

# INHERITED BONE MARROW FAILURE AND/OR PREDISPOSITION PANEL TO HEMATOLOGICAL MALIGNANCIES DG-4.2.0 (202 GENES)

| <i>Gene</i> | <i>Twist X2 covered 10x</i> | <i>Twist X2 covered 20x</i> | <i>srWGS covered 10x</i> | <i>srWGS covered 15x</i> | <i>srWGS covered 20x</i> | <i>Associated Phenotype description and OMIM disease ID</i>                                                  |
|-------------|-----------------------------|-----------------------------|--------------------------|--------------------------|--------------------------|--------------------------------------------------------------------------------------------------------------|
| ABCB7       | 100%                        | 99.9%                       | 99%                      | 89.5%                    | 69.4%                    | Anemia, sideroblastic, with ataxia, 301310                                                                   |
| ABCD4       | 100%                        | 100%                        | 100%                     | 100%                     | 98.9%                    | Methylmalonic aciduria and homocystinuria, cblJ type, 614857                                                 |
| ACBD5       | 85.6%                       | 85.6%                       | 100%                     | 100%                     | 99%                      | Retinal dystrophy with leukodystrophy, 618863                                                                |
| ACD         | 100%                        | 100%                        | 100%                     | 100%                     | 99.4%                    | ?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553 |
| ALAS2       | 100%                        | 99.1%                       | 98.6%                    | 87.5%                    | 66.6%                    | Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752                            |

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| AMN     | 100%  | 100%  | 100%  | 100%  | 98.9% | Imerslund-Grasbeck syndrome 2, 618882                                                                                                                                                                                                                  |
| ANKRD26 | 100%  | 100%  | 100%  | 100%  | 99.5% | Thrombocytopenia 2, 188000                                                                                                                                                                                                                             |
| AP3B1   | 100%  | 100%  | 100%  | 100%  | 99.5% | Hermansky-Pudlak syndrome 2, 608233                                                                                                                                                                                                                    |
| ASXL1   | 100%  | 100%  | 100%  | 99.9% | 99.2% | Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039                                                                                                                                                                               |
| ATR     | 100%  | 100%  | 99.7% | 99.2% | 98.3% | Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564                                                                                                                                                              |
| BLM     | 96.8% | 96.6% | 100%  | 100%  | 99.5% | Bloom syndrome, 210900                                                                                                                                                                                                                                 |
| BRAF    | 100%  | 100%  | 99.9% | 99.1% | 97.1% | Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Non-small cell lung cancer, somatic, 211980 |

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|-------|------|------|------|------|-------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| BRCA1 | 100% | 100% | 100% | 100% | 99.3% | Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320                                                                                                                                |
| BRCA2 | 100% | 100% | 100% | 100% | 99.6% | Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070 |
| BRIP1 | 96%  | 96%  | 100% | 100% | 99.5% | Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480                                                                                                                                                                       |
| CA2   | 100% | 100% | 100% | 100% | 99.6% | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730                                                                                                                                                                                                     |

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| CAD    | 100% | 100%  | 100%  | 99.9% | 98.8% | Developmental and epileptic encephalopathy 50, 616457                                                                            |
| CALR   | 100% | 100%  | 100%  | 100%  | 98.9% | Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950                                                                  |
| CASP10 | 100% | 99.7% | 100%  | 100%  | 99.5% | Autoimmune lymphoproliferative syndrome, type II, 603909;Gastric cancer, somatic, 613659;Lymphoma, non-Hodgkin, somatic, 605027  |
| CBL    | 100% | 100%  | 100%  | 99.9% | 98.9% | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785 |
| CDAN1  | 100% | 100%  | 100%  | 99.9% | 98.3% | Dyserythropoietic anemia, congenital, type Ia, 224120                                                                            |
| CDIN1  | 100% | 100%  | 100%  | 100%  | 99.5% | Dyserythropoietic anemia, congenital, type Ib, 615631                                                                            |
| CEBPA  | 100% | 99.7% | 99.7% | 97.9% | 91.4% | Leukemia, acute myeloid, somatic, 601626;?Leukemia, acute myeloid, 601626                                                        |

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| CLCN7  | 100% | 100% | 100% | 99.9% | 98.9% | Hypopigmentation, organomegaly, and delayed myelination and development, 618541;Osteopetrosis, autosomal recessive 4, 611490;Osteopetrosis, autosomal dominant 2, 166600                             |
| CLPB   | 100% | 100% | 100% | 99.7% | 98.5% | Neutropenia, severe congenital, 9, autosomal dominant, 619813;3-methylglutaconic aciduria, type VIIB, autosomal recessive, 616271;3-methylglutaconic aciduria, type VIIA, autosomal dominant, 619835 |
| COPZ1  | 100% | 100% | 100% | 100%  | 99.6% |                                                                                                                                                                                                      |
| COX4I2 | 100% | 100% | 100% | 100%  | 99%   | Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714                                                                                                      |
| CSF3R  | 100% | 100% | 100% | 100%  | 99%   | Neutropenia, severe congenital, 7, autosomal recessive, 617014;?Neutrophilia, hereditary, 162830                                                                                                     |

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| CTC1  | 100%  | 100%  | 100% | 99.9% | 98.8% | Cerebroretinal microangiopathy with calcifications and cysts, 612199                                                                                                                                                                                                                  |
| CTLA4 | 93.2% | 93.2% | 100% | 100%  | 99.3% | Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100;{Diabetes mellitus, insulin-dependent, 12}, 601388;{Celiac disease, susceptibility to, 3}, 609755;{Hashimoto thyroiditis}, 140300;{Systemic lupus erythematosus, susceptibility to}, 152700 |
| CUBN  | 100%  | 100%  | 100% | 99.9% | 99.2% | [Proteinuria, chronic benign], 618884;Imerslund-Grasbeck syndrome 1, 261100                                                                                                                                                                                                           |
| CXCR2 | 100%  | 100%  | 100% | 99.6% | 97.8% | ?WHIM syndrome 2, 619407                                                                                                                                                                                                                                                              |
| CXCR4 | 99%   | 99%   | 100% | 100%  | 99.3% | WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670                                                                                                                                                                                                                               |
| CYCS  | 100%  | 100%  | 100% | 100%  | 100%  | Thrombocytopenia 4, 612004                                                                                                                                                                                                                                                            |
| DBF4  | 100%  | 100%  | 100% | 99.9% | 98.5% |                                                                                                                                                                                                                                                                                       |

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| DCLRE1B | 100% | 100%  | 100%  | 100%  | 99%   | Dyskeratosis congenita, autosomal recessive 8, 620133                                                                                                                                    |
| DDX41   | 100% | 100%  | 100%  | 100%  | 98.8% | {Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to}, 616871                                                                                 |
| DHFR    | 100% | 100%  | 100%  | 99.9% | 99.1% | Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839                                                                                                                   |
| DICER1  | 100% | 100%  | 100%  | 100%  | 99.3% | Pleuropulmonary blastoma, 601200;Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800;GLOW syndrome, somatic mosaic, 618272;Rhabdomyosarcoma, embryonal, 2, 180295 |
| DIS3    | 100% | 100%  | 100%  | 100%  | 99.2% |                                                                                                                                                                                          |
| DKC1    | 100% | 99.7% | 98.6% | 87.6% | 67.8% | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000                                                                 |

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| DNAJC21 | 100%  | 100%  | 100% | 99.8% | 99%   | Bone marrow failure syndrome 3, 617052                                                                                                                                        |
| DUT     | 100%  | 100%  | 100% | 99.8% | 99.2% | Bone marrow failure and diabetes mellitus syndrome, 620044                                                                                                                    |
| EFL1    | 99.2% | 99.2% | 100% | 100%  | 99.6% | Shwachman-Diamond syndrome 2, 617941                                                                                                                                          |
| ELANE   | 100%  | 100%  | 100% | 100%  | 99.2% | Neutropenia, cyclic, 162800;Neutropenia, severe congenital 1, autosomal dominant, 202700                                                                                      |
| EPO     | 100%  | 100%  | 100% | 99.9% | 98.4% | {Microvascular complications of diabetes 2}, 612623;Erythrocytosis, familial, 5, 617907;?Diamond-Blackfan anemia-like, 617911                                                 |
| ERCC4   | 100%  | 100%  | 100% | 99.9% | 99.1% | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272 |
| ERCC6L2 | 100%  | 100%  | 100% | 100%  | 99.7% | Bone marrow failure syndrome 2, 615715                                                                                                                                        |

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| ERG     | 100%  | 100%  | 100%  | 100%  | 99.4% | Lymphatic malformation 14, 620602                                   |
| ETV6    | 100%  | 100%  | 100%  | 100%  | 99%   | Thrombocytopenia 5, 616216;Leukemia, acute myeloid, somatic, 601626 |
| EZH2    | 100%  | 100%  | 100%  | 100%  | 99.5% | Weaver syndrome, 277590                                             |
| FAAP100 | 100%  | 100%  | 100%  | 100%  | 99.3% |                                                                     |
| FANCA   | 100%  | 100%  | 100%  | 100%  | 99%   | Fanconi anemia, complementation group A, 227650                     |
| FANCB   | 96.2% | 96.1% | 98.7% | 88.6% | 69.8% | Fanconi anemia, complementation group B, 300514                     |
| FANCC   | 100%  | 100%  | 100%  | 99.9% | 99.3% | Fanconi anemia, complementation group C, 227645                     |
| FANCD2  | 100%  | 100%  | 100%  | 100%  | 99.3% | Fanconi anemia, complementation group D2, 227646                    |
| FANCE   | 100%  | 100%  | 100%  | 100%  | 98.9% | Fanconi anemia, complementation group E, 600901                     |
| FANCF   | 100%  | 100%  | 100%  | 100%  | 99.6% | Fanconi anemia, complementation group F, 603467                     |
| FANCG   | 100%  | 100%  | 100%  | 99.9% | 98.9% | Fanconi anemia, complementation group G, 614082                     |

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| FANCI | 100%  | 100%  | 100%  | 100%  | 99.5% | Fanconi anemia, complementation group I, 609053                                                                                                                |
| FANCL | 91.1% | 88.6% | 99.8% | 98.8% | 96.6% | Fanconi anemia, complementation group L, 614083                                                                                                                |
| FANCM | 100%  | 100%  | 100%  | 100%  | 99.6% | Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086                                                                                          |
| FAS   | 100%  | 99.9% | 100%  | 99.9% | 99.5% | Squamous cell carcinoma, burn scar-related, somatic;Autoimmune lymphoproliferative syndrome, type IA, 601859;{Autoimmune lymphoproliferative syndrome}, 601859 |
| FASLG | 100%  | 100%  | 100%  | 99.9% | 99.2% | Autoimmune lymphoproliferative syndrome, type IB, 601859;{Lung cancer, susceptibility to}, 211980                                                              |
| G6PC3 | 96.7% | 96.7% | 100%  | 100%  | 99.3% | Dursun syndrome, 612541;Neutropenia, severe congenital 4, autosomal recessive, 612541                                                                          |

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| GALE  | 100%  | 100%  | 100% | 100%  | 99.4% | Thrombocytopenia 13, syndromic, 620776;Galactose epimerase deficiency, 230350                                                                                                                                                                                                                                                                             |
| GATA1 | 100%  | 99.8% | 97%  | 86.4% | 64.8% | Anemia, congenital, nonspherocytic hemolytic, 9, 301083;Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 159595;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 | 85.7% | 85.7% | 100% | 100%  | 99.2% | {Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286                                                                                                                                                                                          |

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| GBA1  | 100% | 100% | 100% | 100%  | 99.3% | {Lewy body dementia, susceptibility to}, 127750;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;{Parkinson disease, late-onset, susceptibility to}, 168600 |
| GFI1  | 100% | 100% | 100% | 100%  | 98.8% | ?Neutropenia, nonimmune chronic idiopathic, of adults, 607847;Neutropenia, severe congenital 2, autosomal dominant, 613107                                                                                                                                                                 |
| GINS4 | 100% | 100% | 100% | 99.9% | 99.1% |                                                                                                                                                                                                                                                                                            |

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| GP1BA  | 100% | 100% | 100% | 99.3% | 96.3% | Bernard-Soulier syndrome, type A1 (recessive), 231200;Bernard-Soulier syndrome, type A2 (dominant), 153670;von Willebrand disease, platelet-type, 177820;{Nonarteritic anterior ischemic optic neuropathy, susceptibility to}, 258660 |
| GP1BB  | 100% | 100% | 100% | 100%  | 98.8% | Giant platelet disorder, isolated, 231200;Bernard-Soulier syndrome, type B, 231200                                                                                                                                                    |
| GRHL2  | 100% | 100% | 100% | 100%  | 99.1% | Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031                                                                                      |
| HAVCR2 | 100% | 100% | 100% | 100%  | 99.5% | T-cell lymphoma, subcutaneous panniculitis-like, 618398                                                                                                                                                                               |
| HAX1   | 100% | 100% | 100% | 99.8% | 98.7% | Neutropenia, severe congenital 3, autosomal recessive, 610738                                                                                                                                                                         |

