HEREDITARY BONE MARROW FAILURE GENE PANEL DG 3.5.0 (176 genes)

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Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCB7	100%	99%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100%	100%	Retinal dystrophy with leukodystrophy, 618863
ACD	100%	100%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ALAS2	100%	100%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
AMN	100%	100%	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	97%	97%	Thrombocytopenia 2, 188000
AP3B1	100%	100%	Hermansky-Pudlak syndrome 2, 608233
ASXL1	100%	100%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ATR	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	100%	100%	Bloom syndrome, 210900
BRAF	100%	100%	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%	Fanconi anemia, complementation group S, 617883
BRCA2	100%	100%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070

BRIP1	100%	100%	Fanconi anemia, complementation group J, 609054
CAD	100%	100%	Developmental and epileptic encephalopathy 50, 616457
CASP10	100%	100%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CBL	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CDAN1	100%	100%	Dyserythropoietic anemia, congenital, type Ia, 224120
C15orf41	100%	100%	Dyserythropoietic anemia, congenital, type Ib, 615631
CEBPA	100%	100%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CLPB	100%	100%	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%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CTC1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100%	100%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CUBN	100%	100%	Imerslund-Grasbeck syndrome 1, 261100
CXCR2	100%	100%	?WHIM syndrome 2, 619407
CXCR4	100%	100%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
DBF4	100%	100%	No OMIM disease ID
DDX41	100%	100%	No OMIM disease ID
DHFR	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100%	100%	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%	100%	Dyskeratosis congenita, X-linked, 305000
DNAJC21	100%	100%	Bone marrow failure syndrome 3, 617052

EFL1	100%	100%	Shwachman-Diamond syndrome 2, 617941
ELANE	100%	100%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	100%	100%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
ERCC4	100%	100%	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%	Bone marrow failure syndrome 2, 615715
ETV6	100%	100%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EZH2	100%	100%	Weaver syndrome, 277590
FANCA	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	100%	100%	Fanconi anemia, complementation group B, 300514
FANCC	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	100%	100%	Fanconi anemia, complementation group D2, 227646
FANCE	100%	100%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	100%	100%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	100%	100%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100%	100%	Autoimmune lymphoproliferative syndrome, type IB, 601859
G6PC3	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	100%	100%	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%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100%	100%	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%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GP1BA	100%	100%	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	100%	100%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100%	100%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	100%	100%	Immunodeficiency, common variable, 13, 616873
IKZF2	100%	100%	No OMIM disease ID
IKZF5	100%	100%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100%	100%	Isovaleric acidemia, 243500
JAGN1	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KIF23	100%	100%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KLF1	100%	100%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673

KRAS	100%	100%	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%	No OMIM disease ID
LIG4	100%	100%	LIG4 syndrome, 606593
LPIN2	100%	100%	Majeed syndrome, 609628
MAD2L2	100%	100%	?Fanconi anemia, complementation group V, 617243
MBD4	100%	100%	Tumor predisposition syndrome 2, 619975
MCM4	95%	95%	Immunodeficiency 54, 609981
MDM4	100%	100%	?Bone marrow failure syndrome 6, 618849
MECOM	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLH1	100%	100%	Lynch syndrome 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MPL	100%	100%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MSH2	100%	100%	Lynch syndrome 1, 120435 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	Lynch syndrome 5, 614350 Mismatch repair cancer syndrome 3, 619097
MVK	90%	90%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377

MYH9	100%	100%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYSM1	100%	100%	Bone marrow failure syndrome 4, 618116
NBEAL2	100%	100%	Gray platelet syndrome, 139090
NBN	100%	100%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NF1	100%	100%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFE2	100%	100%	No OMIM disease ID
NHP2	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPAT	100%	100%	No OMIM disease ID
NPM1	100%	100%	Leukemia, acute myeloid, somatic, 601626
NRAS	100%	100%	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%	Fanconi anemia, complementation group N, 610832
PARN	97%	96%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARP4	100%	100%	No OMIM disease ID
PAX5	100%	100%	No OMIM disease ID
PMS2	100%	100%	Lynch syndrome 4, 614337 Mismatch repair cancer syndrome 4, 619101
POT1	100%	100%	No OMIM disease ID

PRDX2	100%	100%	No OMIM disease ID
PRF1	100%	100%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89%	89%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	100%	100%	Fanconi anemia, complementation group O, 613390
RBBP6	100%	100%	No OMIM disease ID
RBM8A	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100%	100%	?Fanconi anemia, complementation group W, 617784
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RPA1	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6, 619767
RPL11	100%	100%	Diamond-Blackfan anemia 7, 612562
RPL15	100%	97%	?Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	?Diamond-Blackfan anemia 18, 618310
RPL26	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL31	100%	100%	No OMIM disease ID
RPL35	100%	100%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100%	100%	Diamond-Blackfan anemia 5, 612528
RPL4	100%	100%	No OMIM disease ID
RPL5	100%	100%	Diamond-Blackfan anemia 6, 612561
RPL9	100%	100%	No OMIM disease ID
RPS10	100%	100%	Diamond-Blackfan anemia 9, 613308
RPS15A	80%	80%	?Diamond-Blackfan anemia 20, 618313
RPS17	100%	100%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	100%	Diamond-Blackfan anemia 1, 105650

RPS24	100%	100%	Diamond-blackfan anemia 3, 610629
RPS26	100%	99%	Diamond-Blackfan anemia 10, 613309
RPS27	100%	100%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	100%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	100%	Diamond-Blackfan anemia 13, 615909
RPS7	100%	100%	Diamond-Blackfan anemia 8, 612563
RTEL1	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RUNX1	100%	100%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	100%	100%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100%	100%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SBDS	100%	100%	Shwachman-Diamond syndrome 1, 260400
SEC23B	100%	100%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SH2B3	100%	100%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100%	100%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC46A1	100%	100%	Folate malabsorption, hereditary, 229050
SLX4	100%	100%	Fanconi anemia, complementation group P, 613951
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRP54	100%	100%	Neutropenia, severe congenital, 8, autosomal dominant, 618752

SRP72	100%	100%	Bone marrow failure syndrome 1, 614675
STIM1	100%	100%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STN1	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	100%	100%	Barth syndrome, 302060
TBXAS1	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	100%	96%	No OMIM disease ID
TERT	100%	100%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TET2	100%	99%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
THPO	100%	100%	Thrombocythemia 1, 187950
TINF2	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR8	100%	100%	Immunodeficiency 98 with autoinflammation, X-linked, 301078
TP53	95%	95%	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%	100%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TYK2	100%	100%	Immunodeficiency 35, 611521
UBA1	100%	100%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UBE2T	100%	100%	Fanconi anemia, complementation group T, 616435
USB1	100%	100%	Poikiloderma with neutropenia, 604173
VPS13B	100%	99%	Cohen syndrome, 216550
VPS45	95%	95%	Neutropenia, severe congenital, 5, autosomal recessive, 615285

VPS4A	100%	100%	CIMDAG syndrome, 619273
WAS	100%	99%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WRAP53	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	100%	100%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
YARS2	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	100%	100%	?Pulmonary fibrosis and/or bone marrow failure, 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.
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TWIST X2 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors