# HEREDITARY BONE MARROW FAILURE PANEL DG 2.16 (107 genes)

Releasedate: 07-06-2019

| *Gene* | *Median coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|
| ABCB7 | 126,2 | 99.9% | 98.6% | Anemia, sideroblastic, with ataxia, 301310 |
| ABCD4 | 129 | 99.8% | 98.4% | Methylmalonic aciduria and homocystinuria, cblJ type, 614857 |
| ACBD5 | 145,1 | 99.6% | 98.0% | No OMIM phenotype<br>Thrombocytopaenia (Punzo (2010) J Thromb Haemost 8,2085)<br>?Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) |
| ACD | 159,6 | 100.0% | 100.0% | ?Dyskeratosis congenita, autosomal dominant 6, 616553<br>?Dyskeratosis congenita, autosomal recessive 7, 616553 |
| AMN | 101,5 | 98.1% | 90.6% | Megaloblastic anemia-1, Norwegian type, 261100 |
| ANKRD26 | 83,3 | 95.3% | 90.1% | Thrombocytopenia 2, 188000 |
| ATR | 144,6 | 99.8% | 98.6% | ?Cutaneous telangiectasia and cancer syndrome, familial, 614564<br>Seckel syndrome 1, 210600 |
| BRCA1 | 161,4 | 99.1% | 98.1% | Fanconi anemia, complementation group S, 617883<br>{Breast-ovarian cancer, familial, 1}, 604370<br>{Pancreatic cancer, susceptibility to, 4}, 614320 |
| BRCA2 | 106,2 | 99.6% | 98.7% | Fanconi anemia, complementation group D1, 605724<br>Wilms tumor, 194070<br>{Breast cancer, male, susceptibility to}, 114480<br>{Breast-ovarian cancer, familial, 2}, 612555<br>{Glioblastoma 3}, 613029<br>{Medulloblastoma}, 155255<br>{Pancreatic cancer 2}, 613347<br>{Prostate cancer}, 176807 |
| BRIP1 | 125,8 | 99.7% | 98.8% | Fanconi anemia, complementation group J, 609054<br>{Breast cancer, early-onset, susceptibility to}, 114480 |
| CSF3R | 105,4 | 99.7% | 98.6% | Neutropenia, severe congenital, 7, autosomal recessive, 617014 |
| CTC1 | 105,5 | 100.0% | 99.3% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTLA4 | 141 | 100.0% | 100.0% | Autoimmune lymphoproliferative syndrome, type V, 616100<br>{Celiac disease, susceptibility to, 3}, 609755<br>{Diabetes mellitus, insulin-dependent, 12}, 601388<br>{Hashimoto thyroiditis}, 140300<br>{Systemic lupus erythematosus, susceptibility to}, 152700 |

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| CUBN | 103,2 | 99.6% | 97.6% | Megaloblastic anemia-1, Finnish type, 261100 |
| DHFR | 50 | 94.1% | 83.1% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839 |
| DKC1 | 91,2 | 99.8% | 97.7% | Dyskeratosis congenita, X-linked, 305000 |
| DNAJC21 | 128,1 | 99.9% | 99.5% | Bone marrow failure syndrome 3, 617052 |
| EFL1 | 150,5 | 99.5% | 98.1% | Shwachman-Diamond syndrome 2, 617941 |
| ELANE | 141,5 | 100.0% | 99.3% | Neutropenia, cyclic, 162800<br>Neutropenia, severe congenital 1, autosomal dominant, 202700 |
| ERCC4 | 132 | 100.0% | 99.8% | Fanconi anemia, complementation group Q, 615272<br>Xeroderma pigmentosum, group F, 278760<br>Xeroderma pigmentosum, type F/Cockayne syndrome, 278760<br>XFE progeroid syndrome, 610965 |
| ERCC6L2 | 121,7 | 99.9% | 99.0% | Bone marrow failure syndrome 2, 615715 |
| ETV6 | 148,3 | 100.0% | 99.4% | Leukemia, acute myeloid, somatic, 601626<br>Thrombocytopenia 5, 616216 |
| FANCA | 112,4 | 99.9% | 98.9% | Fanconi anemia, complementation group A, 227650 |
| FANCB | 76,4 | 98.6% | 93.2% | Fanconi anemia, complementation group B, 300514 |
| FANCC | 100,8 | 99.7% | 99.2% | Fanconi anemia, complementation group C, 227645 |
| FANCD2 | 115,6 | 99.1% | 96.6% | Fanconi anemia, complementation group D2, 227646 |
| FANCE | 118,2 | 96.6% | 89.9% | Fanconi anemia, complementation group E, 600901 |
| FANCF | 244,4 | 100.0% | 100.0% | Fanconi anemia, complementation group F, 603467 |
| FANCG | 140,7 | 100.0% | 99.8% | Fanconi anemia, complementation group G, 614082 |
| FANCI | 136,2 | 99.9% | 98.9% | Fanconi anemia, complementation group I, 609053 |
| FANCL | 105,8 | 99.7% | 98.0% | Fanconi anemia, complementation group L, 614083 |
| FANCM | 100,6 | 99.3% | 97.1% | ?Premature ovarian failure 15, 618096<br>Spermatogenic failure 28, 618086 |
| G6PC3 | 114,6 | 100.0% | 100.0% | Dursun syndrome, 612541<br>Neutropenia, severe congenital 4, autosomal recessive, 612541 |
| GATA1 | 92,9 | 99.9% | 98.2% | Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835<br>Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685<br>Thrombocytopenia with beta-thalassemia, X-linked, 314050<br>Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 |
| GATA2 | 115 | 100.0% | 99.0% | Emberger syndrome, 614038<br>Immunodeficiency 21, 614172<br>{Leukemia, acute myeloid, susceptibility to}, 601626<br>{Myelodysplastic syndrome, susceptibility to}, 614286 |
| GBA | 169,8 | 100.0% | 100.0% | Gaucher disease, perinatal lethal, 608013<br>Gaucher disease, type I, 230800 |

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| | | | | Gaucher disease, type II, 230900<br>Gaucher disease, type III, 231000<br>Gaucher disease, type IIIC, 231005<br>{Lewy body dementia, susceptibility to}, 127750<br>{Parkinson disease, late-onset, susceptibility to}, 168600 |
| GFI1 | 105,7 | 100.0% | 100.0% | ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847<br>?Neutropenia, severe congenital 2, autosomal dominant, 613107 |
| GP1BA | 136,8 | 98.7% | 95.7% | Bernard-Soulier syndrome, type A1 (recessive), 231200<br>Bernard-Soulier syndrome, type A2 (dominant), 153670<br>von Willebrand disease, platelet-type, 177820<br>{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 |
| GP1BB | 68,6 | 94.5% | 83.1% | Bernard-Soulier syndrome, type B, 231200<br>Giant platelet disorder, isolated, 231200 |
| GRHL2 | 116,8 | 100.0% | 100.0% | Corneal dystrophy, posterior polymorphous, 4, 618031<br>Deafness, autosomal dominant 28, 608641<br>Ectodermal dysplasia/short stature syndrome, 616029 |
| HAX1 | 137,4 | 100.0% | 100.0% | Neutropenia, severe congenital 3, autosomal recessive, 610738 |
| HOXA11 | 88,3 | 100.0% | 98.0% | Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432 |
| IVD | 100 | 100.0% | 99.9% | Isovaleric acidemia, 243500 |
| JAGN1 | 118,5 | 100.0% | 100.0% | Neutropenia, severe congenital, 6, autosomal recessive, 616022 |
| KLF1 | 115,3 | 100.0% | 99.9% | Blood group--Lutheran inhibitor, 111150<br>Dyserythropoietic anemia, congenital, type IV, 613673<br>[Hereditary persistence of fetal hemoglobin], 613566 |
| LIG4 | 173,4 | 100.0% | 99.8% | LIG4 syndrome, 606593<br>{Multiple myeloma, resistance to}, 254500 |
| MAD2L2 | 139,1 | 100.0% | 99.8% | ?Fanconi anemia, complementation group V, 617243 |
| MECOM | 131,2 | 100.0% | 99.6% | Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738 |
| MPL | 125,8 | 100.0% | 99.8% | Myelofibrosis with myeloid metaplasia, somatic, 254450<br>Thrombocythemia 2, 601977<br>Thrombocytopenia, congenital amegakaryocytic, 604498 |
| MYH9 | 128,5 | 99.6% | 98.5% | Deafness, autosomal dominant 17, 603622<br>Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 |
| MYSM1 | 111 | 99.8% | 98.4% | Bone marrow failure syndrome 4, 618116 |
| NBEAL2 | 166 | 100.0% | 99.5% | Gray platelet syndrome, 139090 |
| NHP2 | 121,9 | 100.0% | 99.2% | Dyskeratosis congenita, autosomal recessive 2, 613987 |
| NOP10 | 120,5 | 100.0% | 100.0% | Dyskeratosis congenita, autosomal recessive 1, 224230 |
| PALB2 | 143,5 | 100.0% | 99.9% | Fanconi anemia, complementation group N, 610832 |

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| | | | | {Breast cancer, susceptibility to}, 114480<br>{Pancreatic cancer, susceptibility to, 3}, 613348 |
| PARN | 127,3 | 99.9% | 99.5% | Dyskeratosis congenita, autosomal recessive 6, 616353<br>Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 |
| POT1 | 97,7 | 99.9% | 98.5% | {Glioma susceptibility 9}, 616568<br>{Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 |
| PRF1 | 138,1 | 91.2% | 90.6% | Aplastic anemia, 609135<br>Hemophagocytic lymphohistiocytosis, familial, 2, 603553<br>Lymphoma, non-Hodgkin, 605027 |
| RAD51 | 100,6 | 89.4% | 89.4% | ?Fanconi anemia, complementation group R, 617244<br>Mirror movements 2, 614508<br>{Breast cancer, susceptibility to}, 114480 |
| RAD51C | 140,6 | 99.9% | 99.5% | Fanconi anemia, complementation group O, 613390<br>{Breast-ovarian cancer, familial, susceptibility to, 3}, 613399 |
| RBM8A | 87,4 | 99.8% | 97.4% | Thrombocytopenia-absent radius syndrome, 274000 |
| RPL11 | 85,4 | 100.0% | 99.3% | Diamond-Blackfan anemia 7, 612562 |
| RPL15 | 32 | 87.2% | 72.1% | ?Diamond-Blackfan anemia 12, 615550 |
| RPL18 | 89,3 | 100.0% | 99.5% | ?Diamond-Blackfan anemia 18, 618310 |
| RPL26 | 31 | 91.7% | 68.9% | ?Diamond-Blackfan anemia 11, 614900 |
| RPL27 | 32,5 | 72.7% | 54.6% | ?Diamond-Blackfan anemia 16, 617408 |
| RPL31 | 72,9 | 98.6% | 93.7% | No OMIM phenotype |
| RPL35A | 75,4 | 96.4% | 84.6% | Diamond-Blackfan anemia 5, 612528 |
| RPL5 | 34,7 | 85.0% | 67.7% | Diamond-Blackfan anemia 6, 612561 |
| RPL9 | 67,9 | 98.4% | 86.4% | No OMIM phenotype |
| RPS10 | 91,8 | 98.8% | 91.8% | Diamond-Blackfan anemia 9, 613308 |
| RPS15A | 58,3 | 97.1% | 86.3% | ?Diamond-Blackfan anemia 20, 618313 |
| RPS17 | 38,2 | 87.0% | 68.9% | Diamond-Blackfan anemia 4, 612527 |
| RPS19 | 76,7 | 99.9% | 96.6% | Diamond-Blackfan anemia 1, 105650 |
| RPS24 | 84,5 | 95.2% | 89.7% | Diamond-blackfan anemia 3, 610629 |
| RPS26 | 75,9 | 89.2% | 75.8% | Diamond-Blackfan anemia 10, 613309 |
| RPS27 | 34,4 | 89.5% | 57.5% | ?Diamond-Blackfan anemia 17, 617409 |
| RPS28 | 54,1 | 99.7% | 95.0% | Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 |
| RPS29 | 90,8 | 98.7% | 94.6% | Diamond-Blackfan anemia 13, 615909 |
| RPS7 | 76,6 | 84.8% | 70.0% | Diamond-Blackfan anemia 8, 612563 |
| RTEL1 | 131,1 | 99.7% | 97.7% | Dyskeratosis congenita, autosomal dominant 4, 615190<br>Dyskeratosis congenita, autosomal recessive 5, 615190<br>Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 |

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| RUNX1 | 84,6 | 99.6% | 96.3% | Leukemia, acute myeloid, 601626<br>Platelet disorder, familial, with associated myeloid malignancy, 601399 |
| SAMD9 | 163,9 | 100.0% | 99.9% | MIRAGE syndrome, 617053<br>Tumoral calcinosis, familial, normophosphatemic, 610455 |
| SAMD9L | 171,8 | 100.0% | 100.0% | Ataxia-pancytopenia syndrome, 159550 |
| SBDS | 166,2 | 100.0% | 100.0% | Shwachman-Diamond syndrome, 260400<br>{Aplastic anemia, susceptibility to}, 609135 |
| SH2D1A | 108,9 | 97.8% | 92.4% | Lymphoproliferative syndrome, X-linked, 1, 308240 |
| SLC19A2 | 101,3 | 100.0% | 99.6% | Thiamine-responsive megaloblastic anemia syndrome, 249270 |
| SLC25A38 | 94,5 | 99.1% | 95.2% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 |
| SLC37A4 | 114,3 | 100.0% | 99.6% | Glycogen storage disease Ib, 232220<br>Glycogen storage disease Ic, 232240 |
| SLC46A1 | 111,1 | 99.9% | 98.4% | Folate malabsorption, hereditary, 229050 |
| SLX4 | 124,2 | 100.0% | 99.7% | Fanconi anemia, complementation group P, 613951 |
| SRP72 | 69,2 | 95.7% | 85.6% | Bone marrow failure syndrome 1, 614675 |
| STIM1 | 120,7 | 99.8% | 96.8% | Immunodeficiency 10, 612783<br>Myopathy, tubular aggregate, 1, 160565<br>Stormorken syndrome, 185070 |
| STN1 | 82,2 | 100.0% | 99.6% | Cerebroretinal microangiopathy with calcifications and cysts 2, 617341 |
| TBXAS1 | 128,8 | 100.0% | 100.0% | ?Thromboxane synthase deficiency, 614158<br>Ghosal hematodiaphyseal syndrome, 231095 |
| TCIRG1 | 131,4 | 99.2% | 96.6% | Osteopetrosis, autosomal recessive 1, 259700 |
| TERC | NC | NC | NC | Dyskeratosis congenita, autosomal dominant 1, 127550<br>{Aplastic anemia}, 614743<br>{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 |
| TERT | 144,1 | 99.7% | 97.6% | {Dyskeratosis congenita, autosomal dominant 2}, 613989<br>{Dyskeratosis congenita, autosomal recessive 4}, 613989<br>{Leukemia, acute myeloid}, 601626<br>{Melanoma, cutaneous malignant, 9}, 615134<br>{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 |
| THPO | 97,3 | 100.0% | 99.7% | Thrombocythemia 1, 187950 |
| TINF2 | 177,1 | 100.0% | 100.0% | Dyskeratosis congenita, autosomal dominant 3, 613990<br>Revesz syndrome, 268130 |
| TSR2 | 76,1 | 100.0% | 99.4% | ?Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946 |
| UBE2T | 91,1 | 100.0% | 99.3% | Fanconi anemia, complementation group T, 616435 |
| USB1 | 118,2 | 99.8% | 97.2% | Poikiloderma with neutropenia, 604173 |
| VPS45 | 126,5 | 97.3% | 94.4% | Neutropenia, severe congenital, 5, autosomal recessive, 615285 |

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| WAS | 70,4 | 94.2% | 83.6% | Neutropenia, severe congenital, X-linked, 300299<br>Thrombocytopenia, X-linked, 313900<br>Thrombocytopenia, X-linked, intermittent, 313900<br>Wiskott-Aldrich syndrome, 301000 |
| WRAP53 | 162,8 | 100.0% | 100.0% | Dyskeratosis congenita, autosomal recessive 3, 613988 |
| XRCC2 | 171,8 | 99.8% | 96.5% | ?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 : May 8th, 2019.*
*This list is accurate for panel version DG 2.16*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*