HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 3.4.0 (155 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
A2M	100,0%	100,0%	No OMIM Disease ID
ABCG5	100,0%	100,0%	Sitosterolemia 2, 618666
ABCG8	100,0%	100,0%	Sitosterolemia 1, 210250
ACBD5	100,0%	100,0%	Retinal dystrophy with leukodystrophy, 618863
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTN1	100,0%	100,0%	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	100,0%	100,0%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ANKRD26	97,2%	97,2%	Thrombocytopenia 2, 188000
ANO6	100,0%	100,0%	Scott syndrome, 262890
AP3B1	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100,0%	100,0%	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	100,0%	100,0%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
BLOC1S3	100,0%	100,0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100,0%	100,0%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100,0%	100,0%	?Hermansky-Pudlak syndrome 9, 614171
BRAF	100,0%	100,0%	Melanoma, malignant, somatic, 155600 LEOPARD syndrome 3, 613707 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 Noonan syndrome 7, 613706 Colorectal cancer, somatic, 114500 Nonsmall cell lung cancer, somatic, 211980
C3	100,0%	100,0%	C3 deficiency, 613779
CALR	100,0%	100,0%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950

CBL	100,0%	100,0%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CD36	100,0%	100,0%	Platelet glycoprotein IV deficiency, 608404
CD46	100,0%	100,0%	No OMIM Disease ID
CDC42	100,0%	100,0%	Takenouchi-Kosaki syndrome, 616737
CFB	100,0%	100,0%	?Complement factor B deficiency, 615561
CFH	100,0%	100,0%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFI	100,0%	100,0%	Complement factor I deficiency, 610984
CHST14	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	100,0%	100,0%	Osteogenesis imperfecta, type II, 166210 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL3A1	100,0%	100,0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329
COL5A2	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	100,0%	100,0%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CYCS	100,0%	100,0%	Thrombocytopenia 4, 612004
DGKE	100,0%	100,0%	Nephrotic syndrome, type 7, 615008
DIAPH1	100,0%	100,0%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	100,0%	100,0%	No OMIM Disease ID
DTNBP1	100,0%	100,0%	Hermansky-Pudlak syndrome 7, 614076
ENG	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	100,0%	99,9%	?Bleeding disorder, platelet-type, 22, 618462
ETV6	100,0%	100,0%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
F10	100,0%	100,0%	Factor X deficiency, 227600
F11	100,0%	100,0%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416

F12	100,0%	100,0%	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
F13A1	100,0%	100,0%	Factor XIIIA deficiency, 613225
F13B	100,0%	100,0%	Factor XIIIB deficiency, 613235
F2	100,0%	100,0%	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia 1 due to thrombin defect, 188050
F2RL3	100,0%	100,0%	No OMIM Disease ID
F5	100,0%	100,0%	Thrombophilia 2 due to activated protein C resistance, 188055 Factor V deficiency, 227400
F7	100,0%	100,0%	Factor VII deficiency, 227500
F8	100,0%	100,0%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071 Hemophilia A, 306700
F9	100,0%	99,8%	Hemophilia B, 306900 Thrombophilia 8, X-linked, due to factor IX defect, 300807
FBN1	100,0%	100,0%	Geleophysic dysplasia 2, 614185 Weill-Marchesani syndrome 2, dominant, 608328 Ectopia lentis, familial, 129600 MASS syndrome, 604308 Marfan lipodystrophy syndrome, 616914 Acromicric dysplasia, 102370 Marfan syndrome, 154700 Stiff skin syndrome, 184900
FERMT3	100,0%	100,0%	Leukocyte adhesion deficiency, type III, 612840
FGA	100,0%	100,0%	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400
FGB	100,0%	100,0%	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGG	100,0%	100,0%	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400
FLI1	100,0%	100,0%	Bleeding disorder, platelet-type, 21, 617443
FLNA	100,0%	100,0%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048

			Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FYB1	100,0%	100,0%	Thrombocytopenia 3, 273900
GATA1	100,0%	100,0%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	100,0%	100,0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100,0%	100,0%	Gaucher disease, type II, 230900 Gaucher disease, type IIIC, 231005 Gaucher disease, type III, 231000 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013
GDF2	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	100,0%	100,0%	Bleeding disorder, platelet-type, 17, 187900
GGCX	100,0%	100,0%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GNE	100,0%	100,0%	Sialuria, 269921 Nonaka myopathy, 605820
GP1BA	100,0%	100,0%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	100,0%	100,0%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	99,0%	96,3%	Bleeding disorder, platelet-type, 11, 614201
GP9	100,0%	100,0%	Bernard-Soulier syndrome, type C, 231200
HABP2	100,0%	100,0%	No OMIM Disease ID
HOXA11	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	100,0%	100,0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100,0%	100,0%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100,0%	100,0%	Hermansky-Pudlak syndrome 4, 614073

HPS5	100,0%	100,0%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100,0%	100,0%	Hermansky-Pudlak syndrome 6, 614075
HRG	100,0%	100,0%	Thrombophilia 11 due to HRG deficiency, 613116
IKZF5	100,0%	100,0%	Thrombocytopenia, autosomal dominant, 7, 619130
ITGA2	100,0%	100,0%	No OMIM Disease ID
ITGA2B	100,0%	100,0%	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGB3	100,0%	100,0%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,
JAK2	100,0%	100,0%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
KDSR	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 4, 617526
KLKB1	100,0%	100,0%	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	100,0%	100,0%	Angioedema, hereditary, 6, 619363
KRAS	100,0%	100,0%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 Arteriovenous malformation of the brain, somatic, 108010 Lung cancer, somatic, 211980 Pancreatic carcinoma, somatic, 260350 Leukemia, acute myeloid, somatic, 601626 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Cardiofaciocutaneous syndrome 2, 615278 Bladder cancer, somatic, 109800
LMAN1	100,0%	100,0%	Combined factor V and VIII deficiency, 227300
LYST	100,0%	100,0%	Chediak-Higashi syndrome, 214500
LZTR1	100,0%	100,0%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MASTL	100,0%	100,0%	No OMIM Disease ID
MCFD2	100,0%	100,0%	Factor V and factor VIII, combined deficiency of, 613625

MECOM	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	100,0%	100,0%	Griscelli syndrome, type 3, 609227
MPIG6B	100,0%	100,0%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100,0%	100,0%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	100,0%	100,0%	Homocystinuria due to MTHFR deficiency, 236250
MYH9	100,0%	100,0%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO5A	100,0%	100,0%	Griscelli syndrome, type 1, 214450
NBEA	100,0%	100,0%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	100,0%	100,0%	Gray platelet syndrome, 139090
NFE2	100,0%	100,0%	No OMIM Disease ID
NRAS	100,0%	100,0%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Melanocytic nevus syndrome, congenital, somatic, 137550 Epidermal nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Colorectal cancer, somatic, 114500
OCRL	100,0%	100,0%	Dent disease 2, 300555 Lowe syndrome, 309000
ORAI1	100,0%	100,0%	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
P2RY12	100,0%	100,0%	Bleeding disorder, platelet-type, 8, 609821
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PLA2G4A	100,0%	100,0%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	100,0%	100,0%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAT	100,0%	100,0%	No OMIM Disease ID
PLAU	100,0%	100,0%	Quebec platelet disorder, 601709
PLG	100,0%	100,0%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PRKACG	100,0%	100,0%	?Bleeding disorder, platelet-type, 19, 616176

PROC	100,0%	100,0%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860 Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PROS1	98,4%	98,4%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514 Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PROZ	100,0%	100,0%	No OMIM Disease ID
PTGS1	100,0%	100,0%	No OMIM Disease ID
PTPN11	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN22	100,0%	100,0%	No OMIM Disease ID
PTPRJ	100,0%	100,0%	Colon cancer, somatic, 114500
RAB27A	100,0%	100,0%	Griscelli syndrome, type 2, 607624
RAF1	100,0%	100,0%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RASGRP2	100,0%	100,0%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	100,0%	100,0%	Thrombocytopenia-absent radius syndrome, 274000
RIT1	100,0%	100,0%	Noonan syndrome 8, 615355
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RUNX1	100,0%	100,0%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SERPINC1	100,0%	100,0%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPIND1	100,0%	100,0%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356
SERPINE1	100,0%	100,0%	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF2	100,0%	100,0%	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	100,0%	100,0%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SLFN14	100,0%	100,0%	Bleeding disorder, platelet-type, 20, 616913
SOS1	100,0%	100,0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100,0%	100,0%	Noonan syndrome 9, 616559
SRC	100,0%	100,0%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500

STAB2	100,0%	100,0%	No OMIM Disease ID
STIM1	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STXBP2	99,8%	98,7%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
TALDO1	100,0%	100,0%	Transaldolase deficiency, 606003
TBX1	98,1%	95,9%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBXA2R	100,0%	99,7%	No OMIM Disease ID
TBXAS1	100,0%	100,0%	Ghosal hematodiaphyseal syndrome, 231095
THBD	100,0%	100,0%	Thrombophilia 12 due to thrombomodulin defect, 614486
THPO	100,0%	100,0%	Thrombocythemia 1, 187950
TNXB	100,0%	100,0%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	100,0%	100,0%	No OMIM Disease ID
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TUBB1	100,0%	100,0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	93,1%	93,0%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	100,0%	100,0%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WAS	100,0%	100,0%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WIPF1	100,0%	100,0%	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.
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.
TWIST is the chemistry used for WES analysis.

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 coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors