INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DG-4.1.0 (192 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*
ABCB7	100%	99.7%	99.1%	91.3%	74.9%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100%	100%	100%	99.9%	99.2%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	85.6%	85.6%	100%	99.9%	99.4%	Retinal dystrophy with leukodystrophy, 618863
ACD	100%	100%	100%	99.9%	98.9%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ALAS2	100%	99.3%	99%	88.9%	71.3%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752

AMN	100%	100%	100%	99.7%	97.2%	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	100%	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000
AP3B1	100%	100%	100%	100%	99.7%	Hermansky-Pudlak syndrome 2, 608233
ASXL1	100%	100%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATR	100%	100%	99.7%	99.3%	98.7%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	96.8%	96.6%	100%	100%	99.7%	Bloom syndrome, 210900
BRAF	100%	100%	99.1%	97.9%	96.2%	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

BRCA1	100%	100%	100%	100%	99.7%	Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	100%	100%	100%	100%	99.7%	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	96%	96%	100%	100%	99.7%	Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480
CAD	100%	100%	100%	99.9%	98.8%	Developmental and epileptic encephalopathy 50, 616457

CALR	100%	100%	100%	99.9%	99.3%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CASP10	100%	99.5%	100%	100%	99.6%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027
CBL	100%	100%	100%	100%	99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CDAN1	100%	100%	100%	99.8%	98.4%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDIN1	100%	100%	100%	100%	99.6%	Dyserythropoietic anemia, congenital, type Ib, 615631
CEBPA	100%	100%	99.6%	93.5%	80.9%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626

CLPB	100%	100%	100%	99.9%	98.9%	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%	100%	100%	99.9%	98.7%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100%	100%	100%	100%	99.3%	Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830
CTC1	100%	100%	100%	100%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199

CTLA4	93.2%	93.2%	100%	100%	99.4%	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%	100%	100%	100%	99.6%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CXCR2	100%	100%	100%	100%	99.6%	?WHIM syndrome 2, 619407
CXCR4	99%	99%	100%	100%	99.2%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYCS	100%	100%	100%	100%	99.9%	Thrombocytopenia 4, 612004
DBF4	100%	100%	100%	100%	99.5%	
DCLRE1B	100%	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133

DDX41	100%	100%	100%	99.9%	98.7%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DHFR	100%	100%	100%	100%	99.5%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	99.7%	
DKC1	100%	99.4%	99%	89.6%	70.2%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNAJC21	100%	100%	100%	99.7%	98.4%	Bone marrow failure syndrome 3, 617052

DUT	100%	100%	100%	100%	98.9%	Bone marrow failure and diabetes mellitus syndrome, 620044
EFL1	99.2%	99.2%	100%	100%	99.8%	Shwachman-Diamond syndrome 2, 617941
ELANE	100%	100%	100%	100%	98.6%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	100%	100%	100%	99.9%	98.8%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
ERCC4	100%	100%	100%	100%	99.5%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC6L2	100%	100%	100%	100%	99.8%	Bone marrow failure syndrome 2, 615715
ERG	100%	100%	100%	100%	99.6%	Lymphatic malformation 14, 620602

ETV6	100%	100%	100%	99.9%	99.5%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EZH2	100%	100%	100%	100%	99.5%	Weaver syndrome, 277590
FANCA	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group A, 227650
FANCB	96.2%	95.9%	99.1%	91%	73.4%	Fanconi anemia, complementation group B, 300514
FANCC	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	100%	98.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	99.4%	Fanconi anemia, complementation group F, 603467
FANCG	100%	100%	100%	99.9%	98.9%	Fanconi anemia, complementation group G, 614082
FANCI	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group I, 609053

FANCL	91%	88.1%	99.8%	98.7%	97.1%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAS	100%	99.8%	100%	99.9%	99.3%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	99.9%	99.3%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980
G6PC3	96.7%	96.7%	100%	100%	99.7%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
GALE	100%	100%	100%	100%	99.2%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350

GATA1	100%	99.7%	98%	86.1%	66.6%	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%	99.9%	99.3%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286

GBA1	100%	100%	100%	100%	99.1%	{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%	100%	100%	99.9%	99.2%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS4	100%	100%	100%	100%	99.8%	

GP1BA	100%	100%	99.9%	99.3%	96.9%	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%	99.5%	96.1%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200
GRHL2	100%	100%	100%	100%	99.5%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100%	100%	100%	100%	99.6%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	99.4%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HEATR3	100%	100%	100%	100%	99.3%	Diamond-Blackfan anemia 21, 620072
HOXA11	100%	100%	100%	99.9%	97.5%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	100%	100%	100%	100%	98.6%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	100%	100%	99.6%	
IKZF5	100%	100%	100%	100%	99.6%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100%	100%	100%	100%	99.4%	Isovaleric acidemia, 243500
JAGN1	100%	100%	100%	100%	99.7%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
JAK2	100%	100%	100%	100%	99.5%	{Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300

KDM1A	96.9%	96.9%	100%	100%	99.3%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990
KIF23	100%	100%	100%	100%	99.7%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KIT	100%	100%	100%	100%	99.8%	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%	100%	100%	99.8%	98.6%	Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969

KRAS	100%	100%	100%	100%	99.8%	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%	99.8%	100%	100%	99.7%	
LCP1	100%	100%	100%	100%	99.8%	

LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LPIN2	99.5%	99.2%	100%	100%	99.5%	Majeed syndrome, 609628
MAD2L2	100%	100%	100%	100%	99.1%	?Fanconi anemia, complementation group V, 617243
MBD4	100%	100%	100%	100%	99.8%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MCM4	95.3%	95.3%	100%	100%	99.6%	Immunodeficiency 54, 609981
MDM4	100%	100%	100%	100%	99.6%	?Bone marrow failure syndrome 6, 618849
MECOM	100%	100%	100%	99.9%	99.6%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLH1	100%	100%	100%	99.9%	99.1%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300

MPL	100%	100%	100%	99.9%	98.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MSH2	100%	100%	100%	100%	99.3%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	100%	99.9%	98.4%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MVK	100%	100%	100%	100%	99.2%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYH9	97.3%	97.2%	100%	99.9%	99.1%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622

MYSM1	100%	100%	100%	100%	99.8%	Bone marrow failure syndrome 4, 618116
NAF1	100%	100%	100%	99.9%	99%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NBEAL2	100%	100%	100%	100%	99.1%	Gray platelet syndrome, 139090
NBN	97.5%	97.5%	100%	100%	99.9%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NF1	99.4%	99.4%	100%	100%	99.7%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFE2	100%	100%	100%	100%	99.2%	
NHP2	100%	100%	100%	99.9%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987

NOP10	92.5%	92.5%	100%	100%	98.8%	?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%	100%	100%	100%	99.8%	
NPM1	87.6%	87.6%	100%	99.9%	98.7%	Leukemia, acute myeloid, somatic, 601626

NRAS	100%	100%	100%	100%	99.6%	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%	100%	100%	100%	99.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832

PARN	97.3%	95.3%	100%	100%	99.7%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP4	100%	100%	100%	100%	99.5%	
PAX5	100%	100%	100%	100%	99.1%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PMS2	93.7%	93.4%	100%	99.7%	98.8%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
POT1	100%	100%	100%	100%	99.8%	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%	100%	100%	100%	99.5%	

PRF1	100%	100%	100%	99.9%	99%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PTPN11	89.7%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89.3%	89.3%	100%	100%	99.4%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244
RAD51C	90.3%	90.3%	100%	100%	99.4%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RBBP6	100%	100%	100%	100%	99.6%	
RBM8A	100%	100%	100%	100%	99.6%	Thrombocytopenia-absent radius syndrome, 274000

RFWD3	100%	100%	100%	100%	99.8%	?Fanconi anemia, complementation group W, 617784
RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RPA1	100%	100%	100%	100%	99.3%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPL11	100%	100%	100%	100%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL15	97.6%	89.4%	100%	100%	99.6%	Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	100%	98.8%	?Diamond-Blackfan anemia 18, 618310
RPL26	100%	100%	100%	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	100%	99.8%	100%	99.8%	99.3%	?Diamond-Blackfan anemia 16, 617408
RPL31	100%	99.7%	100%	100%	99.8%	
RPL35	100%	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	100%	100%	99.8%	Diamond-Blackfan anemia 5, 612528
RPL4	100%	100%	100%	100%	99.6%	

RPL5	100%	100%	100%	100%	99.8%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	100%	100%	99.9%	
RPS10	100%	100%	100%	100%	99.5%	Diamond-Blackfan anemia 9, 613308
RPS15A	79.7%	79.7%	100%	100%	99.8%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	100%	100%	99.1%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	100%	100%	99.2%	Diamond-Blackfan anemia 1, 105650
RPS24	100%	100%	100%	100%	99.7%	Diamond-blackfan anemia 3, 610629
RPS26	100%	98.1%	100%	100%	99.7%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	100%	100%	99.5%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	100%	100%	99.2%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	100%	100%	99.7%	Diamond-Blackfan anemia 13, 615909
RPS7	100%	100%	100%	99.8%	98.2%	Diamond-Blackfan anemia 8, 612563

RTEL1	100%	100%	100%	100%	99.1%	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%	100%	99%	96.9%	92.2%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
SAMD9	100%	100%	100%	100%	99.6%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMD9L	100%	100%	100%	100%	99.8%	Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270

SBDS	100%	100%	100%	100%	99.7%	{Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400
SEC23B	100%	100%	100%	100%	99.7%	?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SH2B3	100%	100%	100%	99.9%	98.7%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	99.1%	92.1%	73.8%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100%	100%	100%	100%	99.8%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	100%	100%	100%	100%	99.7%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100%	100%	100%	100%	99.5%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240

SLC46A1	100%	100%	100%	99.9%	98.7%	Folate malabsorption, hereditary, 229050
SLX4	100%	100%	100%	99.9%	99%	Fanconi anemia, complementation group P, 613951
SOS1	98.7%	98.3%	100%	100%	99.7%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SRP54	100%	100%	100%	100%	99.8%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100%	100%	100%	100%	99.5%	Bone marrow failure syndrome 1, 614675
STIM1	100%	99.8%	100%	100%	99.2%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STN1	87.1%	87%	100%	100%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAFAZZIN	100%	99.6%	97.2%	85.8%	66.7%	Barth syndrome, 302060
TBXAS1	100%	100%	100%	100%	99.4%	Ghosal hematodiaphyseal syndrome, 231095

TCIRG1	100%	100%	100%	99.9%	99%	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	98.5%	91.1%	100%	100%	99%	
TERT	100%	100%	100%	99.9%	98.4%	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%	100%	100%	100%	99.6%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126

THPO	100%	100%	100%	100%	99.6%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TINF2	100%	100%	100%	100%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TLR8	100%	100%	99.1%	91.9%	75.4%	Immunodeficiency 98 with autoinflammation, X-linked, 301078

TP53	94.7%	94.7%	100%	99.8%	98.8%	{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%	99.8%	100%	99.9%	98.4%	Bleeding disorder, platelet-type, 25, 620486
TSR2	100%	100%	99.4%	89.2%	71.2%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TUBB1	100%	100%	100%	99.9%	99.3%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112

TYK2	100%	100%	100%	99.9%	98.1%	Immunodeficiency 35, 611521
UBA1	99.9%	99.3%	98.7%	88.4%	68.8%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UBE2T	100%	100%	100%	99.9%	99%	Fanconi anemia, complementation group T, 616435
USB1	95.5%	93.2%	100%	100%	99.2%	Poikiloderma with neutropenia, 604173
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS45	87.3%	86.8%	100%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100%	100%	100%	99.7%	96.7%	CIMDAG syndrome, 619273
WAS	97.7%	89.9%	97.7%	85.6%	65.4%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900

WRAP53	100%	100%	100%	99.9%	98.8%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	100%	100%	100%	100%	99.5%	Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247
YARS2	100%	100%	100%	100%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	100%	100%	100%	100%	99.5%	?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 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 4.1.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors