HEREDITARY BONE MARROW FAILURE GENE PANEL DG 3.1.0 (154 genes)

Releasedate: 23-03-2021

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	98,2	99,3	98,8	Anemia, sideroblastic, with ataxia, 301310
ABCD4	99,9	98,6	100	100	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100	99,2	100	100	Retinal dystrophy with leukodystrophy, 618863
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AMN	89,7	80	100	100	Imerslund-Grasbeck syndrome 2, 618882
ANKRD26	95	89,3	97,2	97,2	Thrombocytopenia 2, 188000
ASXL1	99,8	99,3	99,8	99,8	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ATR	99,9	99,4	100	100	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BLM	99,8	98,3	100	100	Bloom syndrome, 210900
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
BRCA1	99,4	98,8	100	100	Fanconi anemia, complementation group S, 617883 {Pancreatic cancer, susceptibility to, 4}, 614320 {Breast-ovarian cancer, familial, 1}, 604370
BRCA2	99,8	98,5	100	100	{Pancreatic cancer 2}, 613347 {Breast cancer, male, susceptibility to}, 114480 {Glioblastoma 3}, 613029 Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724

					{Medulloblastoma}, 155255 {Prostate cancer}, 176807 {Breast-ovarian cancer, familial, 2}, 612555
BRIP1	99,9	99	100	100	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
CASP10	99,5	97,3	100	100	Lymphoma, non-Hodgkin, somatic, 605027 Gastric cancer, somatic, 613659 Autoimmune lymphoproliferative syndrome, type II, 603909
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CEBPA	98,6	83,9	99,3	94,7	Leukemia, acute myeloid, somatic, 601626 ?Leukemia, acute myeloid, 601626
CLPB	94,9	94,9	100	100	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271
CSF3R	99,6	98,2	100	100	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100	100	100	100	{Systemic lupus erythematosus, susceptibility to}, 152700 {Diabetes mellitus, insulin-dependent, 12}, 601388 {Celiac disease, susceptibility to, 3}, 609755 Autoimmune lymphoproliferative syndrome, type V, 616100 {Hashimoto thyroiditis}, 140300
CUBN	99,7	98,3	100	100	[Proteinuria, chronic benign], 618884 Imerslund-Grasbeck syndrome 1, 261100
CXCR4	100	100	100	100	WHIM syndrome, 193670 Myelokathexis, isolated, 0
DBF4	96,6	89,6	100	100	No OMIM disease ID
DDX41	100	100	100	100	{Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871
DHFR	92,1	78,9	100	100	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DICER1	99,8	99	100	100	GLOW syndrome, somatic mosaic, 618272 Rhabdomyosarcoma, embryonal, 2, 180295 Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800 Pleuropulmonary blastoma, 601200
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
DNAJC21	99,8	98,7	100	100	Bone marrow failure syndrome 3, 617052
EFL1	99,6	98,5	100	100	Shwachman-Diamond syndrome 2, 617941
ELANE	99,7	97,4	100	100	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800

EPO	99,9	97,6	100	100	Erythrocytosis, familial, 5, 617907 {Microvascular complications of diabetes 2}, 612623 ?Diamond-Blackfan anemia-like, 617911
ERCC4	100	99,9	100	100	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC6L2	99,9	99,2	100	100	Bone marrow failure syndrome 2, 615715
ETV6	100	99,9	100	100	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
EZH2	100	99,5	100	100	Weaver syndrome, 277590
FANCA	100	99,4	100	100	Fanconi anemia, complementation group A, 227650
FANCB	98,6	94,1	100	100	Fanconi anemia, complementation group B, 300514
FANCC	97,2	96,6	97,3	97,3	Fanconi anemia, complementation group C, 227645
FANCD2	99,5	97,5	98,8	98,8	Fanconi anemia, complementation group D2, 227646
FANCE	89,8	85,1	100	99,9	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	99,9	99,2	100	100	Fanconi anemia, complementation group I, 609053
FANCL	100	98,6	100	100	Fanconi anemia, complementation group L, 614083
FANCM	99,6	97,3	100	100	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAS	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IA, 601859 {Autoimmune lymphoproliferative syndrome}, 601859 Squamous cell carcinoma, burn scar-related, somatic, 0
FASLG	100	99,6	100	100	Autoimmune lymphoproliferative syndrome, type IB, 601859 {Lung cancer, susceptibility to}, 211980
G6PC3	100	99,9	100	100	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	99,8	98,4	100	100	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	100	98,3	100	100	Emberger syndrome, 614038 {Myelodysplastic syndrome, susceptibility to}, 614286 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626

GBA	100	100	100	100	Gaucher disease, type III, 231000 {Parkinson disease, late-onset, susceptibility to}, 168600 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900 {Lewy body dementia, susceptibility to}, 127750
GFI1	100	99,2	100	100	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GP1BA	98,6	95,9	100	100	Bernard-Soulier syndrome, type A1 (recessive), 231200 {Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,9	59,6	99,5	95	Giant platelet disorder, isolated, 231200 Bernard-Soulier syndrome, type B, 231200
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
HAVCR2	100	100	100	100	T-cell lymphoma, subcutaneous panniculitis-like, 618398
HAX1	100	100	100	100	Neutropenia, severe congenital 3, autosomal recessive, 610738
HOXA11	97,1	87,5	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	100	100	100	Isovaleric acidemia, 243500
JAGN1	100	100	99,7	98	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KLF1	100	97,8	100	100	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Dyserythropoietic anemia, congenital, type IV, 613673
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215

					Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
LAPTM5	97,9	92,9	100	100	No OMIM disease ID
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
MAD2L2	100	99,9	100	100	?Fanconi anemia, complementation group V, 617243
MDM4	99,9	99	100	100	?Bone marrow failure syndrome 6, 618849
MECOM	100	99,9	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	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977
MSH2	99	96,9	100	100	Mismatch repair cancer syndrome 2, 619096 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSH6	100	99,8	100	100	{Endometrial cancer, familial}, 608089 Mismatch repair cancer syndrome 3, 619097 Colorectal cancer, hereditary nonpolyposis, type 5, 614350
MYH9	100	99,3	100	100	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYSM1	96,4	95,5	96,4	96,4	Bone marrow failure syndrome 4, 618116
NBEAL2	99,4	99,4	100	100	Gray platelet syndrome, 139090
NBN	99,9	98,6	100	100	Leukemia, acute lymphoblastic, 613065 Nijmegen breakage syndrome, 251260 Aplastic anemia, 609135
NF1	92,6	90,2	100	100	Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Watson syndrome, 193520 Neurofibromatosis, type 1, 162200
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPAT	99,8	98,7	100	100	No OMIM disease ID
NPM1	98,2	85,3	100	100	Leukemia, acute myeloid, somatic, 601626

NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
PALB2	100	100	100	100	{Pancreatic cancer, susceptibility to, 3}, 613348 Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
PAX5	98,7	96,1	100	100	{Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545
PMS2	84,3	82,8	100	100	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
POT1	99,9	99	100	100	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PRF1	91,2	90,8	100	100	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
RAD51	89,4	89,4	89,4	89,4	{Breast cancer, susceptibility to}, 114480 Mirror movements 2, 614508 Fanconi anemia, complementation group R, 617244
RAD51C	100	99,8	100	100	{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 Fanconi anemia, complementation group O, 613390
RBBP6	97,8	95,9	100	100	No OMIM disease ID
RBM8A	99,8	97,9	100	100	Thrombocytopenia-absent radius syndrome, 274000
RFWD3	100	99,8	100	100	?Fanconi anemia, complementation group W, 617784
RPL11	100	100	100	100	Diamond-Blackfan anemia 7, 612562
RPL15	86,8	78	100	100	?Diamond-Blackfan anemia 12, 615550
RPL18	100	100	100	100	?Diamond-Blackfan anemia 18, 618310
RPL26	97,2	84,4	100	100	?Diamond-Blackfan anemia 11, 614900
RPL27	73,6	56,5	100	100	?Diamond-Blackfan anemia 16, 617408

RPL31	99,3	94,6	100	100	No OMIM disease ID
RPL35	86,4	75	100	100	?Diamond-Blackfan anemia 19, 618312
RPL35A	97,1	88,7	100	100	Diamond-Blackfan anemia 5, 612528
RPL4	87,6	78,3	100	100	No OMIM disease ID
RPL5	86,2	70	100	100	Diamond-Blackfan anemia 6, 612561
RPL9	98,9	92	100	100	No OMIM disease ID
RPS10	98	92,5	100	100	Diamond-Blackfan anemia 9, 613308
RPS15A	96,9	86,7	80,5	80,4	?Diamond-Blackfan anemia 20, 618313
RPS17	84,2	69,8	100	100	Diamond-Blackfan anemia 4, 612527
RPS19	100	99,6	100	100	Diamond-Blackfan anemia 1, 105650
RPS24	98,4	93,1	100	100	Diamond-blackfan anemia 3, 610629
RPS26	95,7	84,9	100	100	Diamond-Blackfan anemia 10, 613309
RPS27	85,9	60,6	100	100	?Diamond-Blackfan anemia 17, 617409
RPS28	100	94,8	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	82	74,7	100	100	Diamond-Blackfan anemia 13, 615909
RPS7	80	68,7	100	100	Diamond-Blackfan anemia 8, 612563
RTEL1	99,5	96,8	100	100	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX1	99,3	94,9	100	100	Platelet disorder, familial, with associated myeloid malignancy, 601399 Leukemia, acute myeloid, 601626
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SAMD9L	100	100	100	100	Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270 Ataxia-pancytopenia syndrome, 159550
SBDS	100	100	100	100	{Aplastic anemia, susceptibility to}, 609135 Shwachman-Diamond syndrome, 260400
SH2B3	99,4	95,1	100	99,9	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950 Erythrocytosis, somatic, 133100
SH2D1A	97,2	94	100	100	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100	99,7	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	97,9	95,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100	99,2	100	100	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050

SLX4	100	99,8	100	100	Fanconi anemia, complementation group P, 613951
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SRP54	99,5	96,5	100	100	Neutropenia, severe congenital, 8, autosomal dominant, 618752
SRP72	97,6	89,7	100	100	Bone marrow failure syndrome 1, 614675
STIM1	99,8	98	100	100	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STN1	100	100	100	100	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341
TAZ	99,1	95,5	100	100	Barth syndrome, 302060
TBXAS1	100	100	100	100	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	97,6	90,1	100	100	Osteopetrosis, autosomal recessive 1, 259700
TERC	NC	NC	NC	NC	{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	99,9	97,8	83,7	83,7	No OMIM disease ID
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989
TET2	100	100	100	100	Immunodeficiency 75, 619126 Myelodysplastic syndrome, somatic, 614286
THPO	81,4	81	100	100	Thrombocythemia 1, 187950
TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TP53	99,9	97,7	91,7	91,7	{Adrenocortical carcinoma, pediatric}, 202300 {Glioma susceptibility 1}, 137800 {Basal cell carcinoma 7}, 614740 Bone marrow failure syndrome 5, 618165 {Colorectal cancer}, 114500 Nasopharyngeal carcinoma, somatic, 607107 Breast cancer, somatic, 114480 {Osteosarcoma}, 259500 {Choroid plexus papilloma}, 260500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, somatic, 114550 Pancreatic cancer, somatic, 260350

TSR2	100	100	100	99,9	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
TYK2	99,9	99	100	100	Immunodeficiency 35, 611521
UBA1	99,4	98,2	99,8	99	VEXAS syndrome, somatic, 301054 Spinal muscular atrophy, X-linked 2, infantile, 301830
UBE2T	100	99,9	100	100	Fanconi anemia, complementation group T, 616435
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
VPS45	99,2	95,7	95,3	95,3	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	95,9	85,3	100	99,8	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WRAP53	100	100	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988
XRCC2	99,8	97,4	100	100	Spermatogenic failure, 619145 ?Premature ovarian failure 17, 619146 ?Fanconi anemia, complementation group U, 617247
YARS2	100	99,8	100	100	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561
ZCCHC8	99,8	98,7	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.
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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors