HEREDITARY BONE MARROW FAILURE GENE PANEL DG 3.4.0 (173 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ABCB7	99,7%	99,5%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100,0%	100,0%	Retinal dystrophy with leukodystrophy, 618863
ACD	100,0%	100,0%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ALAS2	100,0%	100,0%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
AMN	100,0%	100,0%	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	97,2%	97,2%	Thrombocytopenia 2, 188000
AP3B1	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
ASXL1	99,9%	99,9%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ATR	100,0%	100,0%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	100,0%	100,0%	Bloom syndrome, 210900
BRAF	100,0%	100,0%	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%	Fanconi anemia, complementation group S, 617883
BRCA2	100,0%	100,0%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRIP1	100,0%	100,0%	Fanconi anemia, complementation group J, 609054
CAD	100,0%	100,0%	Developmental and epileptic encephalopathy 50, 616457

CASP10	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CBL	100,0%	100,0%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CDAN1	100,0%	100,0%	Dyserythropoietic anemia, congenital, type Ia, 224120
C15orf41	100,0%	100,0%	Dyserythropoietic anemia, congenital, type Ib, 615631
CEBPA	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CLPB	100,0%	100,0%	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%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
CSF3R	100,0%	100,0%	Neutropenia, severe congenital, 7, autosomal recessive, 617014 ?Neutrophilia, hereditary, 162830
CTC1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100,0%	100,0%	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100
CUBN	100,0%	100,0%	Imerslund-Grasbeck syndrome 1, 261100
CXCR4	100,0%	100,0%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
DBF4	100,0%	100,0%	No OMIM Disease ID
DDX41	100,0%	100,0%	No OMIM Disease ID
DHFR	100,0%	100,0%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	100,0%	100,0%	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%	Dyskeratosis congenita, X-linked, 305000
DNAJC21	100,0%	100,0%	Bone marrow failure syndrome 3, 617052
EFL1	100,0%	100,0%	Shwachman-Diamond syndrome 2, 617941
ELANE	100,0%	100,0%	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	100,0%	100,0%	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
ERCC4	100,0%	100,0%	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%	100,0%	Bone marrow failure syndrome 2, 615715
ETV6	100,0%	100,0%	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EZH2	100,0%	100,0%	Weaver syndrome, 277590
FANCA	100,0%	100,0%	Fanconi anemia, complementation group A, 227650
FANCB	100,0%	100,0%	Fanconi anemia, complementation group B, 300514
FANCC	97,3%	97,3%	Fanconi anemia, complementation group C, 227645
FANCD2	98,8%	98,8%	Fanconi anemia, complementation group D2, 227646
FANCE	100,0%	100,0%	Fanconi anemia, complementation group E, 600901
FANCF	100,0%	100,0%	Fanconi anemia, complementation group F, 603467
FANCG	100,0%	100,0%	Fanconi anemia, complementation group G, 614082
FANCI	100,0%	100,0%	Fanconi anemia, complementation group I, 609053
FANCL	100,0%	100,0%	Fanconi anemia, complementation group L, 614083
FANCM	100,0%	100,0%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100,0%	100,0%	Autoimmune lymphoproliferative syndrome, type IB, 601859
G6PC3	100,0%	100,0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	100,0%	100,0%	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
GATA2	100,0%	100,0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100,0%	100,0%	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%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107

GP1BA	100,0%	100,0%	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%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100,0%	100,0%	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100,0%	100,0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	100,0%	100,0%	Immunodeficiency, common variable, 13, 616873
IKZF5	100,0%	100,0%	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100,0%	100,0%	Isovaleric acidemia, 243500
JAGN1	100,0%	99,8%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KIF23	100,0%	100,0%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KLF1	100,0%	100,0%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
KRAS	100,0%	100,0%	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%	No OMIM Disease ID
LIG4	100,0%	100,0%	LIG4 syndrome, 606593
LPIN2	100,0%	100,0%	Majeed syndrome, 609628
MAD2L2	100,0%	100,0%	?Fanconi anemia, complementation group V, 617243
MCM4	95,5%	95,5%	Immunodeficiency 54, 609981
MDM4	100,0%	100,0%	?Bone marrow failure syndrome 6, 618849
MECOM	100,0%	100,0%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738

MLH1	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MPL	100,0%	100,0%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MSH2	100,0%	100,0%	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH6	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MVK	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYH9	100,0%	100,0%	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYSM1	96,4%	96,4%	Bone marrow failure syndrome 4, 618116
NBEAL2	100,0%	100,0%	Gray platelet syndrome, 139090
NBN	100,0%	100,0%	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NF1	100,0%	100,0%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFE2	100,0%	100,0%	No OMIM Disease ID
NHP2	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPAT	100,0%	100,0%	No OMIM Disease ID
NPM1	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626
NRAS	100,0%	100,0%	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%	Fanconi anemia, complementation group N, 610832
PARN	89,5%	87,8%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PARP4	100,0%	100,0%	No OMIM Disease ID
PAX5	100,0%	100,0%	No OMIM Disease ID
PMS2	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
POT1	100,0%	100,0%	No OMIM Disease ID
PRDX2	100,0%	100,0%	No OMIM Disease ID
PRF1	100,0%	100,0%	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PTPN11	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89,4%	89,4%	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	100,0%	100,0%	Fanconi anemia, complementation group O, 613390
RBBP6	100,0%	100,0%	No OMIM Disease ID
RBM8A	100,0%	100,0%	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100,0%	100,0%	?Fanconi anemia, complementation group W, 617784
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RPA1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 6, 619767
RPL11	100,0%	100,0%	Diamond-Blackfan anemia 7, 612562
RPL15	100,0%	100,0%	?Diamond-Blackfan anemia 12, 615550
RPL18	100,0%	100,0%	?Diamond-Blackfan anemia 18, 618310
RPL26	100,0%	100,0%	?Diamond-Blackfan anemia 11, 614900
RPL27	100,0%	100,0%	?Diamond-Blackfan anemia 16, 617408
RPL31	100,0%	100,0%	No OMIM Disease ID
RPL35	100,0%	100,0%	?Diamond-Blackfan anemia 19, 618312
RPL35A	100,0%	100,0%	Diamond-Blackfan anemia 5, 612528
RPL4	100,0%	100,0%	No OMIM Disease ID

RPL5	100,0%	100,0%	Diamond-Blackfan anemia 6, 612561
RPL9	100,0%	100,0%	No OMIM Disease ID
RPS10	100,0%	100,0%	Diamond-Blackfan anemia 9, 613308
RPS15A	80,4%	80,4%	?Diamond-Blackfan anemia 20, 618313
RPS17	100,0%	100,0%	Diamond-Blackfan anemia 4, 612527
RPS19	100,0%	100,0%	Diamond-Blackfan anemia 1, 105650
RPS24	100,0%	100,0%	Diamond-blackfan anemia 3, 610629
RPS26	100,0%	100,0%	Diamond-Blackfan anemia 10, 613309
RPS27	100,0%	100,0%	?Diamond-Blackfan anemia 17, 617409
RPS28	100,0%	100,0%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100,0%	100,0%	Diamond-Blackfan anemia 13, 615909
RPS7	100,0%	100,0%	Diamond-Blackfan anemia 8, 612563
RTEL1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RUNX1	100,0%	100,0%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100,0%	100,0%	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Spinocerebellar ataxia 49, 619806
SBDS	100,0%	100,0%	Shwachman-Diamond syndrome, 260400
SEC23B	100,0%	100,0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SH2B3	100,0%	100,0%	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	100,0%	100,0%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100,0%	100,0%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	100,0%	100,0%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100,0%	100,0%	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC46A1	100,0%	100,0%	Folate malabsorption, hereditary, 229050
SLX4	100,0%	100,0%	Fanconi anemia, complementation group P, 613951

SOS1	100,0%	100,0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRP54	100,0%	100,0%	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	100,0%	100,0%	Bone marrow failure syndrome 1, 614675
STIM1	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STN1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	100,0%	100,0%	Barth syndrome, 302060
TBXAS1	100,0%	100,0%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	100,0%	100,0%	Osteopetrosis, autosomal recessive 1, 259700
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	83,7%	83,7%	No OMIM Disease ID
TERT	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TET2	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
THPO	100,0%	100,0%	Thrombocythemia 1, 187950
TINF2	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR8	100,0%	100,0%	No OMIM Disease ID
TP53	91,7%	91,7%	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%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TYK2	100,0%	100,0%	Immunodeficiency 35, 611521
UBA1	100,0%	99,8%	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UBE2T	100,0%	100,0%	Fanconi anemia, complementation group T, 616435
USB1	100,0%	100,0%	Poikiloderma with neutropenia, 604173
VPS13B	99,5%	99,4%	Cohen syndrome, 216550
VPS45	95,3%	95,3%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
VPS4A	100,0%	100,0%	CIMDAG syndrome, 619273

WAS	100,0%	100,0%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WRAP53	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	100,0%	100,0%	Spermatogenic failure 50, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
YARS2	100,0%	100,0%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	100,0%	100,0%	?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 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors