HEREDITARY BONE MARROW FAILURE GENE PANEL DG 3.2.0 (159 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ABCB7	99,5	97,1	99,5	98,7	Anemia, sideroblastic, with ataxia, 301310
ABCD4	99,8	97,7	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100	98,4	100	99,9	Retinal dystrophy with leukodystrophy, 618863
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
AMN	92,5	82,9	100	100	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	94,6	88,5	97,2	97	Thrombocytopenia 2, 188000
AP3B1	99,2	96,4	100	99,9	Hermansky-Pudlak syndrome 2, 608233
ASXL1	99,8	98,9	100	100	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ATR	99,7	99	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	99,3	97,7	100	100	Bloom syndrome, 210900
BRAF	89,4	77,6	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	99,4	98,4	100	100	Fanconi anemia, complementation group S, 617883
BRCA2	99,1	98,2	100	100	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
BRIP1	99,4	98,5	100	100	Fanconi anemia, complementation group J, 609054

CASP10	99,3	96,8	100	100	Autoimmune lymphoproliferative syndrome, type II, 603909 Gastric cancer, somatic, 613659 Lymphoma, non-Hodgkin, somatic, 605027
CBL	97,3	96,9	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CEBPA	95,9	80,1	99,7	97,3	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CLPB	94,9	94	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CSF3R	99,8	98,5	100	100	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100	99,1	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	Autoimmune lymphoproliferative syndrome, type V, 616100
CUBN	99,2	97,1	100	100	Imerslund-Grasbeck syndrome 1, 261100
CXCR4	100	100	100	100	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
DBF4	96	88,9	100	99,9	No OMIM disease ID
DDX41	100	100	100	100	No OMIM disease ID
DHFR	88,9	76,3	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	99,5	98,5	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	99,7	97,2	100	99,6	Dyskeratosis congenita, X-linked, 305000
DNAJC21	99,5	97,4	100	100	Bone marrow failure syndrome 3, 617052
EFL1	99,3	97,7	100	100	Shwachman-Diamond syndrome 2, 617941
ELANE	99,9	98,8	100	100	Neutropenia, cyclic, 162800 Neutropenia, severe congenital 1, autosomal dominant, 202700
EPO	99,9	97,8	100	100	Erythrocytosis, familial, 5, 617907 ?Diamond-Blackfan anemia-like, 617911
ERCC4	100	99,9	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	99,6	98,6	100	100	Bone marrow failure syndrome 2, 615715

ETV6	100	99,3	100	100	Thrombocytopenia 5, 616216 Leukemia, acute myeloid, somatic, 601626
EZH2	99,7	98	100	100	Weaver syndrome, 277590
FANCA	99,9	98,7	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98	91,7	100	99,6	Fanconi anemia, complementation group B, 300514
FANCC	96,9	95,7	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	98,7	95,9	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	90,7	85,5	100	100	Fanconi anemia, complementation group E, 600901
FANCF	100	100	100	100	Fanconi anemia, complementation group F, 603467
FANCG	100	99,9	100	100	Fanconi anemia, complementation group G, 614082
FANCI	99,8	98,6	100	100	Fanconi anemia, complementation group I, 609053
FANCL	99,4	97,6	100	100	Fanconi anemia, complementation group L, 614083
FANCM	98,9	96,3	100	100	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 Squamous cell carcinoma, burn scar-related, somatic,
FASLG	100	99,1	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	99,9	98,5	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
GATA2	99,8	97	100	100	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100	100	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	99,9	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107

GP1BA	97,6	94,3	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 Bernard-Soulier syndrome, type A2 (dominant), 153670 von Willebrand disease, platelet-type, 177820
GP1BB	77,8	66,9	100	99,5	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	100	99,9	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
HAVCR2	100	99,8	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	97,1	88,3	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IKZF1	99,3	99,3	100	100	Immunodeficiency, common variable, 13, 616873
IKZF5	100	100	100	100	Thrombocytopenia, autosomal dominant, 7, 619130
IVD	100	99,9	100	100	Isovaleric acidemia, 243500
JAGN1	100	100	100	99,2	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KLF1	100	99,6	100	100	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
KRAS	99	97,8	100	100	Gastric cancer, somatic, 137215 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	96,9	91,6	100	100	No OMIM disease ID
LIG4	99,8	99,3	100	100	LIG4 syndrome, 606593
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MCM4	95,3	95	95,5	95,5	Immunodeficiency 54, 609981
MDM4	100	98,8	100	100	?Bone marrow failure syndrome 6, 618849

MECOM	100	99,6	100	100	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MLH1	100	99,9	100	100	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MPL	100	99,8	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
MSH2	98,5	94,5	100	100	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSH6	100	99,3	100	100	Colorectal cancer, hereditary nonpolyposis, type 5, 614350 Mismatch repair cancer syndrome 3, 619097
MYH9	99,9	98,9	100	100	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 Deafness, autosomal dominant 17, 603622
MYSM1	96,1	95,4	96,4	96,3	Bone marrow failure syndrome 4, 618116
NBEAL2	99,5	99,3	100	100	Gray platelet syndrome, 139090
NBN	99,2	97,8	100	99,9	Leukemia, acute lymphoblastic, 613065 Aplastic anemia, 609135 Nijmegen breakage syndrome, 251260
NF1	91,8	89,3	100	100	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFE2	100	100	100	100	No OMIM disease ID
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPAT	99,7	98,8	100	100	No OMIM disease ID
NPM1	95,3	84,9	100	100	Leukemia, acute myeloid, somatic, 601626
NRAS	100	100	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	99,9	100	100	Fanconi anemia, complementation group N, 610832
PARN	81,1	80,4	88,3	87,6	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
PAX5	98,8	95,4	100	100	No OMIM disease ID
PMS2	83,9	81,6	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
POT1	99,5	98,5	100	100	No OMIM disease ID
PRF1	91,2	90,1	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89,4	89,4	89,4	89,4	Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	99,8	99,4	100	100	Fanconi anemia, complementation group O, 613390
RBBP6	97,9	95,2	100	100	No OMIM disease ID
RBM8A	99,6	95,3	100	100	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100	99,4	100	100	?Fanconi anemia, complementation group W, 617784
RMRP	0	0	0	0	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RPL11	99,9	97,9	100	100	Diamond-Blackfan anemia 7, 612562
RPL15	84,9	70,4	100	99,5	?Diamond-Blackfan anemia 12, 615550
RPL18	100	99,9	100	100	?Diamond-Blackfan anemia 18, 618310
RPL26	94,2	75,5	100	100	?Diamond-Blackfan anemia 11, 614900
RPL27	68	56,6	100	100	?Diamond-Blackfan anemia 16, 617408
RPL31	97,6	87,4	100	100	No OMIM disease ID
RPL35	90,1	79,1	100	100	?Diamond-Blackfan anemia 19, 618312

RPL35A	94,7	84,9	100	100	Diamond-Blackfan anemia 5, 612528
RPL4	83,3	73,3	100	100	No OMIM disease ID
RPL5	81,9	59,7	100	100	Diamond-Blackfan anemia 6, 612561
RPL9	98,6	88	100	100	No OMIM disease ID
RPS10	96,6	87,6	100	100	Diamond-Blackfan anemia 9, 613308
RPS15A	95,3	84,1	80,4	80,4	?Diamond-Blackfan anemia 20, 618313
RPS17	85	67,8	100	100	Diamond-Blackfan anemia 4, 612527
RPS19	100	99,9	100	100	Diamond-Blackfan anemia 1, 105650
RPS24	96,2	90,3	100	100	Diamond-blackfan anemia 3, 610629
RPS26	93,2	81,2	100	100	Diamond-Blackfan anemia 10, 613309
RPS27	95,5	70	100	100	?Diamond-Blackfan anemia 17, 617409
RPS28	99,7	86,3	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	78	70,5	100	100	Diamond-Blackfan anemia 13, 615909
RPS7	81,7	66,9	100	100	Diamond-Blackfan anemia 8, 612563
RTEL1	99,7	97,2	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	98,6	93	100	100	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	99,9	99,8	100	100	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMD9L	100	99,9	100	100	Ataxia-pancytopenia syndrome, 159550 Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270
SBDS	100	99,9	100	100	Shwachman-Diamond syndrome, 260400
SH2B3	99	94,7	100	100	Thrombocythemia, somatic, 187950 Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100
SH2D1A	97,8	92,9	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100	98,5	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	97,4	93,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950

SLC37A4	99,8	97,6	100	100	Glycogen storage disease Ib, 232220 Congenital disorder of glycosylation, type IIw, 619525 Glycogen storage disease Ic, 232240
SLC46A1	100	98,5	100	100	Folate malabsorption, hereditary, 229050
SLX4	100	99,9	100	100	Fanconi anemia, complementation group P, 613951
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRP54	98	93,4	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	98	90,1	100	100	Bone marrow failure syndrome 1, 614675
STIM1	99,9	97,5	100	100	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STN1	99,9	99,8	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	99,3	93,7	100	100	Barth syndrome, 302060
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	98,5	93,4	100	100	Osteopetrosis, autosomal recessive 1, 259700
TERC	0	0	0	0	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	100	99,9	83,7	83,7	No OMIM disease ID
TERT	97	94,8	100	100	No OMIM disease ID
TET2	100	100	100	100	Myelodysplastic syndrome, somatic, 614286 Immunodeficiency 75, 619126
THPO	81,4	78,7	100	100	Thrombocythemia 1, 187950
TINF2	100	100	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TLR8	99,9	99,8	100	100	No OMIM disease ID
TP53	99	95,2	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	99,9	98,3	100	100	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TYK2	100	99,3	100	100	Immunodeficiency 35, 611521

UBA1	99,2	97,3	99,9	99,3	Spinal muscular atrophy, X-linked 2, infantile, 301830 VEXAS syndrome, somatic, 301054
UBE2T	99,9	99,3	100	100	Fanconi anemia, complementation group T, 616435
USB1	100	98,8	100	100	Poikiloderma with neutropenia, 604173
VPS45	97,8	95,1	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	94,1	83,7	100	100	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WRAP53	100	100	100	99,9	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	99,6	95,7	100	100	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
YARS2	99,9	99,4	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	99,7	98	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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Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors