HEMOSTATIC/THROMBOTIC DISORDERS GENE PANEL DG 3.1.0 (157 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
A2M	99,9	99,6	100	100	{Alzheimer disease, susceptibility to}, 104300
ABCG5	100	100	100	100	Sitosterolemia 2, 618666
ABCG8	99,1	97,3	100	100	{Gallbladder disease 4}, 611465 Sitosterolemia 1, 210250
ACBD5	100	99,2	100	100	Retinal dystrophy with leukodystrophy, 618863
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTN1	100	100	100	100	Bleeding disorder, platelet-type, 15, 615193
ACVRL1	100	98,9	100	100	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAMTS13	97,1	93,8	99,9	99,5	Thrombotic thrombocytopenic purpura, hereditary, 274150
ANKRD26	95	89,3	97,2	97,2	Thrombocytopenia 2, 188000
ANO6	99,9	98,7	100	100	Scott syndrome, 262890
AP3B1	99,2	95,8	100	100	Hermansky-Pudlak syndrome 2, 608233
AP3D1	99,8	98,6	100	100	?Hermansky-Pudlak syndrome 10, 617050
ARPC1B	100	100	100	100	Immunodeficiency 71 with inflammatory disease and congenital thrombocytopenia, 617718
BLOC1S3	98,5	81,3	100	100	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,9	97,8	100	100	?Hermansky-pudlak syndrome 9, 614171
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
C3	99,9	99,2	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 5}, 612925 C3 deficiency, 613779 {Macular degeneration, age-related, 9}, 611378

CALR	94,8	89,1	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CD36	99,7	98,7	100	100	{Coronary heart disease, susceptibility to, 7}, 610938 {Malaria, cerebral, susceptibility to}, 611162 Platelet glycoprotein IV deficiency, 608404 {Malaria, cerebral, reduced risk of}, 611162
CD46	99,9	99,4	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}, 612922
CDC42	97,9	90,9	100	100	Takenouchi-Kosaki syndrome, 616737
CFB	100	100	100	100	?Complement factor B deficiency, 615561 {Macular degeneration, age-related, 14, reduced risk of}, 615489 {Hemolytic uremic syndrome, atypical, susceptibility to, 4}, 612924
CFH	99,9	99	100	99,9	Basal laminar drusen, 126700 Complement factor H deficiency, 609814 {Hemolytic uremic syndrome, atypical, susceptibility to, 1}, 235400 {Macular degeneration, age-related, 4}, 610698
CFHR1	96,4	94,9	95,4	93,8	{Hemolytic uremic syndrome, atypical, susceptibility to}, 235400 {Macular degeneration, age-related, reduced risk of}, 603075
CFHR3	94	92,2	96	95,2	{Macular degeneration, age-related, reduced risk of}, 603075 {Hemolytic uremic syndrome, atypical, susceptibility to}, 235400
CFI	99,2	96,8	100	100	Complement factor I deficiency, 610984 {Hemolytic uremic syndrome, atypical, susceptibility to, 3}, 612923 {Macular degeneration, age-related, 13, susceptibility to}, 615439
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
COL1A1	99,9	98,6	100	100	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1, 619115 Osteogenesis imperfecta, type I, 166200 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type II, 166210 {Bone mineral density variation QTL, osteoporosis}, 166710 Caffey disease, 114000 Ehlers-Danlos syndrome, arthrochalasia type, 1, 130060 Osteogenesis imperfecta, type III, 259420
COL3A1	99,6	97,6	100	100	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	98,8	98	100	99,9	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100	99,5	100	100	Ehlers-Danlos syndrome, classic type, 2, 130010

CTLA4	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Celiac disease, susceptibility to, 3}, 609755 Autoimmune lymphoproliferative syndrome, type V, 616100 {Hashimoto thyroiditis}, 140300
CYCS	99,1	94,9	100	100	Thrombocytopenia 4, 612004
DGKE	99,8	98,1	100	100	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}, 615008 Nephrotic syndrome, type 7, 615008
DIAPH1	99,8	99	99,5	98	Seizures, cortical blindness, microcephaly syndrome, 616632 Deafness, autosomal dominant 1, with or without thrombocytopenia, 124900
DNASE1	100	99,9	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700
DTNBP1	99,8	98,7	100	100	Hermansky-Pudlak syndrome 7, 614076
ENG	99,6	96	100	100	Telangiectasia, hereditary hemorrhagic, type 1, 187300
EPHB2	98,1	98,1	99,8	98,8	{Prostate cancer/brain cancer susceptibility, somatic}, 603688 ?Bleeding disorder, platelet-type, 22, 618462
ETV6	100	99,9	100	100	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
F10	99,8	99,1	100	100	Factor X deficiency, 227600
F11	100	100	100	100	Factor XI deficiency, autosomal dominant, 612416 Factor XI deficiency, autosomal recessive, 612416
F12	99,9	98,8	100	100	Factor XII deficiency, 234000 Angioedema, hereditary, type III, 610618
F13A1	100	100	100	100	{Myocardial infarction, protection against}, 608446 Factor XIIIA deficiency, 613225 {Venous thrombosis, protection against}, 188050
F13B	98,7	93,5	100	100	Factor XIIIB deficiency, 613235
F2	99,9	97,9	100	100	{Pregnancy loss, recurrent, susceptibility to, 2}, 614390 Hypoprothrombinemia, 613679 Dysprothrombinemia, 613679 Thrombophilia due to thrombin defect, 188050 {Stroke, ischemic, susceptibility to}, 601367
F2RL3	100	100	100	100	No OMIM disease ID
F5	99,4	98,5	100	100	{Pregnancy loss, recurrent, susceptibility to, 1}, 614389 Thrombophilia due to activated protein C resistance, 188055 Factor V deficiency, 227400 {Budd-Chiari syndrome}, 600880 {Stroke, ischemic, susceptibility to}, 601367 {Thrombophilia, susceptibility to, due to factor V Leiden}, 188055

F7	100	100	100	100	Factor VII deficiency, 227500 {Myocardial infarction, decreased susceptibility to}, 608446
F8	97,3	96,1	100	99,9	Hemophilia A, 306700
F9	99,6	97,6	99,9	98,8	Thrombophilia, X-linked, due to factor IX defect, 300807 {Deep venous thrombosis, protection against}, 300807 Hemophilia B, 306900 {Warfarin sensitivity}, 301052
FBN1	100	99,9	100	100	Marfan lipodystrophy syndrome, 616914 Marfan syndrome, 154700 MASS syndrome, 604308 Ectopia lentis, familial, 129600 Acromicric dysplasia, 102370 Weill-Marchesani syndrome 2, dominant, 608328 Geleophysic dysplasia 2, 614185 Stiff skin syndrome, 184900
FCGR2A	100	100	100	100	{Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis}, 219700 {Lupus nephritis, susceptibility to}, 152700 {Malaria, severe, susceptibility to}, 611162
FCGR2B	99,5	95,4	100	100	{Malaria, resistance to}, 611162 {Systemic lupus erythematosus, susceptibility to}, 152700
FCGR2C	98,9	98,6	98,1	97,2	No OMIM disease ID
FERMT3	100	100	100	100	Leukocyte adhesion deficiency, type III, 612840
FGA	99,1	97,2	100	100	Dysfibrinogenemia, congenital, 616004 Amyloidosis, familial visceral, 105200 Hypodysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FGB	99,8	99,1	100	100	Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400 Hypofibrinogenemia, congenital, 202400
FGG	99,7	98,2	100	100	Hypodysfibrinogenemia, 616004 Hypofibrinogenemia, congenital, 202400 Dysfibrinogenemia, congenital, 616004 Afibrinogenemia, congenital, 202400
FLI1	99,5	98,2	100	100	Bleeding disorder, platelet-type, 21, 617443
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048

					Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FYB1	99,4	97	100	100	Thrombocytopenia 3, 273900
GATA1	99,8	98,4	100	100	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	100	98,3	100	100	Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626
GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GDF2	100	100	100	100	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GFI1B	98,5	96,7	100	100	Bleeding disorder, platelet-type, 17, 187900
GGCX	100	99,9	100	100	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GNE	100	99,7	100	100	Sialuria, 269921 Nonaka myopathy, 605820
GP1BA	98,6	95,9	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,9	59,6	99,5	95	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GP6	100	100	94,9	91,7	Bleeding disorder, platelet-type, 11, 614201
GP9	96,5	89,3	100	100	Bernard-Soulier syndrome, type C, 231200
HABP2	100	99,9	100	100	{?Thyroid cancer, nonmedullary, 5}, 616535 {Venous thromboembolism, susceptibility to}, 188050

HOXA11	97,1	87,5	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
HPS1	100	100	100	100	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,7	97,5	100	100	Hermansky-Pudlak syndrome 3, 614072
HPS4	100	100	100	100	Hermansky-Pudlak syndrome 4, 614073
HPS5	100	99,7	100	100	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,1	88,9	100	100	Hermansky-Pudlak syndrome 6, 614075
HRG	95	94,2	100	100	Thrombophilia due to HRG deficiency, 613116
IKZF5	100	100	100	100	Thrombocytopenia, autosomal dominant, 7, 619130
ITGA2	99,6	98,1	100	100	No OMIM disease ID
ITGA2B	99,7	97,8	100	100	Glanzmann thrombasthenia, 273800 Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 Thrombocytopenia, neonatal alloimmune, BAK antigen related, 0
ITGB3	100	99,4	100	100	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800 {Myocardial infarction, susceptibility to}, 608446 Glanzmann thrombasthenia, 273800 Purpura, posttransfusion, 0 Thrombocytopenia, neonatal alloimmune, 0
JAK2	98,1	95,8	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 {Budd-Chiari syndrome, somatic}, 600880 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
KDSR	100	99,5	100	100	Erythrokeratodermia variabilis et progressiva 4, 617526
KLKB1	100	99,5	100	100	Fletcher factor (prekallikrein) deficiency, 612423
KNG1	100	100	100	100	[Kininogen deficiency], 228960 [High molecular weight kininogen deficiency], 228960
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215

					Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LMAN1	99,8	99,2	100	100	Combined factor V and VIII deficiency, 227300
LYST	99,6	98,3	100	100	Chediak-Higashi syndrome, 214500
LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MASTL	100	100	100	100	No OMIM disease ID
MCFD2	99,5	96,9	100	100	Factor V and factor VIII, combined deficiency of, 613625
MECOM	100	99,9	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLPH	100	98,8	100	100	Griscelli syndrome, type 3, 609227
MPIG6B	100	100	100	100	?Thrombocytopenia, anemia, and myelofibrosis, 617441
MPL	100	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977
MTHFR	97,3	96	100	100	{Schizophrenia, susceptibility to}, 181500 Homocystinuria due to MTHFR deficiency, 236250 {Neural tube defects, susceptibility to}, 601634 {Thromboembolism, susceptibility to}, 188050 {Vascular disease, susceptibility to}, 0
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYO5A	99,8	98,9	100	100	Griscelli syndrome, type 1, 214450
NBEA	92	90,6	100	100	Neurodevelopmental disorder with or without early-onset generalized epilepsy, 619157
NBEAL2	99,4	99,4	100	100	Gray platelet syndrome, 139090
NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
ORAI1	99,1	96,4	99,6	97,1	Myopathy, tubular aggregate, 2, 615883 Immunodeficiency 9, 612782
P2RY12	100	100	100	100	Bleeding disorder, platelet-type, 8, 609821

PIGA	93,8	86,7	100	100	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PLA2G4A	99,9	99,4	100	100	Gastrointestinal ulceration, recurrent, with dysfunctional platelets, 618372
PLA2G7	99,9	99	100	100	{Asthma, susceptibility to}, 600807 Platelet-activating factor acetylhydrolase deficiency, 614278 {Atopy, susceptibility to}, 147050
PLAT	100	99,1	100	100	Hyperfibrinolysis, familial, due to increased release of PLAT, 612348 Thrombophilia, familial, due to decreased release of PLAT, 612348
PLAU	100	99,8	100	100	Quebec platelet disorder, 601709 {Alzheimer disease, late-onset, susceptibility to}, 104300
PLG	87,8	87,5	100	100	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PRKACG	100	99,9	100	100	?Bleeding disorder, platelet-type, 19, 616176
PROC	100	100	100	100	Thrombophilia due to protein C deficiency, autosomal dominant, 176860 Thrombophilia due to protein C deficiency, autosomal recessive, 612304
PROS1	96,7	92,1	98,4	98,4	Thrombophilia due to protein S deficiency, autosomal recessive, 614514 Thrombophilia due to protein S deficiency, autosomal dominant, 612336
PROZ	100	99,8	100	100	[Protein Z deficiency], 614024
PTGS1	100	99,8	100	100	No OMIM disease ID
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN22	99,5	97,1	100	100	{Diabetes, type 1, susceptibility to}, 222100 {Systemic lupus erythematosus susceptibility to}, 152700 {Rheumatoid arthritis, susceptibility to}, 180300
PTPRJ	97,7	97,2	99,9	99,6	Colon cancer, somatic, 114500
RAB27A	100	100	100	100	Griscelli syndrome, type 2, 607624
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RASGRP2	99,7	97,3	100	100	?Bleeding disorder, platelet-type, 18, 615888
RBM8A	99,8	97,9	100	100	Thrombocytopenia-absent radius syndrome, 274000
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RNU4ATAC	NC	NC	NC	NC	Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651

RUNX1	99,3	94,9	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 {Transcription of plasminogen activator inhibitor, modulator of}, 0
SERPINF2	100	99,8	100	100	Alpha-2-plasmin inhibitor deficiency, 262850
SH2B3	99,4	95,1	100	99,9	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SLFN14	100	100	100	100	Bleeding disorder, platelet-type, 20, 616913
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100	99,2	100	100	Noonan syndrome 9, 616559
SRC	100	99,8	100	100	Colon cancer, advanced, somatic, 114500 ?Thrombocytopenia 6, 616937
STAB2	100	99,9	100	100	No OMIM disease ID
STIM1	99,8	98	100	100	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STXBP2	82,1	79,7	99,3	97,1	Hemophagocytic lymphohistiocytosis, familial, 5, 613101
TALDO1	100	97,9	100	100	Transaldolase deficiency, 606003
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBXA2R	97,6	93,8	99,8	98	{Bleeding disorder, platelet-type, 13, susceptibility to}, 614009
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
THBD	100	99,7	100	100	Thrombophilia due to thrombomodulin defect, 614486 {Hemolytic uremic syndrome, atypical, susceptibility to, 6}, 612926
THPO	81,4	81	100	100	Thrombocythemia 1, 187950
TNXB	99,1	93,7	100	99,9	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TPM4	82,9	70,4	100	100	No OMIM disease ID
TREX1	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 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	100	93	93	Warfarin resistance, 122700 Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473
VPS33B	100	100	100	100	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
VWF	99,8	98,6	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	95,9	85,3	100	99,8	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WIPF1	100	99,9	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.
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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors