HEMOSTATIC/THROMBOTIC DISORDERS DG 2.16 (150 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
A2M	107,3	100.0%	99.3%	Alpha-2-macroglobulin deficiency, 614036 {Alzheimer disease, susceptibility to}, 104300
ABCG5	139,5	100.0%	99.9%	Sitosterolemia, 210250
ABCG8	133,9	99.7%	98.2%	Sitosterolemia, 210250 {Gallbladder disease 4}, 611465
ACBD5	145,1	99.6%	98.0%	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACTN1	131,5	100.0%	100.0%	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	113,6	100.0%	98.4%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	103,8	98.1%	95.2%	Thrombotic thrombocytopenic purpura, familial, 274150
ANKRD26	83,3	95.3%	90.1%	Thrombocytopenia 2, 188000
ANO6	133,3	99.8%	98.0%	Scott syndrome, 262890
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
AP3D1	125,2	98.4%	97.9%	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	139,6	100.0%	100.0%	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease, 617718
BLOC1S3	67,4	100.0%	99.9%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706
C3	141,6	100.0%	99.4%	C3 deficiency, 613779 {Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 {Macular degeneration, age-related, 9}, 611378
CALR	111,8	98.1%	91.7%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950

CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CD36	119,8	99.7%	99.1%	Platelet glycoprotein IV deficiency, 608404 [Macrothrombocytopenia], 0 {Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, reduced risk of}, 611162 {Malaria, cerebral, susceptibility to}, 611162
CD46	125,5	99.7%	98.7%	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CFB	119,4	100.0%	99.9%	?Complement factor B deficiency, 615561 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924 {Macular degeneration, age-related, 14, reduced risk of}, 615489
CFH	155,4	99.4%	97.9%	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	158,4	93.6%	90.8%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	98,4	93.8%	91.6%	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFI	139	99.5%	97.0%	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	141	99.8%	98.4%	Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Bone mineral density variation QTL, osteoporosis}, 166710
COL3A1	99,2	99.3%	96.8%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	136,4	99.9%	98.9%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100,2	99.9%	99.4%	Ehlers-Danlos syndrome, classic type, 2, 130010
CTLA4	141	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100 {Celiac disease, susceptibility to, 3}, 609755 {Diabetes mellitus, insulin-dependent, 12}, 601388

				{Hashimoto thyroiditis}, 140300 {Systemic lupus erythematosus, susceptibility to}, 152700
CYCS	61	99.1%	93.1%	Thrombocytopenia 4, 612004
DGKE	127,8	99.8%	98.3%	Nephrotic syndrome, type 7, 615008 {Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008
DIAPH1	101,7	99.9%	99.6%	Deafness, autosomal dominant 1, 124900 Seizures, cortical blindness, microcephaly syndrome, 616632
DNASE1	168,5	100.0%	100.0%	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	113,4	99.8%	97.9%	Hermansky-Pudlak syndrome 7, 614076
ENG	128,4	99.9%	98.7%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	190,2	98.1%	98.1%	{Prostate cancer/brain cancer susceptibility, somatic}, 603688
ETV6	148,3	100.0%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
F10	173,6	99.8%	99.1%	Factor X deficiency, 227600
F11	126,7	100.0%	99.9%	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	151,3	99.9%	99.5%	Angioedema, hereditary, type III, 610618 Factor XII deficiency, 234000
F13A1	112,9	100.0%	99.6%	Factor XIIIA deficiency, 613225 {Myocardial infarction, protection against}, 608446 {Venous thrombosis, protection against}, 188050
F13B	105,8	98.6%	92.9%	Factor XIIIB deficiency, 613235
F2	128,1	99.8%	97.1%	Dysprothrombinemia, 613679 Hypoprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Pregnancy loss, recurrent, susceptibility to, 2}, 614390 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	130,4	100.0%	100.0%	No OMIM phenotype Impaired thrombin-induced platelet response (Bianchi et al. (2016) Blood 127(10):1249-1259)
F5	145,5	99.5%	97.7%	Factor V deficiency, 227400 Thrombophilia due to activated protein C resistance, 188055 {Budd-Chiari syndrome}, 600880 {Pregnancy loss, recurrent, susceptibility to, 1}, 614389 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055
F7	162	100.0%	100.0%	Factor VII deficiency, 227500

				{Myocardial infarction, decreased susceptibility to}, 608446
F8	106	99.4%	97.2%	Hemophilia A, 306700
F9	109,5	99.9%	98.4%	Hemophilia B, 306900 Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 {Warfarin sensitivity}, 122700
FBN1	137,1	100.0%	99.8%	Acromicric dysplasia, 102370 Ectopia lentis, familial, 129600 Geleophysic dysplasia 2, 614185 Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Stiff skin syndrome, 184900 Weill-Marchesani syndrome 2, dominant, 608328
FCGR2A	164,6	100.0%	100.0%	{Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162 {Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700
FCGR2B	119,7	99.9%	96.9%	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	148,2	98.2%	97.8%	Thrombocytopenic purpura, autoimmune, 188030
FERMT3	144,9	100.0%	99.9%	Leukocyte adhesion deficiency, type III, 612840
FGA	137	99.3%	97.3%	Afibrinogenemia, congenital, 202400 Amyloidosis, familial visceral, 105200 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, congenital, 616004
FGB	136,7	99.7%	98.2%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypofibrinogenemia, congenital, 202400
FGG	126,4	99.8%	98.0%	Afibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400
FLI1	153,8	99.3%	97.9%	Bleeding disorder, platelet-type, 21, 617443
FLNA	142,7	100.0%	99.9%	?FG syndrome 2, 300321 Cardiac valvular dysplasia, X-linked, 314400 Congenital short bowel syndrome, 300048 Frontometaphyseal dysplasia 1, 305620

				Heterotopia, periventricular, 1, 300049 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Otopalatodigital syndrome, type I, 311300 Otopalatodigital syndrome, type II, 304120 Terminal osseous dysplasia, 300244
FYB1	96,3	99.5%	96.8%	Thrombocytopenia 3, 273900
GATA1	92,9	99.9%	98.2%	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 dyserythropoietic anemia, 300367
GATA2	115	100.0%	99.0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GBA	169,8	100.0%	100.0%	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GDF2	142,4	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	170,9	99.9%	98.2%	Bleeding disorder, platelet-type, 17, 187900
GGCX	101,2	100.0%	99.4%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GNE	113,8	100.0%	99.3%	Nonaka myopathy, 605820 Sialuria, 269921
GP1BA	136,8	98.7%	95.7%	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	68,6	94.5%	83.1%	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GP6	123,2	100.0%	99.7%	Bleeding disorder, platelet-type, 11, 614201
GP9	134,3	99.9%	98.3%	Bernard-Soulier syndrome, type C, 231200
HABP2	109,3	100.0%	99.4%	{?Thyroid cancer, nonmedullary, 5}, 616535

				{Venous thromboembolism, susceptibility to}, 188050
HOXA11	88,3	100.0%	98.0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	115,8	100.0%	99.9%	Hermansky-Pudlak syndrome 1, 203300
HPS3	132,7	99.9%	98.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	128,1	100.0%	99.9%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122,8	99.9%	98.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	164,6	99.9%	97.8%	Hermansky-Pudlak syndrome 6, 614075
HRG	126,1	95.0%	94.2%	Thrombophilia due to elevated HRG, 613116 Thrombophilia due to HRG deficiency, 613116
ITGA2	134,1	99.7%	97.9%	?Glycoprotein Ia deficiency, 614200
ITGA2B	124,4	99.9%	98.9%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGB3	112,8	100.0%	99.8%	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0 {Myocardial infarction, susceptibility to}, 608446
JAK2	103,5	97.6%	95.0%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
KLKB1	132,3	100.0%	99.5%	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	150,7	100.0%	100.0%	[High molecular weight kininogen deficiency], 228960 [Kininogen deficiency], 228960
KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 60162 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470

				Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
LMAN1	144,3	99.8%	99.4%	Combined factor V and VIII deficiency, 227300
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
LZTR1	143,6	100.0%	99.7%	Noonan syndrome 10, 616564 Noonan syndrome 2, 605275 {Schwannomatosis-2, susceptibility to}, 615670
MASTL	134,9	100.0%	100.0%	?Thrombocytopenia-2, 188000
MCFD2	91,4	97.6%	91.0%	Factor V and factor VIII, combined deficiency of, 613625
MECOM	131,2	100.0%	99.6%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	97,4	99.7%	97.2%	Griscelli syndrome, type 3, 609227
MPIG6B	119	100.0%	99.8%	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	125,8	100.0%	99.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MTHFR	114,9	98.2%	96.4%	Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Schizophrenia, susceptibility to}, 181500 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MYH9	128,5	99.6%	98.5%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	109	99.7%	98.6%	Griscelli syndrome, type 1, 214450
NBEA	125,1	91.9%	90.2%	No OMIM phenotype Autism, idiopathic (Castermans (2003) J Med Genet 40, 352) ?Schizophrenia (Fromer (2014) Nature 506, 179) ?Obesity, extreme (Mariman (2015) Physiol Genomics 47,225) ?Tetralogy of Fallot (Silversides (2012) PloS Genet 8)
NBEAL2	166	100.0%	99.5%	Gray platelet syndrome, 139090
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
ORAI1	198,9	99.8%	98.2%	Immunodeficiency 9, 612782

				Myopathy, tubular aggregate, 2, 615883
P2RY12	186,2	100.0%	100.0%	Bleeding disorder, platelet-type, 8, 609821
PLA2G4A	124,5	99.8%	99.3%	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	120,9	99.8%	98.6%	Platelet-activating factor acetylhydrolase deficiency, 614278 {Asthma, susceptibility to}, 600807 {Atopy, susceptibility to}, 147050
PLAT	92	100.0%	99.2%	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	99,6	100.0%	99.0%	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	93,4	87.8%	86.8%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	204,5	100.0%	100.0%	?Bleeding disorder, platelet-type, 19, 616176
PROC	142,6	100.0%	100.0%	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	95,8	97.7%	92.7%	Thrombophilia due to protein S deficiency, autosomal dominant, 612336 Thrombophilia due to protein S deficiency, autosomal recessive, 614514
PROZ	130	99.8%	98.8%	[Protein Z deficiency], 614024
PTGS1	137,6	100.0%	99.4%	No OMIM phenotype
PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN22	127,8	99.6%	95.9%	{Diabetes, type 1, susceptibility to}, 222100 {Rheumatoid arthritis, susceptibility to}, 180300 {Systemic lupus erythematosus susceptibility to}, 152700
PTPRJ	137,4	97.7%	96.3%	Colon cancer, somatic, 114500
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RASGRP2	102,5	100.0%	99.7%	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	87,4	99.8%	97.4%	Thrombocytopenia-absent radius syndrome, 274000
RIT1	139,2	100.0%	100.0%	Noonan syndrome 8, 615355
RUNX1	84,6	99.6%	96.3%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SERPINC1	117,9	100.0%	100.0%	Thrombophilia due to antithrombin III deficiency, 613118

SERPIND1	134,8	100.0%	99.9%	Thrombophilia due to heparin cofactor II deficiency, 612356
SERPINE1	134	100.0%	100.0%	Plasminogen activator inhibitor-1 deficiency, 613329 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF2	151,6	100.0%	99.9%	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	108,4	99.9%	97.6%	Erythrocytosis, somatic, 133100 Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
SLFN14	172	100.0%	100.0%	Bleeding disorder, platelet-type, 20, 616913
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOS2	99,7	99.7%	97.9%	Noonan syndrome 9, 616559
SRC	124,1	100.0%	99.5%	?Thrombocytopenia 6, 616937 Colon cancer, advanced, somatic, 114500
STIM1	120,7	99.8%	96.8%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STXBP2	100,2	83.7%	80.4%	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TALDO1	148,2	100.0%	99.6%	Transaldolase deficiency, 606003
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBXA2R	119,6	97.2%	93.9%	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	128,8	100.0%	100.0%	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
THBD	181,1	100.0%	100.0%	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THPO	97,3	100.0%	99.7%	Thrombocythemia 1, 187950
TNXB	105,6	99.5%	95.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	59,5	96.9%	86.3%	No OMIM phenotype
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TUBB1	150	100.0%	100.0%	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112
VIPAS39	114,7	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404

VKORC1	146,5	100.0%	99.9%	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 Warfarin resistance, 122700
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	98,2	99.9%	99.1%	von Willebrand disease, type 1, 193400 von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554 von Willibrand disease, type 3, 277480
WAS	70,4	94.2%	83.6%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WIPF1	89,1	100.0%	99.1%	?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.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions: May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors