HEMOSTATIC/THROMBOTIC DISORDERS PANEL DG-3.9.0 (47 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
A2M	100.0%	100.0%	100.0%	99.1%	
ABCG5	100.0%	100.0%	100.0%	98.3%	Sitosterolemia 2, 618666
ABCG8	100.0%	100.0%	100.0%	99.2%	Sitosterolemia 1, 210250;{Gallbladder disease 4}, 611465
ACBD5	100.0%	100.0%	100.0%	98.3%	Retinal dystrophy with leukodystrophy, 618863
ACTB	100.0%	100.0%	100.0%	99.0%	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, 620479
ACTN1	100.0%	100.0%	100.0%	99.4%	Bleeding disorder, platelet-type, 15, 615193

ACVRL1	100.0%	100.0%	100.0%	99.0%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	100.0%	100.0%	100.0%	98.4%	Thrombotic thrombocytopenic purpura, hereditary, 274150
ANKRD26	97.2%	97.2%	100.0%	97.0%	Thrombocytopenia 2, 188000
ANO6	100.0%	100.0%	100.0%	98.3%	Scott syndrome, 262890
AP3B1	100.0%	100.0%	100.0%	98.8%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	100.0%	100.0%	100.0%	99.2%	?Hermansky-Pudlak syndrome 10, 617050
APOLD1	100.0%	100.0%	100.0%	93.4%	?Bleeding disorder, vascular-type, 620715
ARPC1B	100.0%	100.0%	100.0%	99.9%	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
HRG	100.0%	100.0%	100.0%	98.7%	Thrombophilia 11 due to HRG deficiency, 613116
IKZF5	100.0%	100.0%	100.0%	97.6%	Thrombocytopenia, autosomal dominant, 7, 619130
ITGA2	100.0%	99.9%	100.0%	98.0%	

ITGA2B	100.0%	100.0%	100.0%	99.3%	Glanzmann thrombasthenia 1, 273800;Bleeding disorder, platelet-type, 16, autosomal dominant, 187800;Thrombocytopenia, neonatal alloimmune, BAK antigen related,
ITGB3	100.0%	100.0%	100.0%	98.5%	Bleeding disorder, platelet-type, 24, autosomal dominant, 619271;{Myocardial infarction, susceptibility to}, 608446;Glanzmann thrombasthenia 2, 619267;Thrombocytopenia, neonatal alloimmune, ;Purpura, posttransfusion,
JAK2	100.0%	100.0%	100.0%	98.3%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300
KNG1	100.0%	100.0%	100.0%	98.3%	[Kininogen deficiency], 228960;Angioedema, hereditary, 6, 619363;[High molecular weight kininogen deficiency], 228960

KRAS	100.0%	100.0%	100.0%	99.7%	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%	100.0%	98.0%	Combined factor V and VIII deficiency, 227300
LYST	100.0%	99.8%	100.0%	98.8%	Chediak-Higashi syndrome, 214500
LZTR1	100.0%	100.0%	100.0%	99.7%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MASTL	100.0%	100.0%	100.0%	98.5%	

MCFD2	100.0%	100.0%	100.0%	96.8%	Factor V and factor VIII, combined deficiency of, 613625
MECOM	100.0%	100.0%	100.0%	98.9%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	100.0%	100.0%	100.0%	99.2%	Griscelli syndrome, type 3, 609227
MYH9	100.0%	100.0%	100.0%	98.8%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622
MYO5A	100.0%	100.0%	100.0%	98.4%	Griscelli syndrome, type 1, 214450
NBEA	99.7%	99.2%	100.0%	98.1%	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	100.0%	100.0%	100.0%	99.4%	Gray platelet syndrome, 139090
NFE2	100.0%	100.0%	100.0%	99.0%	

NRAS	100.0%	100.0%	100.0%	99.7%	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%	97.8%	69.7%	Dent disease 2, 300555;Lowe syndrome, 309000
PIGA	100.0%	100.0%	97.7%	73.6%	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%	100.0%	98.7%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372

PLA2G7	100.0%	100.0%	100.0%	96.7%	Platelet-activating factor acetylhydrolase deficiency, 614278
PLAT	100.0%	100.0%	100.0%	99.5%	
PLAU	100.0%	100.0%	100.0%	98.1%	Quebec platelet disorder, 601709;{Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	100.0%	100.0%	100.0%	98.8%	Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PRKACG	100.0%	100.0%	100.0%	96.2%	?Bleeding disorder, platelet-type, 19, 616176
RAB27A	100.0%	100.0%	100.0%	99.1%	Griscelli syndrome, type 2, 607624
RAF1	100.0%	100.0%	100.0%	98.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RASGRP2	100.0%	100.0%	100.0%	98.7%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	100.0%	100.0%	99.9%	97.6%	Thrombocytopenia-absent radius syndrome, 274000

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 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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 3.9.0

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