HEMOSTATIC/THROMBOTIC DISORDERS PANEL DG-5.1.0 (161 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
A2M	96.1%	96%	100%	100%	99.8%	
ABCG5	100%	100%	100%	100%	99.5%	Sitosterolemia 2, 618666
ABCG8	100%	100%	100%	100%	99.7%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465
ACBD5	85.6%	85.6%	100%	100%	99.5%	Retinal dystrophy with leukodystrophy, 618863
ACTB	100%	100%	100%	99.9%	99.4%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTN1	95.9%	95.9%	100%	100%	99.4%	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	91.2%	91.2%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	100%	100%	100%	100%	99.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150

ANKRD26	100%	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000
ANO6	95.9%	94.9%	100%	100%	99.8%	Scott syndrome, 262890
AP3B1	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100%	100%	100%	100%	99.7%	?Hermansky-Pudlak syndrome 10, 617050
APOLD1	100%	99.4%	100%	99.9%	97.5%	?Bleeding disorder, vascular-type, 620715
ARPC1B	99.4%	95.9%	100%	100%	99.3%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
BLOC1S3	100%	100%	100%	100%	98.5%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S5	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 11, 619172
BLOC1S6	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 9, 614171
BRAF	100%	100%	100%	99.8%	98.2%	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	97%	97%	100%	99.9%	99.2%	C3 deficiency, 613779;{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925;{Macular degeneration, age-related, 9}, 611378

CALR	100%	100%	100%	100%	99.4%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CBL	100%	100%	100%	100%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CD36	100%	100%	100%	100%	99.8%	Platelet glycoprotein IV deficiency, 608404;{Coronary heart disease, susceptibility to, 7}, 610938;{Malaria, cerebral, susceptibility to}, 611162;{Malaria, cerebral, reduced risk of}, 611162
CD46	100%	100%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CDC42	100%	100%	100%	100%	99.6%	Takenouchi-Kosaki syndrome, 616737
CFB	100%	100%	100%	100%	99.4%	Complement factor B deficiency, 615561;{Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924;{Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	97.5%	97.5%	100%	100%	99.9%	{Macular degeneration, age-related, 4}, 610698;Basal laminar drusen, 126700;Complement factor H deficiency, 609814;{Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400

CFI	98.6%	95.9%	100%	100%	99.8%	{Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923;{Macular degeneration, age-related, 13, susceptibility to}, 615439;Complement factor I deficiency, 610984
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CLASP1	100%	100%	100%	100%	99.6%	
COL1A1	100%	100%	100%	100%	99.5%	Osteogenesis imperfecta, type II, 166210;Caffey disease, 114000;Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060;Osteogenesis imperfecta, type I, 166200;{Bone mineral density variation QTL, osteoporosis}, 166710;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type III, 259420
COL3A1	100%	100%	100%	100%	99.7%	Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	100%	100%	100%	100%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329

COL5A2	100%	100%	100%	100%	99.8%	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	93.2%	93.2%	100%	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CYCS	100%	100%	100%	100%	99.9%	Thrombocytopenia 4, 612004
DGKE	100%	100%	100%	100%	99.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008;Nephrotic syndrome, type 7, 615008
DIAPH1	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900;Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	100%	100%	100%	100%	99.4%	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	96.4%	96.4%	100%	100%	99.7%	Hermansky-Pudlak syndrome 7, 614076
ENG	97.3%	96.8%	100%	99.9%	98.6%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	100%	100%	100%	100%	99.1%	?Bleeding disorder, platelet-type, 22, 618462;{Prostate cancer/brain cancer susceptibility, somatic}, 603688
ERG	100%	100%	100%	100%	99.4%	Lymphatic malformation 14, 620602

ETV6	100%	100%	100%	100%	99.3%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
F10	100%	100%	100%	100%	99.4%	Factor X deficiency, 227600
F11	92.5%	92.5%	100%	100%	99.6%	Factor XI deficiency, autosomal dominant, 612416;Factor XI deficiency, autosomal recessive, 612416
F12	100%	100%	100%	100%	99.7%	Angioedema, hereditary, 3, 610618;Factor XII deficiency, 234000
F13A1	100%	100%	100%	100%	99.7%	Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050
F13B	100%	100%	100%	100%	99.9%	Factor XIIIB deficiency, 613235
F2	90.8%	90.8%	100%	99.9%	99.4%	Hypoprothrombinemia, 613679;{Pregnancy loss, recurrent, susceptibility to, 2}, 614390;Dysprothrombinemia, 613679;Thrombophilia 1 due to thrombin defect, 188050;{Stroke, ischemic, susceptibility to}, 601367
F2RL3	100%	100%	100%	100%	99.5%	

F5	100%	100%	100%	100%	99.8%	Thrombophilia 2 due to activated protein C resistance, 188055;{Pregnancy loss, recurrent, susceptibility to, 1}, 614389;{Thrombophilia, susceptibility to, due to factor V Leiden}, 188055;{Budd-Chiari syndrome}, 600880;{Stroke, ischemic, susceptibility to}, 601367;Factor V deficiency, 227400
F7	99.5%	96.6%	100%	100%	99.6%	{Myocardial infarction, decreased susceptibility to}, 608446;Factor VII deficiency, 227500
F8	100%	100%	99.3%	92.3%	74.9%	Thrombophilia 13, X-linked, due to factor VIII defect, 301071;Hemophilia A, 306700
F9	100%	100%	99.7%	92.5%	75.6%	{Deep venous thrombosis, protection against}, 300807;Hemophilia B, 306900;Thrombophilia 8, X-linked, due to factor IX defect, 300807;{Warfarin sensitivity}, 301052
FBN1	100%	100%	100%	100%	99.8%	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%	100%	100%	100%	99.4%	Leukocyte adhesion deficiency, type III, 612840
FGA	100%	99.7%	100%	100%	99.7%	Amyloidosis, hereditary systemic 2, 105200;Hypodysfibrinogenemia, congenital, 616004;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGB	100%	100%	100%	100%	99.6%	Hypofibrinogenemia, congenital, 202400;Dysfibrinogenemia, congenital, 616004;Afibrinogenemia, congenital, 202400
FGG	100%	100%	100%	100%	99.8%	Dysfibrinogenemia, congenital, 616004;Hypodysfibrinogenemia, 616004;Hypofibrinogenemia, congenital, 202400;Afibrinogenemia, congenital, 202400
FLI1	91.9%	91.9%	100%	100%	99.5%	Bleeding disorder, platelet-type, 21, 617443
FLNA	100%	100%	98.3%	87.4%	68.7%	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%	100%	100%	100%	99.9%	Thrombocytopenia 3, 273900
GALE	100%	100%	100%	100%	99.6%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GATA1	100%	100%	98.9%	88.4%	68.2%	Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;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	85.7%	85.7%	100%	100%	99.1%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286

GBA1	100%	98.3%	100%	100%	99.5%	{Lewy body dementia, susceptibility to}, 127750;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;{Parkinson disease, late-onset, susceptibility to}, 168600
GDF2	100%	100%	100%	100%	99.1%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	100%	100%	100%	100%	99.3%	Bleeding disorder, platelet-type, 17, 187900
GGCX	100%	100%	100%	100%	99.4%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GNE	100%	100%	100%	100%	99.8%	Sialuria, 269921;Thrombocytopenia 12 with or without myopathy, 620757;Nonaka myopathy, 605820

GP1BA	100%	100%	99.9%	99.7%	98.6%	Bernard-Soulier syndrome, type A1 (recessive), 231200;Bernard-Soulier syndrome, type A2 (dominant), 153670;von Willebrand disease, platelet-type, 177820;{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660
GP1BB	100%	100%	100%	99.4%	97.5%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GP6	100%	98.2%	100%	100%	99.6%	Bleeding disorder, platelet-type, 11, 614201
GP9	100%	100%	100%	100%	99%	Bernard-Soulier syndrome, type C, 231200
HABP2	100%	100%	100%	100%	99.9%	{?Thyroid cancer, nonmedullary, 5}, 616535;{Venous thromboembolism, susceptibility to}, 188050
HOXA11	100%	100%	100%	100%	99.1%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100%	100%	100%	100%	99.8%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100%	100%	100%	100%	99.4%	Hermansky-Pudlak syndrome 6, 614075

HRG	95.1%	93.3%	100%	100%	99.4%	Thrombophilia 11 due to HRG deficiency, 613116
IKZF5	100%	100%	100%	100%	99.8%	Thrombocytopenia, autosomal dominant, 7, 619130
ITGA2	100%	100%	100%	100%	99.9%	Fetomaternal alloimmune thrombocytopenia 3, 621267
ITGA2B	100%	100%	100%	100%	99.4%	Fetomaternal alloimmune thrombocytopenia 2, 621266;Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800
ITGB3	100%	100%	100%	100%	99.5%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;Glanzmann thrombasthenia 2, 619267;Fetomaternal alloimmune thrombocytopenia 1, 621264
JAK2	100%	100%	100%	99.9%	99.2%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300
KDSR	91.6%	91.6%	100%	100%	98.4%	Erythrokeratodermia variabilis et progressiva 4, 617526
KLKB1	100%	100%	100%	100%	99.8%	Fletcher factor (prekallikrein) deficiency, 612423

KNG1	100%	100%	100%	100%	99.7%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960
KRAS	100%	100%	100%	100%	100%	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%	100%	100%	100%	99.7%	Combined factor V and VIII deficiency, 227300
LYST	99.5%	99.5%	100%	100%	99.9%	Chediak-Higashi syndrome, 214500
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MASTL	100%	100%	100%	100%	99.8%	

MCFD2	100%	100%	100%	100%	99.6%	Factor V and factor VIII, combined deficiency of, 613625
MECOM	100%	100%	100%	100%	99.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	95.4%	95.4%	100%	100%	99.3%	Griscelli syndrome, type 3, 609227
MPIG6B	100%	100%	100%	100%	99.9%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100%	100%	100%	99.9%	99.3%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MTHFR	100%	100%	100%	100%	99.5%	{Vascular disease, susceptibility to};Homocystinuria due to MTHFR deficiency, 236250;{Thromboembolism, susceptibility to}, 188050;{Schizophrenia, susceptibility to}, 181500;{Neural tube defects, susceptibility to}, 601634
MYH9	97.3%	97%	100%	100%	99.4%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO5A	99%	99%	100%	100%	99.7%	Griscelli syndrome, type 1, 214450
NBEA	97.6%	97.6%	100%	100%	99.7%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157

NBEAL2	100%	100%	100%	100%	99.2%	Gray platelet syndrome, 139090
NFE2	100%	100%	100%	100%	99.7%	
NRAS	89.4%	89.4%	100%	100%	99.9%	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%	100%	99.3%	92.4%	74.8%	Dent disease 2, 300555;Lowe syndrome, 309000
ORAI1	100%	100%	100%	99.8%	98.6%	Immunodeficiency 9, 612782;Myopathy, tubular aggregate, 2, 615883
P2RY12	100%	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 8, 609821
PIGA	100%	100%	99.2%	90.8%	73.4%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072

PLA2G4A	100%	100%	100%	100%	99.8%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	100%	100%	100%	100%	99.6%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAT	96.1%	96.1%	100%	100%	99.4%	
PLAU	100%	100%	100%	100%	99.4%	Quebec platelet disorder, 601709;{Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	89.3%	89.3%	100%	100%	99.8%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PRKACG	100%	100%	100%	99.6%	98.3%	?Bleeding disorder, platelet-type, 19, 616176
PROC	100%	100%	100%	100%	99.6%	Thrombophilia 3 due to protein C deficiency, autosomal dominant, 176860;Thrombophilia 3 due to protein C deficiency, autosomal recessive, 612304
PROS1	100%	100%	100%	100%	99.7%	Thrombophilia 5 due to protein S deficiency, autosomal recessive, 614514;Thrombophilia 5 due to protein S deficiency, autosomal dominant, 612336
PROZ	97.7%	94.5%	100%	100%	99.1%	[Protein Z deficiency], 614024
PTGS1	96.6%	96.6%	100%	100%	99.7%	

PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN22	96.8%	96.8%	100%	100%	99.9%	
PTPRJ	100%	100%	100%	99.9%	99.5%	Colon cancer, somatic, 114500;Thrombocytopenia 10, 620484
RAB27A	100%	100%	100%	100%	99.6%	Griscelli syndrome, type 2, 607624
RAF1	96.1%	93.8%	100%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RAP1B	100%	100%	100%	100%	99.6%	Thrombocytopenia 11 with multiple congenital anomalies and dysmorphic facies, 620654
RASGRP2	98.6%	98%	100%	100%	99.4%	Bleeding disorder, platelet-type, 18, 615888
RBM8A	100%	100%	100%	99.7%	98.9%	Thrombocytopenia-absent radius syndrome, 274000
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355
RNU4ATAC						Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710

RUNX1	100%	100%	100%	99.7%	97.9%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
SERPINC1	90.4%	88.6%	100%	100%	99.9%	Thrombophilia 7 due to antithrombin III deficiency, 613118
SERPIND1	100%	100%	100%	100%	99.8%	Thrombophilia 10 due to heparin cofactor II deficiency, 612356
SERPINE1	100%	100%	100%	100%	99.6%	Plasminogen activator inhibitor-1 deficiency, 613329;{Transcription of plasminogen activator inhibitor, modulator of}
SERPINF2	92.1%	91.2%	100%	100%	99.4%	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	100%	100%	100%	99.9%	99.3%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SLFN14	100%	100%	100%	100%	99.8%	Bleeding disorder, platelet-type, 20, 616913
SMAD4	95.4%	95.4%	100%	100%	99.8%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	95%	95%	100%	100%	99.5%	Noonan syndrome 9, 616559

SRC	96.5%	96.5%	100%	99.9%	99.3%	?Thrombocytopenia 6, 616937;Colon cancer, advanced, somatic, 114500
STAB2	100%	100%	100%	100%	99.8%	
STIM1	100%	99.9%	100%	99.9%	99.7%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STXBP2	100%	100%	100%	99.9%	99.3%	Hemophagocytic lymphohistiocytosis, familial, 5, with or without microvillus inclusion disease, 613101
TALDO1	89.1%	89.1%	100%	100%	99.7%	Transaldolase deficiency, 606003
TBX1	98.6%	95.7%	100%	99.4%	96.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBXA2R	100%	99.6%	100%	100%	99.5%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	100%	100%	100%	100%	99.8%	Ghosal hematodiaphyseal syndrome, 231095
THBD	100%	100%	100%	99.8%	98.7%	Thrombophilia 12 due to thrombomodulin defect, 614486;{Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926

THPO	100%	100%	100%	100%	99.3%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TNXB	100%	100%	100%	99.9%	98.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TPM4	89.4%	88.4%	100%	100%	99%	Bleeding disorder, platelet-type, 25, 620486
TREX1	100%	100%	100%	100%	99.5%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448
TUBB1	100%	100%	100%	100%	99.8%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112
VIPAS39	99.8%	97.9%	100%	100%	99.7%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VKORC1	98.6%	93.5%	100%	100%	99.2%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473;Warfarin resistance, 122700

VPS33B	100%	100%	100%	100%	99.9%	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	100%	100%	100%	100%	99.5%	von Willebrand disease, type 1, 193400;von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554;von Willebrand disease, type 3, 277480
WAS	99.6%	95.8%	98.5%	87.7%	68.3%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WIPF1	100%	100%	100%	100%	99.7%	Wiskott-Aldrich syndrome 2, 614493

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors