INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DG-5.0.0 (204 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%	100%	99.2%	90.7%	71.4%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100%	99.9%	100%	99.9%	99.2%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	85.6%	85.6%	100%	100%	99.7%	Retinal dystrophy with leukodystrophy, 618863
ACD	100%	100%	100%	99.9%	99.2%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ADA2	96.2%	93.5%	100%	100%	99.6%	Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ALAS2	100%	100%	98.4%	87.7%	67.8%	Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752
AMN	100%	100%	100%	100%	99.4%	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	100%	100%	100%	100%	99.7%	Thrombocytopenia 2, 188000
AP3B1	100%	100%	100%	100%	99.9%	Hermansky-Pudlak syndrome 2, 608233

ASXL1	100%	100%	100%	99.9%	99.4%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ATR	100%	100%	99.8%	99.4%	98.9%	Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	95.2%	94.7%	100%	100%	99.8%	Bloom syndrome, 210900
BRAF	100%	100%	100%	99.7%	98.3%	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
CA2	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAD	100%	99.1%	100%	99.9%	99.2%	Developmental and epileptic encephalopathy 50, 616457
CALR	100%	100%	100%	100%	99.6%	Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950
CASP10	100%	100%	100%	100%	99.9%	Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027
CBL	100%	100%	100%	100%	99.5%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CDAN1	100%	100%	100%	100%	99.4%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDIN1	100%	100%	100%	100%	99.8%	Dyserythropoietic anemia, congenital, type Ib, 615631
CEBPA	100%	100%	100%	99.4%	93.6%	Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626

CLCN7	97.5%	97.5%	100%	100%	99.4%	Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600
CLPB	98.2%	98.2%	100%	99.9%	99.5%	Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835
COPZ1	90%	86.7%	100%	100%	99.4%	
COX4I2	100%	99.6%	100%	100%	99.2%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100%	100%	100%	100%	99.4%	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%	99.5%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CUBN	100%	100%	100%	100%	99.8%	[Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100
CXCR2	100%	100%	100%	100%	98.8%	?WHIM syndrome 2, 619407

CXCR4	99%	99%	100%	100%	99.9%	WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670
CYCS	100%	100%	100%	100%	100%	Thrombocytopenia 4, 612004
DBF4	99.9%	98.5%	100%	99.9%	99.4%	
DCLRE1B	100%	100%	100%	100%	99.5%	Dyskeratosis congenita, autosomal recessive 8, 620133
DDX41	100%	100%	100%	99.9%	99.2%	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DHFR	100%	100%	100%	100%	99.7%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100%	100%	100%	100%	99.8%	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%	99.6%	
DKC1	100%	100%	98.8%	87.4%	68.7%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DNAJC21	95%	94.7%	100%	99.9%	99.3%	Bone marrow failure syndrome 3, 617052
DUT	100%	100%	100%	100%	100%	Bone marrow failure and diabetes mellitus syndrome, 620044

EFL1	96.3%	96.3%	100%	100%	99.8%	Shwachman-Diamond syndrome 2, 617941
ELANE	100%	100%	100%	100%	99.4%	Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	100%	100%	100%	100%	99.5%	{Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911
ERCC4	100%	100%	100%	100%	99.9%	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%	99.4%	Lymphatic malformation 14, 620602
ETV6	100%	100%	100%	100%	99.8%	Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626
EZH2	100%	100%	100%	100%	99.8%	Weaver syndrome, 277590
FAAP100	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group X, 621258
FANCA	100%	100%	100%	100%	99.3%	Fanconi anemia, complementation group A, 227650

FANCB	96.2%	96.2%	98.9%	90.2%	70.7%	Fanconi anemia, complementation group B, 300514
FANCC	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	99.9%	99.9%	99.3%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	100%	100%	99.3%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	99.8%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99.8%	100%	99.9%	99.2%	Fanconi anemia, complementation group G, 614082
FANCI	100%	100%	100%	100%	99.5%	Fanconi anemia, complementation group I, 609053
FANCL	91.8%	91.1%	100%	99.5%	98.2%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAS	100%	100%	100%	100%	99.4%	Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859
FASLG	100%	100%	100%	100%	99.6%	Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980

G6PC3	96.8%	96.8%	100%	100%	99.8%	Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541
GALE	100%	100%	100%	100%	99.8%	Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350
GATA1	100%	100%	98.3%	86.3%	64.4%	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%	100%	99.2%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286

GBA1	100%	98.8%	100%	99.9%	99.2%	{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%	100%	99.6%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS4	100%	100%	100%	100%	99.9%	
GP1BA	100%	100%	100%	99.6%	97.4%	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%	100%	100%	99.8%	96.6%	Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200

GRHL2	100%	100%	100%	99.9%	99.7%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100%	100%	100%	100%	99.7%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	100%	100%	99.5%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HEATR3	100%	100%	100%	99.9%	99.8%	Diamond-Blackfan anemia 21, 620072
HOXA11	100%	100%	100%	99.8%	99.2%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	100%	100%	100%	99.9%	99.1%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	100%	100%	99.8%	Immunodysregulation, craniofacial anomalies, hearing impairment, athelia, and developmental delay, 621234;Immunodysregulation with variable immunodeficiency and autoimmunity, 621233
IKZF5	100%	100%	100%	100%	99.8%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	90.5%	90%	100%	100%	99.7%	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	93.5%	93.5%	100%	100%	99.6%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990
KIF23	97.7%	97.7%	100%	100%	99.8%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KIT	100%	100%	100%	99.9%	99.7%	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%	100%	99.9%	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%	99.7%	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%	100%	100%	100%	99.8%	
LCP1	100%	100%	100%	100%	99.7%	
LIG4	100%	100%	100%	100%	99.9%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
LPIN2	99.6%	99.2%	100%	100%	99.6%	Majeed syndrome, 609628
MAD2L2	100%	100%	100%	99.9%	98.6%	?Fanconi anemia, complementation group V, 617243
MBD4	100%	100%	100%	100%	99.9%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MCM4	95.4%	95.4%	100%	99.9%	99.5%	Immunodeficiency 54, 609981

MDM4	100%	100%	100%	100%	99.6%	?Bone marrow failure syndrome 6, 618849
MECOM	100%	100%	100%	100%	99.8%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLH1	100%	100%	100%	100%	99.6%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MPL	100%	100%	100%	100%	99.6%	Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977
MSH2	90.6%	90.6%	100%	100%	99.7%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	100%	100%	99.7%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MVK	100%	100%	100%	100%	99.5%	Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYH9	97.4%	97.1%	100%	99.9%	99.4%	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%	100%	99.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NBEAL2	100%	100%	100%	99.9%	99.3%	Gray platelet syndrome, 139090
NBN	97.5%	97.5%	100%	100%	99.8%	Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFE2	100%	100%	100%	100%	99.6%	
NHP2	100%	100%	100%	100%	99.9%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	92.5%	92.5%	100%	100%	99.8%	?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%	99.4%	Leukemia, acute myeloid, somatic, 601626

NRAS	89.6%	89.4%	100%	100%	99.2%	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
OSTM1	100%	100%	100%	100%	99.9%	Osteopetrosis, autosomal recessive 5, 259720
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	98.3%	96.3%	100%	100%	99.9%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
PARP4	100%	100%	100%	100%	99.7%	
PAX5	100%	100%	100%	99.8%	99%	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545

PLEKHM1	100%	100%	99.9%	99.7%	98.7%	?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107
PMS2	94.9%	93.4%	99.9%	99.8%	99.3%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
POT1	100%	100%	100%	100%	100%	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%	99.6%	
PRF1	100%	100%	100%	100%	99.4%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027
PTPN11	90.5%	89.2%	100%	100%	99.6%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89.4%	89.4%	100%	100%	99.5%	Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244

RAD51C	87.4%	87.3%	100%	100%	99.6%	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390
RBBP6	100%	100%	100%	100%	99.4%	
RBM8A	100%	100%	100%	99.6%	99%	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100%	100%	100%	100%	99.4%	?Fanconi anemia, complementation group W, 617784
RMRP						Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RPA1	96.2%	93.5%	100%	100%	99.8%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPL11	98%	94.8%	100%	100%	99.4%	Diamond-Blackfan anemia 7, 612562
RPL15	100%	99.7%	100%	100%	100%	Diamond-Blackfan anemia 12, 615550
RPL17	100%	100%	100%	100%	100%	Diamond-Blackfan anemia 22, 621262
RPL18	100%	100%	100%	100%	98.9%	?Diamond-Blackfan anemia 18, 618310
RPL26	100%	100%	100%	100%	99.9%	?Diamond-Blackfan anemia 11, 614900
RPL27	100%	100%	100%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL31	95.5%	85.7%	100%	100%	99.9%	
RPL35	82.1%	82.1%	100%	100%	98.8%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	100%	100%	100%	Diamond-Blackfan anemia 5, 612528

RPL4	100%	100%	100%	100%	99.9%	
RPL5	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	100%	100%	99.9%	
RPS10	89.4%	89.1%	100%	100%	99.8%	Diamond-Blackfan anemia 9, 613308
RPS15A	63.1%	63%	100%	100%	99.6%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	100%	100%	99.8%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	100%	100%	99.6%	Diamond-Blackfan anemia 1, 105650
RPS24	89.4%	86.9%	100%	100%	99.7%	Diamond-blackfan anemia 3, 610629
RPS26	100%	100%	100%	100%	99.4%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	100%	100%	99.7%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	100%	100%	99.8%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	100%	100%	99.5%	Diamond-Blackfan anemia 13, 615909
RPS7	100%	100%	100%	99.8%	99.6%	Diamond-Blackfan anemia 8, 612563
RTEL1	100%	100%	100%	100%	99.5%	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%	100%	99.7%	97.6%	Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626
SAMD9	100%	100%	100%	100%	99.9%	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%	99.9%	99.6%	{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%	100%	99.2%	Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	98.7%	90.8%	72.2%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100%	100%	100%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	90.6%	90.6%	100%	100%	99.5%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC37A4	100%	100%	100%	100%	99.2%	Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240
SLC46A1	100%	100%	100%	99.9%	98.9%	Folate malabsorption, hereditary, 229050
SLC4A2	100%	100%	100%	99.9%	99.1%	?Osteopetrosis, autosomal recessive 9, 620366
SLX4	100%	100%	100%	100%	99.6%	Fanconi anemia, complementation group P, 613951
SNX10	89.3%	89.3%	100%	100%	99.7%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	98.8%	98.8%	100%	100%	99.8%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SRP54	96.3%	96.3%	100%	100%	99.9%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100%	100%	100%	100%	99.9%	Bone marrow failure syndrome 1, 614675
STIM1	100%	100%	100%	100%	99.7%	Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STN1	87.1%	87.1%	100%	100%	99.8%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAFAZZIN	100%	100%	98%	87.6%	65.4%	Barth syndrome, 302060
TBXAS1	100%	100%	100%	100%	99.7%	Ghosal hematodiaphyseal syndrome, 231095

TCIRG1	100%	100%	100%	100%	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	97%	96%	100%	100%	99.7%	
TERT	99.6%	97.9%	100%	100%	99.4%	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%	99.8%	Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126
THPO	100%	100%	100%	100%	99.9%	Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TINF2	100%	100%	100%	100%	99.6%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TLR8	100%	100%	99%	91.8%	74.3%	Immunodeficiency 98 with autoinflammation, X-linked, 301078

TNFRSF11A	100%	100%	100%	100%	99.4%	Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFSF11	100%	100%	100%	100%	99.7%	Osteopetrosis, autosomal recessive 2, 259710
TP53	95.1%	95.1%	100%	99.9%	99.1%	{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	93.6%	88.5%	100%	99.9%	99%	Bleeding disorder, platelet-type, 25, 620486
TSR2	100%	100%	97.8%	87%	64.6%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TUBB1	100%	100%	100%	100%	99.7%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112
TYK2	100%	99.8%	100%	99.8%	98.5%	Immunodeficiency 35, 611521

UBA1	96.4%	95.9%	98.4%	87.4%	67.6%	Spinal muscular atrophy, X-linked 2, infantile, 301830;VEXAS syndrome, somatic, 301054
UBE2T	92.6%	92.6%	100%	100%	99.4%	Fanconi anemia, complementation group T, 616435
USB1	95.1%	93.3%	100%	100%	99.1%	Poikiloderma with neutropenia, 604173
VPS13B	100%	100%	100%	100%	99.7%	Cohen syndrome, 216550
VPS45	88.9%	87.3%	100%	100%	99.6%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100%	100%	100%	99.9%	98.1%	CIMDAG syndrome, 619273
WAS	99.6%	96.4%	97.2%	84.8%	65.5%	Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WRAP53	100%	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	100%	100%	100%	100%	99.9%	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 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors