WES INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION TO HEMATOLOGICAL MALIGNANCIES DG 3.7

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%	99.3%	77.3%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100.0%	100.0%	100.0%	99.6%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100.0%	100.0%	100.0%	99.5%	Retinal dystrophy with leukodystrophy, 618863
ACD	100.0%	100.0%	100.0%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ALAS2	100.0%	99.8%	98.9%	77.2%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
AMN	100.0%	100.0%	100.0%	100.0%	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	97.2%	97.2%	100.0%	98.6%	Thrombocytopenia 2, 188000
AP3B1	100.0%	100.0%	100.0%	99.7%	Hermansky-Pudlak syndrome 2, 608233
ASXL1	100.0%	100.0%	100.0%	99.6%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039

ATR	100.0%	100.0%	100.0%	99.2%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	100.0%	100.0%	100.0%	99.2%	Bloom syndrome, 210900
BRAF	100.0%	100.0%	100.0%	99.5%	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%	99.1%	Fanconi anemia, complementation group S, 617883
BRCA2	100.0%	100.0%	100.0%	98.6%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRIP1	100.0%	100.0%	100.0%	99.1%	Fanconi anemia, complementation group J, 609054
C15orf41	100.0%	99.9%	100.0%	99.9%	Dyserythropoietic anemia, congenital, type Ib, 615631
CAD	100.0%	100.0%	100.0%	99.8%	Developmental and epileptic encephalopathy 50, 616457
CASP10	100.0%	100.0%	100.0%	99.0%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CBL	100.0%	100.0%	100.0%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785

CDAN1	100.0%	100.0%	100.0%	99.4%	Dyserythropoietic anemia, congenital, type Ia, 224120
CEBPA	100.0%	100.0%	99.9%	92.7%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CLPB	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	98.8%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100.0%	100.0%	100.0%	99.8%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CTC1	100.0%	100.0%	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100.0%	100.0%	100.0%	99.2%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CUBN	100.0%	100.0%	100.0%	99.5%	Imerslund-Grasbeck syndrome 1, 261100
CXCR2	100.0%	100.0%	100.0%	99.8%	?WHIM syndrome 2, 619407
CXCR4	100.0%	100.0%	100.0%	99.2%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
DBF4	100.0%	100.0%	100.0%	98.4%	
DCLRE1B	100.0%	100.0%	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 8, 620133
DDX41	100.0%	100.0%	100.0%	99.8%	

DHFR	100.0%	100.0%	100.0%	99.6%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100.0%	100.0%	100.0%	99.2%	Pleuropulmonary blastoma, 601200 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295
DKC1	100.0%	100.0%	98.0%	73.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 Dyskeratosis congenita, X-linked, 305000
DNAJC21	100.0%	100.0%	100.0%	98.5%	Bone marrow failure syndrome 3, 617052
EFL1	100.0%	100.0%	100.0%	99.6%	Shwachman-Diamond syndrome 2, 617941
ELANE	100.0%	100.0%	100.0%	99.9%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	100.0%	100.0%	100.0%	99.4%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
ERCC4	100.0%	100.0%	100.0%	98.8%	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%	99.1%	Bone marrow failure syndrome 2, 615715
ETV6	100.0%	100.0%	100.0%	99.4%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626

EZH2	100.0%	100.0%	100.0%	99.6%	Weaver syndrome, 277590
FANCA	100.0%	100.0%	100.0%	99.5%	Fanconi anemia, complementation group A, 227650
FANCB	100.0%	100.0%	98.0%	71.0%	Fanconi anemia, complementation group B, 300514
FANCC	100.0%	100.0%	100.0%	99.6%	Fanconi anemia, complementation group C, 227645
FANCD2	100.0%	100.0%	100.0%	99.1%	Fanconi anemia, complementation group D2, 227646
FANCE	100.0%	100.0%	100.0%	99.6%	Fanconi anemia, complementation group E, 600901
FANCF	100.0%	100.0%	100.0%	99.3%	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%	99.2%	Fanconi anemia, complementation group I, 609053
FANCL	100.0%	100.0%	100.0%	99.3%	Fanconi anemia, complementation group L, 614083
FANCM	100.0%	100.0%	100.0%	98.7%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	100.0%	100.0%	100.0%	98.9%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100.0%	100.0%	100.0%	99.4%	Autoimmune lymphoproliferative syndrome, type IB, 601859
G6PC3	100.0%	100.0%	100.0%	99.8%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541

GALE	100.0%	100.0%	100.0%	99.8%	Galactose epimerase deficiency, 230350
GATA1	100.0%	100.0%	98.2%	72.4%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 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%	99.8%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100.0%	100.0%	100.0%	99.6%	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
GFI1	100.0%	100.0%	100.0%	99.7%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GINS4	100.0%	100.0%	100.0%	99.9%	
GP1BA	100.0%	100.0%	99.6%	95.9%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820

GP1BB	100.0%	100.0%	100.0%	100.0%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	100.0%	100.0%	100.0%	99.7%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100.0%	100.0%	100.0%	99.7%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100.0%	100.0%	100.0%	98.6%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HEATR3	100.0%	100.0%	100.0%	99.1%	Diamond-Blackfan anemia 21, 620072
HOXA11	100.0%	100.0%	100.0%	99.3%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	100.0%	100.0%	100.0%	99.7%	Immunodeficiency, common variable, 13, 616873
IKZF2	100.0%	100.0%	100.0%	99.3%	
IKZF5	100.0%	100.0%	100.0%	99.3%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100.0%	100.0%	100.0%	99.8%	Isovaleric acidemia, 243500
JAGN1	100.0%	100.0%	100.0%	99.9%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KIF23	100.0%	100.0%	100.0%	98.9%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KLF1	100.0%	100.0%	100.0%	99.9%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673

KRAS	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.6%	
LIG4	100.0%	100.0%	100.0%	99.4%	LIG4 syndrome, 606593
LPIN2	100.0%	100.0%	100.0%	99.1%	Majeed syndrome, 609628
MAD2L2	100.0%	100.0%	100.0%	99.9%	?Fanconi anemia, complementation group V, 617243
MBD4	100.0%	100.0%	100.0%	99.4%	Tumor predisposition syndrome 2, 619975
MCM4	95.3%	95.3%	100.0%	99.3%	Immunodeficiency 54, 609981
MDM4	100.0%	100.0%	100.0%	99.1%	?Bone marrow failure syndrome 6, 618849
MECOM	100.0%	100.0%	100.0%	99.5%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738

MLH1	100.0%	100.0%	100.0%	99.1%	Lynch syndrome 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MPL	100.0%	100.0%	100.0%	99.5%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MSH2	100.0%	100.0%	100.0%	98.9%	Lynch syndrome 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 2, 619096
MSH6	100.0%	100.0%	100.0%	99.4%	Lynch syndrome 5, 614350 Mismatch repair cancer syndrome 3, 619097
MVK	90.4%	90.4%	100.0%	100.0%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYH9	100.0%	100.0%	100.0%	99.6%	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%	99.1%	Bone marrow failure syndrome 4, 618116
NBEAL2	100.0%	100.0%	100.0%	99.9%	Gray platelet syndrome, 139090
NBN	100.0%	100.0%	100.0%	98.8%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260

NF1	100.0%	100.0%	100.0%	99.3%	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%	99.9%	
NHP2	100.0%	100.0%	100.0%	99.3%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.1%	
NPM1	100.0%	100.0%	100.0%	98.0%	Leukemia, acute myeloid, somatic, 601626
NRAS	100.0%	100.0%	100.0%	99.8%	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%	98.6%	Fanconi anemia, complementation group N, 610832
PARN	97.0%	95.9%	100.0%	99.5%	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%	99.2%	
PAX5	100.0%	100.0%	100.0%	99.7%	
PMS2	100.0%	100.0%	99.7%	96.6%	Lynch syndrome 4, 614337 Mismatch repair cancer syndrome 4, 619101
POT1	100.0%	100.0%	100.0%	99.5%	?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%	99.9%	
PRF1	100.0%	100.0%	100.0%	100.0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PTPN11	100.0%	100.0%	100.0%	98.9%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89.3%	89.3%	100.0%	100.0%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244

RAD51C	100.0%	100.0%	100.0%	98.9%	Fanconi anemia, complementation group O, 613390
RBBP6	100.0%	100.0%	100.0%	98.4%	
RBM8A	100.0%	100.0%	100.0%	99.2%	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100.0%	100.0%	100.0%	99.3%	?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%	99.7%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RPL11	100.0%	100.0%	100.0%	99.1%	Diamond-Blackfan anemia 7, 612562
RPL15	99.6%	96.8%	100.0%	99.7%	Diamond-Blackfan anemia 12, 615550
RPL18	100.0%	100.0%	100.0%	99.9%	?Diamond-Blackfan anemia 18, 618310
RPL26	100.0%	100.0%	100.0%	99.5%	?Diamond-Blackfan anemia 11, 614900
RPL27	100.0%	100.0%	100.0%	99.3%	?Diamond-Blackfan anemia 16, 617408
RPL31	100.0%	100.0%	100.0%	99.3%	
RPL35	100.0%	100.0%	100.0%	100.0%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100.0%	100.0%	100.0%	99.5%	Diamond-Blackfan anemia 5, 612528
RPL4	100.0%	100.0%	100.0%	99.2%	
RPL5	100.0%	100.0%	100.0%	99.5%	Diamond-Blackfan anemia 6, 612561
RPL9	100.0%	100.0%	100.0%	99.8%	
RPS10	100.0%	100.0%	100.0%	99.6%	Diamond-Blackfan anemia 9, 613308

RPS15A	79.7%	79.7%	100.0%	97.6%	?Diamond-Blackfan anemia 20, 618313
RPS17	100.0%	100.0%	100.0%	99.0%	Diamond-Blackfan anemia 4, 612527
RPS19	100.0%	100.0%	100.0%	99.5%	Diamond-Blackfan anemia 1, 105650
RPS24	100.0%	100.0%	100.0%	99.3%	Diamond-blackfan anemia 3, 610629
RPS26	100.0%	98.8%	100.0%	99.0%	Diamond-Blackfan anemia 10, 613309
RPS27	100.0%	100.0%	100.0%	99.5%	?Diamond-Blackfan anemia 17, 617409
RPS28	100.0%	100.0%	100.0%	99.7%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100.0%	100.0%	100.0%	99.1%	Diamond-Blackfan anemia 13, 615909
RPS7	100.0%	100.0%	100.0%	99.0%	Diamond-Blackfan anemia 8, 612563
RTEL1	100.0%	100.0%	100.0%	99.9%	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%	99.7%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	100.0%	100.0%	100.0%	99.0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053

SAMD9L	100.0%	100.0%	100.0%	98.9%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SBDS	100.0%	100.0%	100.0%	99.1%	Shwachman-Diamond syndrome 1, 260400
SEC23B	100.0%	100.0%	100.0%	99.3%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SH2B3	100.0%	100.0%	100.0%	99.1%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100.0%	100.0%	99.8%	79.8%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100.0%	100.0%	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	100.0%	100.0%	100.0%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100.0%	100.0%	100.0%	99.6%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC46A1	100.0%	100.0%	100.0%	99.8%	Folate malabsorption, hereditary, 229050
SLX4	100.0%	100.0%	100.0%	99.6%	Fanconi anemia, complementation group P, 613951
SOS1	100.0%	100.0%	100.0%	99.0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300

SRP54	100.0%	100.0%	100.0%	99.6%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100.0%	100.0%	100.0%	99.0%	Bone marrow failure syndrome 1, 614675
STIM1	100.0%	100.0%	100.0%	99.7%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STN1	100.0%	100.0%	100.0%	99.5%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	100.0%	100.0%	99.3%	74.1%	Barth syndrome, 302060
TBXAS1	100.0%	100.0%	100.0%	99.4%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	100.0%	100.0%	100.0%	100.0%	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%	99.5%	
TERT	100.0%	100.0%	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742
TET2	100.0%	99.4%	100.0%	99.5%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126

THPO	100.0%	100.0%	100.0%	98.3%	Thrombocythemia 1, 187950 Thrombocytopenia 9, 620478 Amegakaryocytic thrombocytopenia, congenital, 2, 620481
TINF2	100.0%	100.0%	100.0%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR8	100.0%	100.0%	98.4%	72.1%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TP53	94.7%	94.7%	100.0%	99.3%	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165
TSR2	100.0%	100.0%	98.6%	76.2%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TUBB1	100.0%	100.0%	100.0%	99.6%	Macrothrombocytopenia, isolated, 1, autosomal dominant, 613112
TYK2	100.0%	100.0%	100.0%	99.8%	Immunodeficiency 35, 611521
UBA1	100.0%	99.7%	99.0%	77.6%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UBE2T	100.0%	100.0%	100.0%	99.9%	Fanconi anemia, complementation group T, 616435
USB1	100.0%	100.0%	100.0%	98.7%	Poikiloderma with neutropenia, 604173
VPS13B	99.6%	99.2%	100.0%	99.4%	Cohen syndrome, 216550

VPS45	95.1%	95.1%	100.0%	99.4%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100.0%	100.0%	100.0%	99.6%	CIMDAG syndrome, 619273
WAS	100.0%	98.8%	98.8%	74.3%	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%	99.6%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	100.0%	100.0%	100.0%	99.5%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
YARS2	100.0%	100.0%	100.0%	99.6%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	100.0%	100.0%	100.0%	98.9%	?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 3.7.0.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors