INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DG-3.9.0 (188 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCB7	99.8%	99.3%	98.3%	74.8%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100.0%	100.0%	100.0%	98.8%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100.0%	100.0%	100.0%	98.3%	Retinal dystrophy with leukodystrophy, 618863
ACD	100.0%	100.0%	100.0%	98.0%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ALAS2	100.0%	99.8%	98.3%	72.9%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752
AMN	100.0%	100.0%	100.0%	97.9%	Imerslund-Grasbeck syndrome 2, 618882

ANKRD26	97.2%	97.2%	100.0%	97.0%	Thrombocytopenia 2, 188000
AP3B1	100.0%	100.0%	100.0%	98.8%	Hermansky-Pudlak syndrome 2, 608233
ASXL1	100.0%	100.0%	100.0%	99.1%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATR	100.0%	100.0%	100.0%	98.2%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	100.0%	100.0%	100.0%	98.3%	Bloom syndrome, 210900
BRAF	100.0%	100.0%	99.9%	96.7%	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
BRCA1	100.0%	100.0%	100.0%	98.3%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320

BRCA2	100.0%	100.0%	100.0%	97.1%	Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070
BRIP1	100.0%	100.0%	100.0%	97.5%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
C15orf41	100.0%	99.9%	100.0%	99.0%	Dyserythropoietic anemia, congenital, type Ib, 615631
CAD	100.0%	100.0%	100.0%	99.4%	Developmental and epileptic encephalopathy 50, 616457
CALR	100.0%	100.0%	100.0%	99.1%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CASP10	100.0%	100.0%	100.0%	98.1%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027

CBL	100.0%	100.0%	100.0%	98.2%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CDAN1	100.0%	100.0%	99.9%	96.7%	Dyserythropoietic anemia, congenital, type Ia, 224120
CEBPA	100.0%	100.0%	98.8%	70.8%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626
CLPB	100.0%	100.0%	99.9%	98.3%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
COX4I2	100.0%	100.0%	100.0%	98.8%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100.0%	100.0%	100.0%	99.5%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830

CTC1	100.0%	100.0%	100.0%	98.8%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100.0%	100.0%	100.0%	98.8%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100;{Diabetes mellitus, insulin-dependent, 12}, 601388;{Celiac disease, susceptibility to, 3}, 609755;{Hashimoto thyroiditis}, 140300;{Systemic lupus erythematosus, susceptibility to}, 152700
CUBN	100.0%	100.0%	100.0%	99.2%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CXCR2	100.0%	100.0%	100.0%	99.4%	?WHIM syndrome 2, 619407
CXCR4	100.0%	100.0%	100.0%	97.4%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
DBF4	100.0%	100.0%	99.9%	96.4%	
DCLRE1B	100.0%	100.0%	100.0%	98.9%	Dyskeratosis congenita, autosomal recessive 8, 620133

DDX41	100.0%	100.0%	100.0%	99.5%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DHFR	100.0%	100.0%	100.0%	98.0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100.0%	100.0%	100.0%	98.5%	Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295
DIS3	100.0%	100.0%	100.0%	97.3%	
DKC1	100.0%	100.0%	97.8%	71.3%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNAJC21	100.0%	100.0%	99.8%	95.0%	Bone marrow failure syndrome 3, 617052
EFL1	100.0%	100.0%	100.0%	99.0%	Shwachman-Diamond syndrome 2, 617941
ELANE	100.0%	100.0%	100.0%	99.5%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700

EPO	100.0%	100.0%	100.0%	98.7%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
ERCC4	100.0%	100.0%	100.0%	97.7%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC6L2	100.0%	99.9%	100.0%	98.1%	Bone marrow failure syndrome 2, 615715
ETV6	100.0%	100.0%	100.0%	98.4%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EZH2	100.0%	100.0%	100.0%	99.0%	Weaver syndrome, 277590
FANCA	100.0%	100.0%	100.0%	98.4%	Fanconi anemia, complementation group A, 227650
FANCB	100.0%	100.0%	96.3%	67.0%	Fanconi anemia, complementation group B, 300514
FANCC	100.0%	100.0%	100.0%	98.7%	Fanconi anemia, complementation group C, 227645

FANCD2	100.0%	100.0%	100.0%	98.7%	Fanconi anemia, complementation group D2, 227646
FANCE	100.0%	100.0%	100.0%	98.0%	Fanconi anemia, complementation group E, 600901
FANCF	100.0%	100.0%	100.0%	98.2%	Fanconi anemia, complementation group F, 603467
FANCG	100.0%	100.0%	100.0%	98.4%	Fanconi anemia, complementation group G, 614082
FANCI	100.0%	100.0%	100.0%	98.4%	Fanconi anemia, complementation group I, 609053
FANCL	100.0%	100.0%	100.0%	98.5%	Fanconi anemia, complementation group L, 614083
FANCM	100.0%	100.0%	100.0%	97.3%	?Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAS	100.0%	100.0%	100.0%	97.3%	Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859;Squamous cell carcinoma, burn scar-related, somatic,

FASLG	100.0%	100.0%	100.0%	99.7%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
G6PC3	100.0%	100.0%	100.0%	99.5%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
GALE	100.0%	100.0%	100.0%	99.3%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GATA1	100.0%	100.0%	97.4%	69.1%	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;Hemolytic anemia due to elevated adenosine deaminase, 301083

GATA2	100.0%	100.0%	100.0%	99.2%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GBA	100.0%	100.0%	100.0%	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
GFI1	100.0%	100.0%	100.0%	98.3%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS4	100.0%	100.0%	100.0%	99.0%	

GP1BA	100.0%	100.0%	99.8%	95.4%	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	100.0%	100.0%	100.0%	98.6%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GRHL2	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100.0%	100.0%	100.0%	98.7%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100.0%	100.0%	100.0%	97.8%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HEATR3	100.0%	100.0%	100.0%	97.3%	Diamond-Blackfan anemia 21, 620072
HOXA11	100.0%	100.0%	100.0%	96.1%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432

IKZF1	100.0%	100.0%	100.0%	99.4%	Immunodeficiency, common variable, 13, 616873
IKZF2	100.0%	100.0%	100.0%	99.0%	
IKZF5	100.0%	100.0%	100.0%	97.6%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100.0%	100.0%	100.0%	99.2%	Isovaleric acidemia, 243500
JAGN1	100.0%	100.0%	100.0%	99.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
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
KDM1A	100.0%	100.0%	100.0%	97.9%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KIF23	100.0%	100.0%	100.0%	98.4%	Anemia, congenital dyserythropoietic, type IIIA, 105600

KIT	100.0%	100.0%	100.0%	99.2%	Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626
KLF1	100.0%	100.0%	100.0%	98.5%	Blood group--Lutheran inhibitor, 111150;Dyserythropoietic anemia, congenital, type IV, 613673;[Hereditary persistence of fetal hemoglobin], 613566

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
LAPTM5	100.0%	100.0%	100.0%	99.1%	
LIG4	100.0%	100.0%	100.0%	97.9%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LPIN2	100.0%	100.0%	100.0%	98.6%	Majeed syndrome, 609628
MAD2L2	100.0%	100.0%	100.0%	99.2%	?Fanconi anemia, complementation group V, 617243

MBD4	100.0%	100.0%	100.0%	98.0%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MCM4	95.3%	95.3%	100.0%	98.6%	Immunodeficiency 54, 609981
MDM4	100.0%	100.0%	100.0%	98.4%	?Bone marrow failure syndrome 6, 618849
MECOM	100.0%	100.0%	100.0%	98.9%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLH1	100.0%	100.0%	100.0%	97.6%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MPL	100.0%	100.0%	100.0%	98.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MSH2	100.0%	100.0%	100.0%	98.0%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096

MSH6	100.0%	100.0%	100.0%	98.1%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MVK	90.4%	90.4%	100.0%	99.7%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
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
MYSM1	100.0%	100.0%	100.0%	98.0%	Bone marrow failure syndrome 4, 618116
NAF1	100.0%	100.0%	99.9%	94.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NBEAL2	100.0%	100.0%	100.0%	99.4%	Gray platelet syndrome, 139090
NBN	100.0%	100.0%	100.0%	97.0%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260

NF1	100.0%	100.0%	100.0%	98.6%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFE2	100.0%	100.0%	100.0%	99.0%	
NHP2	100.0%	100.0%	100.0%	98.7%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100.0%	100.0%	100.0%	96.3%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NPAT	100.0%	100.0%	100.0%	98.2%	
NPM1	100.0%	100.0%	100.0%	96.4%	Leukemia, acute myeloid, somatic, 601626

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
PALB2	100.0%	100.0%	100.0%	96.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PARN	97.0%	95.9%	100.0%	98.5%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP4	100.0%	100.0%	100.0%	98.4%	

PAX5	100.0%	100.0%	100.0%	99.2%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PMS2	100.0%	100.0%	99.3%	95.1%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
POT1	100.0%	100.0%	99.9%	98.3%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
PRDX2	100.0%	100.0%	100.0%	99.4%	
PRF1	100.0%	100.0%	100.0%	99.4%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PTPN11	100.0%	100.0%	100.0%	98.2%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785

RAD51	89.3%	89.3%	100.0%	99.7%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAD51C	100.0%	100.0%	100.0%	97.9%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RBBP6	100.0%	100.0%	100.0%	96.6%	
RBM8A	100.0%	100.0%	99.9%	97.6%	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100.0%	100.0%	100.0%	99.2%	?Fanconi anemia, complementation group W, 617784
RMRP					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RPA1	100.0%	100.0%	100.0%	99.4%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPL11	100.0%	100.0%	100.0%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL15	99.6%	96.8%	100.0%	99.1%	Diamond-Blackfan anemia 12, 615550

RPL18	100.0%	100.0%	100.0%	98.9%	?Diamond-Blackfan anemia 18, 618310
RPL26	100.0%	100.0%	100.0%	99.2%	?Diamond-Blackfan anemia 11, 614900
RPL27	100.0%	100.0%	100.0%	98.6%	?Diamond-Blackfan anemia 16, 617408
RPL31	100.0%	100.0%	100.0%	99.1%	
RPL35	100.0%	100.0%	100.0%	99.4%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100.0%	100.0%	100.0%	99.0%	Diamond-Blackfan anemia 5, 612528
RPL4	100.0%	100.0%	100.0%	98.2%	
RPL5	100.0%	100.0%	100.0%	98.7%	Diamond-Blackfan anemia 6, 612561
RPL9	100.0%	100.0%	100.0%	98.3%	
RPS10	100.0%	100.0%	100.0%	97.5%	Diamond-Blackfan anemia 9, 613308
RPS15A	79.7%	79.7%	100.0%	95.2%	?Diamond-Blackfan anemia 20, 618313
RPS17	100.0%	100.0%	100.0%	97.2%	Diamond-Blackfan anemia 4, 612527
RPS19	100.0%	100.0%	100.0%	97.9%	Diamond-Blackfan anemia 1, 105650
RPS24	100.0%	100.0%	100.0%	98.6%	Diamond-blackfan anemia 3, 610629
RPS26	100.0%	98.8%	100.0%	98.2%	Diamond-Blackfan anemia 10, 613309

RPS27	100.0%	100.0%	100.0%	97.8%	?Diamond-Blackfan anemia 17, 617409
RPS28	100.0%	100.0%	100.0%	98.0%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100.0%	100.0%	100.0%	97.6%	Diamond-Blackfan anemia 13, 615909
RPS7	100.0%	100.0%	100.0%	96.4%	Diamond-Blackfan anemia 8, 612563
RTEL1	100.0%	100.0%	100.0%	99.4%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
RUNX1	100.0%	100.0%	100.0%	97.7%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
SAMD9	100.0%	100.0%	100.0%	97.2%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053

SAMD9L	100.0%	100.0%	100.0%	98.1%	Ataxia-pancytopenia syndrome, 159550;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270;Spinocerebellar ataxia 49, 619806
SBDS	100.0%	100.0%	100.0%	97.9%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SEC23B	100.0%	100.0%	100.0%	98.4%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SH2B3	100.0%	100.0%	100.0%	97.7%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100.0%	100.0%	99.8%	82.0%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100.0%	100.0%	100.0%	99.5%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	100.0%	100.0%	100.0%	99.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC37A4	100.0%	100.0%	100.0%	99.7%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC46A1	100.0%	100.0%	100.0%	98.7%	Folate malabsorption, hereditary, 229050
SLX4	100.0%	100.0%	100.0%	99.0%	Fanconi anemia, complementation group P, 613951
SOS1	100.0%	100.0%	100.0%	96.8%	Noonan syndrome 4, 610733;?Fibromatosis, gingival, 1, 135300
SRP54	100.0%	100.0%	100.0%	99.0%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100.0%	100.0%	100.0%	98.7%	Bone marrow failure syndrome 1, 614675
STIM1	100.0%	100.0%	100.0%	99.2%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STN1	100.0%	100.0%	100.0%	98.7%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	100.0%	100.0%	96.7%	66.1%	Barth syndrome, 302060
TBXAS1	100.0%	100.0%	100.0%	98.7%	Ghosal hematodiaphyseal syndrome, 231095

TCIRG1	100.0%	100.0%	100.0%	99.6%	Osteopetrosis, autosomal recessive 1, 259700
TERC					Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	99.7%	96.0%	100.0%	97.9%	
TERT	100.0%	100.0%	100.0%	99.8%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TET2	100.0%	99.4%	100.0%	98.7%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
THPO	100.0%	100.0%	100.0%	98.2%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481

TINF2	100.0%	100.0%	100.0%	98.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TLR8	100.0%	100.0%	97.8%	69.0%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TP53	94.7%	94.7%	100.0%	97.7%	{Basal cell carcinoma 7}, 614740;{Adrenocortical carcinoma, pediatric}, 202300;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome, 151623;Pancreatic cancer, somatic, 260350;Nasopharyngeal carcinoma, somatic, 607107;{Osteosarcoma}, 259500;{Choroid plexus papilloma}, 260500;{Colorectal cancer}, 114500;{Glioma susceptibility 1}, 137800;Bone marrow failure syndrome 5, 618165
TPM4	100.0%	100.0%	99.9%	97.2%	Bleeding disorder, platelet-type, 25, 620486
TSR2	100.0%	100.0%	97.7%	71.5%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946

TUBB1	100.0%	100.0%	100.0%	99.1%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112
TYK2	100.0%	100.0%	100.0%	99.3%	Immunodeficiency 35, 611521
UBA1	100.0%	99.7%	98.9%	73.2%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UBE2T	100.0%	100.0%	100.0%	98.7%	Fanconi anemia, complementation group T, 616435
USB1	100.0%	100.0%	100.0%	98.6%	Poikiloderma with neutropenia, 604173
VPS13B	99.6%	99.2%	100.0%	98.7%	Cohen syndrome, 216550
VPS45	95.1%	95.1%	100.0%	98.2%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100.0%	100.0%	100.0%	98.0%	CIMDAG syndrome, 619273
WAS	100.0%	98.8%	97.8%	67.8%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WRAP53	100.0%	100.0%	100.0%	98.3%	Dyskeratosis congenita, autosomal recessive 3, 613988

XRCC2	100.0%	100.0%	100.0%	99.1%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
YARS2	100.0%	100.0%	100.0%	97.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	100.0%	100.0%	100.0%	96.5%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674

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