HEREDITARY BONE MARROW FAILURE GENE PANEL
DG 2.18 (108 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ABCB7	99,50%	98,20%	99,80%	99,30%	Anemia, sideroblastic, with ataxia, 301310
ABCD4	99,90%	98,60%	100%	100%	Methylmalonic aciduria and homocystinuria, cblJ type, 614857
ACBD5	100%	99,20%	100%	100%	Retinal dystrophy with leukodystrophy, 618863
ACD	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
AMN	89,70%	80,00%	100%	100%	Megaloblastic anemia-1, Norwegian type, 261100
ANKRD26	95,00%	89,30%	97,20%	97,20%	Thrombocytopenia 2, 188000
ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
BRCA1	99,40%	98,80%	100%	100%	Fanconi anemia, complementation group S, 617883
BRCA2	99,80%	98,50%	100%	100%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
BRIP1	99,90%	99,00%	100%	100%	Fanconi anemia, complementation group J, 609054
CSF3R	99,60%	98,20%	100%	100%	Neutropenia, severe congenital, 7, autosomal recessive, 617014
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTLA4	100%	100%	100%	100%	Autoimmune lymphoproliferative syndrome, type V, 616100
CUBN	99,70%	98,30%	100%	100%	Megaloblastic anemia-1, Finnish type, 261100
DHFR	92,10%	78,90%	100%	100%	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839
DKC1	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DNAJC21	99,90%	99,00%	100%	100%	Bone marrow failure syndrome 3, 617052
EFL1	99,60%	98,50%	100%	100%	Shwachman-Diamond syndrome 2, 617941
ELANE	99,70%	97,40%	100%	100%	Neutropenia, severe congenital 1, autosomal dominant, 202700 Neutropenia, cyclic, 162800
ERCC4	100%	99,90%	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	100%	99,40%	100%	100%	Bone marrow failure syndrome 2, 615715
ETV6	100%	99,90%	100%	100%	Leukemia, acute myeloid, somatic, 601626 Thrombocytopenia 5, 616216
FANCA	100%	99,40%	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	98,60%	94,10%	100%	100%	Fanconi anemia, complementation group B, 300514
FANCC	99,90%	99,30%	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	99,50%	97,50%	98,80%	98,80%	Fanconi anemia, complementation group D2, 227646
FANCE	89,80%	85,10%	100%	99,90%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99,90%	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	99,90%	99,20%	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	98,60%	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	99,60%	97,30%	100%	100%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
G6PC3	100%	99,90%	100%	100%	Dursun syndrome, 612541 Neutropenia, severe congenital 4, autosomal recessive, 612541
GATA1	99,80%	98,40%	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%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GBA	100%	100%	100%	100%	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	100%	99,20%	100%	100%	?Neutropenia, nonimmune chronic idiopathic, of adults, 607847 Neutropenia, severe congenital 2, autosomal dominant, 613107
GP1BA	98,60%	95,90%	100%	100%	Bernard-Soulier syndrome, type A1 (recessive), 231200 von Willebrand disease, platelet-type, 177820 Bernard-Soulier syndrome, type A2 (dominant), 153670
GP1BB	72,90%	59,60%	99,50%	95,00%	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
HAX1	100%	100%	100%	100%	Neutropenia, severe congenital 3, autosomal recessive, 610738

HOXA11	97,10%	87,50%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432
IVD	100%	100%	100%	100%	Isovaleric acidemia, 243500
JAGN1	100%	100%	100%	100%	Neutropenia, severe congenital, 6, autosomal recessive, 616022
KLF1	100%	97,80%	100%	100%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
LIG4	100%	99,90%	100%	100%	LIG4 syndrome, 606593
MAD2L2	100%	99,90%	100%	100%	?Fanconi anemia, complementation group V, 617243
MECOM	100%	99,90%	100%	100%	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738
MPL	100%	99,50%	100%	100%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
MYH9	100%	99,30%	100%	100%	Deafness, autosomal dominant 17, 603622 Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100
MYSM1	100%	99,10%	100%	100%	Bone marrow failure syndrome 4, 618116
NBEAL2	99,40%	99,30%	100%	100%	Gray platelet syndrome, 139090
NHP2	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	98,20%	85,30%	100%	100%	Leukemia, acute myeloid, somatic, 601626
PALB2	100%	100%	100%	100%	Fanconi anemia, complementation group N, 610832
PARN	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
POT1	99,90%	99,00%	100%	100%	No OMIM disease ID
PRF1	91,20%	90,80%	100%	100%	Aplastic anemia, 609135 Lymphoma, non-Hodgkin, 605027 Hemophagocytic lymphohistiocytosis, familial, 2, 603553
RAD51	89,40%	89,40%	89,40%	89,40%	?Fanconi anemia, complementation group R, 617244 Mirror movements 2, 614508
RAD51C	100%	99,80%	100%	100%	Fanconi anemia, complementation group O, 613390
RBM8A	99,80%	97,90%	100%	100%	Thrombocytopenia-absent radius syndrome, 274000
RPL11	100%	100%	100%	100%	Diamond-Blackfan anemia 7, 612562
RPL15	86,80%	78,00%	100%	100%	?Diamond-Blackfan anemia 12, 615550
RPL18	100%	100%	100%	100%	?Diamond-Blackfan anemia 18, 618310
RPL26	97,20%	84,40%	100%	100%	?Diamond-Blackfan anemia 11, 614900
RPL27	73,60%	56,50%	100%	100%	?Diamond-Blackfan anemia 16, 617408
RPL31	99,30%	94,60%	100%	100%	No OMIM disease ID
RPL35A	97,10%	88,70%	100%	100%	Diamond-Blackfan anemia 5, 612528

RPL5	86,20%	70,00%	100%	100%	Diamond-Blackfan anemia 6, 612561
RPL9	98,70%	90,80%	100%	100%	No OMIM disease ID
RPS10	97,70%	91,70%	100%	100%	Diamond-Blackfan anemia 9, 613308
RPS15A	96,90%	86,70%	80,50%	80,40%	?Diamond-Blackfan anemia 20, 618313
RPS17	84,20%	69,80%	100%	100%	Diamond-Blackfan anemia 4, 612527
RPS19	100%	99,60%	100%	100%	Diamond-Blackfan anemia 1, 105650
RPS24	98,50%	93,40%	100%	100%	Diamond-blackfan anemia 3, 610629
RPS26	95,70%	84,90%	100%	100%	Diamond-Blackfan anemia 10, 613309
RPS27	89,30%	60,90%	100%	99,80%	?Diamond-Blackfan anemia 17, 617409
RPS28	100%	94,80%	100%	100%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RPS29	100%	98,20%	100%	100%	Diamond-Blackfan anemia 13, 615909
RPS7	80,00%	68,70%	100%	100%	Diamond-Blackfan anemia 8, 612563
RTEL1	99,50%	96,80%	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	99,30%	94,90%	100%	100%	Leukemia, acute myeloid, 601626 Platelet disorder, familial, with associated myeloid malignancy, 601399
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMD9L	100%	100%	100%	100%	Ataxia-pancytopenia syndrome, 159550
SBDS	100%	100%	100%	100%	Shwachman-Diamond syndrome, 260400
SH2D1A	97,20%	94,00%	100%	100%	Lymphoproliferative syndrome, X-linked, 1, 308240
SLC19A2	100%	99,70%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A38	99,70%	97,10%	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC37A4	100%	99,20%	100%	100%	Glycogen storage disease Ic, 232240 Glycogen storage disease Ib, 232220
SLC46A1	99,90%	98,50%	100%	100%	Folate malabsorption, hereditary, 229050
SLX4	100%	99,80%	100%	100%	Fanconi anemia, complementation group P, 613951
SRP72	97,60%	89,70%	100%	100%	Bone marrow failure syndrome 1, 614675
STIM1	99,80%	98,00%	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
TBXAS1	100%	100%	100%	100%	Ghosal hematodiaphyseal syndrome, 231095
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	96,20%	94,50%	100%	100%	No OMIM disease ID

THPO	100%	99,50%	100%	100%	Thrombocythemia 1, 187950
TINF2	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TSR2	100%	100%	100%	99,90%	?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946
UBE2T	100%	99,90%	100%	100%	Fanconi anemia, complementation group T, 616435
USB1	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
VPS45	99,20%	95,60%	95,10%	95,10%	Neutropenia, severe congenital, 5, autosomal recessive, 615285
WAS	95,90%	85,30%	100%	99,80%	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	99,80%	97,40%	100%	100%	?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.
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors