HEREDITARY BONE MARROW FAILURE PANEL DG 2.14 (103 genes)

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ABCB7	131.5	99.9	98.4	Anemia, sideroblastic, with ataxia, 301310
ABCD4	143.6	99.9	98.3	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	145.3	97.8	96	No OMIM phenotype Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085) ?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236)
ACD	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ANKRD26	81	88.7	76.8	Thrombocytopenia 2, 188000
ATR	138.3	99.4	96.9	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
BRCA1	177.5	98.9	96.9	Fanconi anemia, complementation group S, 617883 {Breast-ovarian cancer, familial, 1}, 604370 {Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	102.7	99	97.4	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070 {Breast cancer, male, susceptibility to}, 114480 {Breast-ovarian cancer, familial, 2}, 612555 {Glioblastoma 3}, 613029 {Medulloblastoma}, 155255 {Pancreatic cancer 2}, 613347 {Prostate cancer}, 176807
BRIP1	117.8	99.8	97.7	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
CSF3R	94.4	99.3	96.5	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DHFR	48.4	91.1	72	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
DNAJC21	125.7	99.8	98.5	Bone marrow failure syndrome 3, 617052
EFL1	174.8	99.4	97.7	Shwachman-Diamond syndrome 2, 617941

ELANE	80.9	99.7	95.9	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
ERCC4	139.2	100	99.5	?XFE progeroid syndrome, 610965 Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760
ERCC6L2	107.6	99.7	97.7	Bone marrow failure syndrome 2, 615715
ETV6	140.1	100	99.9	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
FANCA	123.3	99.8	98.5	Fanconi anemia, complementation group A, 227650
FANCB	68.4	96.7	87.9	Fanconi anemia, complementation group B, 300514
FANCC	121.6	99.4	97.1	Fanconi anemia, complementation group C, 227645
FANCD2	127.6	98.7	95.5	Fanconi anemia, complementation group D2, 227646
FANCE	108	85.9	84.6	Fanconi anemia, complementation group E, 600901
FANCF	166.8	100	100	Fanconi anemia, complementation group F, 603467
FANCG	147.7	100	100	Fanconi anemia, complementation group G, 614082
FANCI	152.1	99.5	97.5	Fanconi anemia, complementation group I, 609053
FANCL	87.8	99.4	94.7	Fanconi anemia, complementation group L, 614083
FANCM	96.8	99.2	94.3	No OMIM phenotype Fanconi anemia, complementation group M, 614087
G6PC3	123.7	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	83.5	99.6	95.7	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GATA2	119.6	99.9	98.5	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286

GBA	240.3	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GFI1	83.1	99	92.9	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 ?Neutropenia, severe congenital 2, autosomal dominant, 613107
GP1BA	153	97	94.3	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	34.5	74.2	64.3	Bernard-Soulier syndrome, type B, 231200 Giant platelet disorder, isolated, 231200
GRHL2	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
HAX1	136.5	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	86.3	88	78.5	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IVD	114.9	100	100	Isovaleric acidemia, 243500
JAGN1	147.3	100	100	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KLF1	52.1	90.8	81.7	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
MAD2L2	125.9	100	100	?Fanconi anemia, complementation group V, 617243
MECOM	143.4	100	99.6	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MPL	136.7	99.6	97.5	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MYH9	130.5	99.4	98.1	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100

NBEAL2	172.7	99.5	99.3	Gray platelet syndrome, 139090
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
PALB2	152.6	100	99.7	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
POT1	90.7	99.6	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	122.5	91.2	90.8	Aplastic anemia, 609135 Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Lymphoma, non-Hodgkin, 605027
RAD51	123.2	89.4	89.4	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508 {Breast cancer, susceptibility to}, 114480
RAD51C	143.4	100	98.9	Fanconi anemia, complementation group O, 613390 {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399
RBM8A	106.6	100	99.4	Thrombocytopenia-absent radius syndrome, 274000
RPL11	99.8	100	99.5	Diamond-Blackfan anemia 7, 612562
RPL15	41.6	88.3	81.2	?Diamond-Blackfan anemia 12, 615550
RPL18	88	100	98.6	No OMIM phenotype
RPL26	46.8	94.8	83.7	?Diamond-Blackfan anemia 11, 614900
RPL27	39.3	80.4	58.3	?Diamond-Blackfan anemia 16, 617408
RPL31	84.7	99.7	95.9	No OMIM phenotype Diamond-Blackfan anemia (Farrar (2014) Am J Hematol 89, 985)
RPL35A	83.9	99.2	91	Diamond-Blackfan anemia 5, 612528
RPL5	43.8	81.8	69.3	Diamond-Blackfan anemia 6, 612561
RPL9	78	99	91.8	No OMIM phenotype Diamond-Blackfan anemia (van Dooijeweert (2018) Eur J Haematol 100, 163)
RPS10	140.1	99.8	97.1	Diamond-Blackfan anemia 9, 613308
RPS15A	80.9	99.2	92.1	No OMIM phenotype Diamond-Blackfan anemia (Ikeda (2017) Haematologica 102,e93)
RPS17	52.4	85	73.7	Diamond-Blackfan anemia 4, 612527

RPS19	82.5	99.7	95.5	Diamond-Blackfan anemia 1, 105650
RPS24	110.4	92.4	87.2	Diamond-blackfan anemia 3, 610629
RPS26	106.8	94.8	82.1	Diamond-Blackfan anemia 10, 613309
RPS27	39.7	86.7	60.3	?Diamond-Blackfan anemia 17, 617409
RPS28	47.7	100	92	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	106.2	97.9	96.6	Diamond-Blackfan anemia 13, 615909
RPS7	93.7	76.9	63.4	Diamond-Blackfan anemia 8, 612563
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX1	92	97.2	89.7	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	159.1	99.9	99.3	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	165.7	100	99.9	Ataxia-pancytopenia syndrome, 159550
SBDS	212.3	100	99.9	Shwachman-Diamond syndrome, 260400 {Aplastic anemia, susceptibility to}, 609135
SH2D1A	104.7	89.9	89.4	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	119.5	99.8	97.8	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	111.4	99.8	98.1	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	140.2	100	99.9	Glycogen storage disease Ib, 232220 Glycogen storage disease Ic, 232240
SLC46A1	106	99.4	96.4	Folate malabsorption, hereditary, 229050
SLX4	114.2	100	99.8	Fanconi anemia, complementation group P, 613951
SRP72	70	93.1	84	Bone marrow failure syndrome 1, 614675
STIM1	145.3	100	99.2	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STN1	94.4	99.9	99.5	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TBXAS1	140.3	100	100	?Thromboxane synthase deficiency, 614158 Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	113.5	95.4	89.4	Osteopetrosis, autosomal recessive 1, 259700
TERC				Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743

				{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	138.3	95.3	92	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
THPO	88.2	100	100	Thrombocythemia 1, 187950
TINF2	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TSR2	81.6	100	99.1	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
UBE2T	107	100	99.3	Fanconi anemia, complementation group T, 616435
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
VPS45	131.5	96.2	94.9	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	66.1	88.2	78.7	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	165.6	93	89.4	?Fanconi anemia, complementation group U, 617247

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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors