HEREDITARY BONE MARROW FAILURE PANEL DG 2.17 (108 genes)

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Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ABCB7	125.6	99.9%	98.8%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	139.9	99.9%	98.5%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	144.7	99.8%	98.1%	No OMIM Disease ID
ACD	180.3	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AMN	118.2	99.1%	93.0%	Megaloblastic anemia-1, Norwegian type, 261100
ANKRD26	81.5	94.9%	89.1%	Thrombocytopenia 2, 188000
ATR	142.1	99.9%	98.9%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BRCA1	164.4	99.2%	98.2%	Fanconi anemia, complementation group S, 617883
BRCA2	103.2	99.7%	98.8%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
BRIP1	122.2	99.9%	98.6%	Fanconi anemia, complementation group J, 609054
CSF3R	116.5	100.0%	99.2%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	146.7	100.0%	100.0%	Autoimmune lymphoproliferative syndrome, type V, 616100
CUBN	103.2	99.6%	98.0%	Megaloblastic anemia-1, Finnish type, 261100
DHFR	48.6	92.6%	80.9%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DNAJC21	130.6	100.0%	99.2%	Bone marrow failure syndrome 3, 617052
EFL1	152.7	99.4%	98.0%	Shwachman-Diamond syndrome 2, 617941
ELANE	156.3	100.0%	99.8%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ERCC4	136.9	100.0%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC6L2	119.8	100.0%	99.1%	Bone marrow failure syndrome 2, 615715
ETV6	157.6	99.9%	99.4%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216

FANCA	118.3	100.0%	99.2%	Fanconi anemia, complementation group A, 227650
FANCB	72.8	98.6%	93.0%	Fanconi anemia, complementation group B, 300514
FANCC	104.4	100.0%	99.3%	Fanconi anemia, complementation group C, 227645
FANCD2	116.2	99.2%	96.5%	Fanconi anemia, complementation group D2, 227646
FANCE	127.9	98.0%	91.8%	Fanconi anemia, complementation group E, 600901
FANCF	269.1	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	149.5	100.0%	100.0%	Fanconi anemia, complementation group G, 614082
FANCI	136.0	100.0%	98.8%	Fanconi anemia, complementation group I, 609053
FANCL	102.9	99.8%	97.9%	Fanconi anemia, complementation group L, 614083
FANCM	99.4	99.5%	96.6%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
G6PC3	126.1	100.0%	100.0%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	102.0	99.9%	99.0%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GATA2	128.7	100.0%	99.7%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	180.2	100.0%	100.0%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GFI1	118.9	100.0%	99.9%	?Neutropenia, severe congenital 2, autosomal dominant, 613107 ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847
GP1BA	144.7	99.1%	96.5%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	85.6	96.1%	86.6%	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
HAX1	146.3	100.0%	100.0%	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	96.7	99.8%	97.7%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IVD	106.7	100.0%	100.0%	Isovaleric acidemia, 243500

JAGN1	129.4	100.0%	100.0%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KLF1	132.7	100.0%	100.0%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593
MAD2L2	150.7	100.0%	99.9%	?Fanconi anemia, complementation group V, 617243
MECOM	133.4	100.0%	99.8%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MPL	134.4	100.0%	99.9%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MYH9	140.9	99.7%	99.0%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYSM1	110.1	99.8%	98.6%	Bone marrow failure syndrome 4, 618116
NBEAL2	182.9	100.0%	99.7%	Gray platelet syndrome, 139090
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	65.4	95.9%	83.8%	Leukemia, acute myeloid, somatic, 601626
PALB2	146.3	100.0%	99.9%	Fanconi anemia, complementation group N, 610832
PARN	127.9	100.0%	99.6%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
POT1	93.9	100.0%	98.7%	No OMIM disease ID
PRF1	154.3	91.2%	90.7%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
RAD51	104.8	89.4%	89.4%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508
RAD51C	141.9	100.0%	99.7%	Fanconi anemia, complementation group O, 613390
RBM8A	89.0	100.0%	98.2%	Thrombocytopenia-absent radius syndrome, 274000
RPL11	88.1	100.0%	99.3%	Diamond-Blackfan anemia 7, 612562
RPL15	34.3	87.5%	74.6%	?Diamond-Blackfan anemia 12, 615550
RPL18	97.9	100.0%	99.5%	?Diamond-Blackfan anemia 18, 618310
RPL26	33.4	93.3%	73.1%	?Diamond-Blackfan anemia 11, 614900
RPL27	35.3	76.1%	57.7%	?Diamond-Blackfan anemia 16, 617408
RPL31	75.6	99.3%	93.1%	No OMIM Disease ID
RPL35A	75.9	97.4%	84.8%	Diamond-Blackfan anemia 5, 612528
RPL5	36.3	86.2%	68.8%	Diamond-Blackfan anemia 6, 612561
RPL9	70.1	97.6%	86.8%	No OMIM Disease ID

RPS10	96.0	99.4%	93.2%	Diamond-Blackfan anemia 9, 613308
RPS15A	58.2	97.8%	87.3%	?Diamond-Blackfan anemia 20, 618313
RPS17	41.4	90.1%	73.3%	Diamond-Blackfan anemia 4, 612527
RPS19	81.5	100.0%	98.3%	Diamond-Blackfan anemia 1, 105650
RPS24	91.0	94.6%	89.4%	Diamond-blackfan anemia 3, 610629
RPS26	84.1	91.3%	76.0%	Diamond-Blackfan anemia 10, 613309
RPS27	34.2	91.2%	59.7%	?Diamond-Blackfan anemia 17, 617409
RPS28	59.6	100.0%	97.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100.0	98.7%	96.1%	Diamond-Blackfan anemia 13, 615909
RPS7	79.0	86.9%	69.3%	Diamond-Blackfan anemia 8, 612563
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX1	92.9	99.9%	97.4%	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	170.6	100.0%	100.0%	Ataxia-pancytopenia syndrome, 159550
SBDS	167.5	100.0%	100.0%	Shwachman-Diamond syndrome, 260400
SH2D1A	108.5	95.7%	90.7%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	103.2	100.0%	99.7%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	98.5	99.0%	95.5%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	122.0	100.0%	99.7%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC46A1	121.7	100.0%	98.0%	Folate malabsorption, hereditary, 229050
SLX4	136.8	100.0%	99.9%	Fanconi anemia, complementation group P, 613951
SRP72	69.4	95.7%	84.1%	Bone marrow failure syndrome 1, 614675
STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STN1	82.8	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TBXAS1	135.5	100.0%	100.0%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	160.1	99.9%	99.0%	No OMIM disease ID
THPO	102.6	100.0%	99.9%	Thrombocythemia 1, 187950

TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TSR2	81.3	100.0%	99.4%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
UBE2T	92.1	100.0%	99.4%	Fanconi anemia, complementation group T, 616435
USB1	122.0	99.8%	98.2%	Poikiloderma with neutropenia, 604173
VPS45	127.2	97.3%	94.1%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	75.4	95.3%	84.4%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WRAP53	178.7	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	169.7	99.8%	95.1%	?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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors