HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 3.2.0 (159 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
A2M	100	99,3	100	100	No OMIM disease ID
ABCG5	99,9	99,9	100	100	Sitosterolemia 2, 618666
ABCG8	99	97,1	100	100	Sitosterolemia 1, 210250
ACBD5	100	98,4	100	99,9	Retinal dystrophy with leukodystrophy, 618863
ACTB	99,9	97,2	100	100	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTN1	100	99,9	100	100	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	99,9	98,1	100	100	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	97	94,3	99,8	98,9	Thrombotic thrombocytopenic purpura, hereditary, 274150
ANKRD26	94,6	88,5	97,2	97	Thrombocytopenia 2, 188000
ANO6	99,2	96,8	100	100	Scott syndrome, 262890
AP3B1	99,2	96,4	100	99,9	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,6	98,4	100	100	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
BLOC1S3	99,9	90,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,3	97,6	100	100	?Hermansky-Pudlak syndrome 9, 614171
BRAF	89,4	77,6	100	100	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	99,9	98,5	100	100	C3 deficiency, 613779
CALR	94,5	87,5	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	97,3	96,9	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CD36	99,6	99,2	100	100	Platelet glycoprotein IV deficiency, 608404
CD46	99,7	98,9	100	99,9	No OMIM disease ID
CDC42	96,3	87,9	100	100	Takenouchi-Kosaki syndrome, 616737
CFB	100	99,6	100	100	?Complement factor B deficiency, 615561
CFH	99,8	98,5	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814
CFHR1	91,7	89,6	96,3	94,1	No OMIM disease ID
CFHR3	89	87,8	97,7	96	No OMIM disease ID
CFI	99,3	96	100	99,9	Complement factor I deficiency, 610984
CHST14	99,9	98,8	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	99,8	98,2	100	100	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	99,6	96,2	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	98,8	97,7	100	99,8	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329
COL5A2	100	98,4	100	100	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	100	100	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100
CYCS	99,4	96,9	100	100	Thrombocytopenia 4, 612004
DGKE	99,7	98,5	100	100	Nephrotic syndrome, type 7, 615008
DIAPH1	99,8	98,4	99,9	99	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	100	100	100	100	No OMIM disease ID

DTNBP1	99,7	98	100	99,9	Hermansky-Pudlak syndrome 7, 614076
ENG	99,8	97	100	100	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	98,1	98,1	99,4	98,7	?Bleeding disorder, platelet-type, 22, 618462
ETV6	100	99,3	100	100	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
F10	99,8	98,4	100	100	Factor X deficiency, 227600
F11	100	100	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	100	98,6	100	100	Angioedema, hereditary, 3, 610618 Factor XII deficiency, 234000
F13A1	100	100	100	100	Factor XIIIA deficiency, 613225
F13B	98,3	92,8	100	99,9	Factor XIIIB deficiency, 613235
F2	99,9	97,8	100	100	Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050
F2RL3	100	100	100	100	No OMIM disease ID
F5	99,9	98,4	100	100	Thrombophilia due to activated protein C resistance, 188055 Factor V deficiency, 227400
F7	100	100	100	100	Factor VII deficiency, 227500
F8	97,2	95,7	100	99,9	Hemophilia A, 306700
F9	99,7	98,3	100	97,9	Thrombophilia, X-linked, due to factor IX defect, 300807 Hemophilia B, 306900
FBN1	100	99,7	100	100	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
FCGR2A	100	100	100	100	No OMIM disease ID
FCGR2B	99,5	96,1	100	100	No OMIM disease ID
FCGR2C	98,2	98,1	99,4	99,3	No OMIM disease ID
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840

FGA	99,1	96,8	100	100	Hypodysfibrinogenemia, congenital, 616004 Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Afibrinogenemia, congenital, 202400
FGB	99,8	98,8	100	100	Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGG	99,4	97,5	100	100	Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Afibrinogenemia, congenital, 202400
FLI1	99,4	97,9	100	100	Bleeding disorder, platelet-type, 21, 617443
FLNA	100	99,9	100	100	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	99,4	95,9	100	100	Thrombocytopenia 3, 273900
GATA1	99,9	98,5	100	100	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	99,8	97	100	100	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100	100	100	100	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	100	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	99	97,3	100	100	Bleeding disorder, platelet-type, 17, 187900

GGCX	100	99,6	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GNE	100	99,5	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GP1BA	97,6	94,3	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	77,8	66,9	100	99,5	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100	99,9	97,7	93,5	Bleeding disorder, platelet-type, 11, 614201
GP9	98,1	91,8	100	100	Bernard-Soulier syndrome, type C, 231200
HABP2	100	99,7	100	100	No OMIM disease ID
HOXA11	97,1	88,3	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,8	97,1	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	99,9	99,3	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,7	88,7	100	100	Hermansky-Pudlak syndrome 6, 614075
HRG	95,4	93,9	100	100	Thrombophilia due to HRG deficiency, 613116
IKZF5	100	100	100	100	Thrombocytopenia, autosomal dominant, 7, 619130
ITGA2	99,1	96,8	100	100	No OMIM disease ID
ITGA2B	99,7	97,2	100	100	Glanzmann thrombasthenia 1, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGB3	100	99	100	100	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271 Glanzmann thrombasthenia 2, 619267 Thrombocytopenia, neonatal alloimmune, Purpura, posttransfusion,
JAK2	97,6	95,2	100	99,9	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
KDSR	99,8	99,5	100	100	Erythrokeratodermia variabilis et progressiva 4, 617526

KLKB1	99,8	99,3	100	99,9	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	100	100	100	100	Angioedema, hereditary, 6, 619363
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 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	99,8	98,5	100	100	Combined factor V and VIII deficiency, 227300
LYST	99,4	97,8	100	100	Chediak-Higashi syndrome, 214500
LZTR1	100	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MASTL	99,9	99,7	100	100	No OMIM disease ID
MCFD2	99,4	94,9	100	100	Factor V and factor VIII, combined deficiency of, 613625
MECOM	100	99,6	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	100	98,5	100	100	Griscelli syndrome, type 3, 609227
MPIG6B	100	99,9	100	100	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100	99,8	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	97,3	95,9	100	100	Homocystinuria due to MTHFR deficiency, 236250
MYH9	99,9	98,9	100	100	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYO5A	99,6	98,3	100	100	Griscelli syndrome, type 1, 214450
NBEA	91,8	90,3	100	100	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	99,5	99,3	100	100	Gray platelet syndrome, 139090
NFE2	100	100	100	100	No OMIM disease ID

NRAS	100	100	100	100	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	99,4	97,6	100	99,9	Dent disease 2, 300555 Lowe syndrome, 309000
ORAI1	99,3	97,1	99,4	96,7	Immunodeficiency 9, 612782 Myopathy, tubular aggregate, 2, 615883
P2RY12	100	100	100	100	Bleeding disorder, platelet-type, 8, 609821
PIGA	91,6	82,5	100	99,8	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868
PLA2G4A	99,5	99,1	100	99,9	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	99,8	99,3	100	100	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAT	100	98,5	100	100	No OMIM disease ID
PLAU	100	99,6	100	100	Quebec platelet disorder, 601709
PLG	87,8	87,6	100	100	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PRKACG	100	99,4	100	100	?Bleeding disorder, platelet-type, 19, 616176
PROC	100	100	100	100	Thrombophilia due to protein C deficiency, autosomal recessive, 612304 Thrombophilia due to protein C deficiency, autosomal dominant, 176860
PROS1	96,4	89,4	98,4	98,4	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	100	99,1	100	100	No OMIM disease ID
PTGS1	100	99,9	100	100	No OMIM disease ID
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN22	99,6	95,2	100	100	No OMIM disease ID
PTPRJ	97,6	96,4	100	99,6	Colon cancer, somatic, 114500

RAB27A	99,5	99,5	100	99,9	Griscelli syndrome, type 2, 607624
RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RASGRP2	100	98,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	99,6	95,3	100	100	Thrombocytopenia-absent radius syndrome, 274000
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RNU4ATAC	0	0	0	0	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
RUNX1	98,6	93	100	100	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SERPINC1	100	100	100	100	Thrombophilia due to antithrombin III deficiency, 613118
SERPIND1	100	100	100	100	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	100	100	100	100	Plasminogen activator inhibitor-1 deficiency, 613329
SERPINF2	100	99,9	100	100	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	99	94,7	100	100	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SLFN14	100	100	100	100	Bleeding disorder, platelet-type, 20, 616913
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	99,6	98,7	100	99,9	Noonan syndrome 9, 616559
SRC	100	99,6	100	100	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
STAB2	100	99,8	100	100	No OMIM disease ID
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STXBP2	82,4	79,9	99,7	98	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
TALDO1	100	98	100	100	Transaldolase deficiency, 606003
TBX1	87,4	77,6	93,7	90,2	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400

					Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBXA2R	97,4	93,9	99,8	98,7	No OMIM disease ID
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
THBD	100	99,9	100	100	Thrombophilia due to thrombomodulin defect, 614486
THPO	81,4	78,7	100	100	Thrombocythemia 1, 187950
TNXB	98,7	93,9	100	100	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	81,5	64,6	100	100	No OMIM disease ID
TREX1	100	100	100	100	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TUBB1	100	100	100	100	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	100	99,8	93	93	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	100	99,9	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	99,9	99	100	100	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willebrand disease, type 3, 277480
WAS	94,1	83,7	100	100	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WIPF1	99,9	98,5	100	100	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.
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Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : September 16th , 2021.

This list is accurate for panel version DG 3.2.0

Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors