPS33B	96.5	100.00%	99.50%	Arthrogryposis,renal dysfunction,and cholestasis 1,208085

VWF	58	78.80%	71.60%	von Willebrand disease, type 1,193400 von Willebrand disease,type 2A,2B,2M and 2N,613554 von Willebrand disease,type 3,277480
WAS	30.2	91.20%	72.00%	Neutropenia,severe congenital,X-linked,300299 Thrombocytopenia,X-linked,313900 Wiskott-Aldrich syndrome,301000
WIPF1	91.4	96.60%	92.50%	?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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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