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| HEATR3 | 100% | 100% | 100% | 100%  | 99.6% | Diamond-Blackfan anemia 21, 620072                                                                                                                                                                           |
| HOXA11 | 100% | 100% | 100% | 100%  | 98.2% | Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432                                                                                                                                        |
| IKZF1  | 100% | 100% | 100% | 100%  | 98.7% | Immunodeficiency, common variable, 13, 616873                                                                                                                                                                |
| IKZF2  | 100% | 100% | 100% | 99.9% | 99.5% |                                                                                                                                                                                                              |
| IKZF5  | 100% | 100% | 100% | 100%  | 99.1% | Thrombocytopenia, autosomal dominant, 7, 619130                                                                                                                                                              |
| IVD    | 100% | 100% | 100% | 100%  | 99.5% | Isovaleric acidemia, 243500                                                                                                                                                                                  |
| JAGN1  | 100% | 100% | 100% | 99.8% | 98.6% | Neutropenia, severe congenital, 6, autosomal recessive, 616022                                                                                                                                               |
| JAK2   | 100% | 100% | 100% | 99.9% | 99.2% | {Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300 |

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| KDM1A | 96.9% | 96.9% | 100% | 99.9% | 99.2% | Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990                                                                           |
| KIF23 | 100%  | 100%  | 100% | 100%  | 99.4% | Anemia, congenital dyserythropoietic, type IIIA, 105600                                                                                                                                                                |
| KIT   | 100%  | 100%  | 100% | 100%  | 99.5% | Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626 |

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| KLF1 | 100% | 100% | 100% | 100% | 99.1% | Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969 |
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| KRAS   | 100% | 100%  | 100% | 100% | 99.6% | Gastric cancer, somatic, 613659;Oculoectoderm al 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;Schimmelpenni ng-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocuta neous syndrome 2, 615278;Bladder cancer, somatic, 109800 |
| LAPTM5 | 100% | 99.8% | 100% | 100% | 99.2% |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| LCP1   | 100% | 100%  | 100% | 100% | 99.5% |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |

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| LIG4   | 100%  | 100%  | 100% | 100%  | 99.8% | LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500                                |
| LPIN2  | 99.4% | 99.2% | 100% | 99.9% | 99.3% | Majeed syndrome, 609628                                                                        |
| MAD2L2 | 100%  | 100%  | 100% | 100%  | 99.2% | ?Fanconi anemia, complementation group V, 617243                                               |
| MBD4   | 100%  | 100%  | 100% | 100%  | 99.2% | {Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975         |
| MCM4   | 95.3% | 95.3% | 100% | 100%  | 99.3% | Immunodeficiency 54, 609981                                                                    |
| MDM4   | 100%  | 100%  | 100% | 100%  | 99.1% | ?Bone marrow failure syndrome 6, 618849                                                        |
| MECOM  | 100%  | 100%  | 100% | 100%  | 99.4% | Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738                          |
| MLH1   | 100%  | 100%  | 100% | 99.9% | 99.4% | Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300 |

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| MPL  | 100%  | 100%  | 100% | 100%  | 99.1% | Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977                 |
| MSH2 | 100%  | 100%  | 100% | 100%  | 99.4% | Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096                                                           |
| MSH6 | 100%  | 100%  | 100% | 99.8% | 98.6% | Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089                                                |
| MVK  | 100%  | 100%  | 100% | 100%  | 99.2% | Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377                                                            |
| MYH9 | 97.2% | 97.2% | 100% | 100%  | 99%   | Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100;Deafness, autosomal dominant 17, 603622 |

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| MYSM1  | 100%  | 100%  | 100% | 100%  | 99.6% | Bone marrow failure syndrome 4, 618116                                                                                                                                                   |
| NAF1   | 100%  | 100%  | 100% | 99.9% | 98.6% | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365                                                                                                      |
| NBEAL2 | 100%  | 100%  | 100% | 99.9% | 99.3% | Gray platelet syndrome, 139090                                                                                                                                                           |
| NBN    | 97.5% | 97.5% | 100% | 100%  | 99.7% | Leukemia, acute lymphoblastic, 613065;Aplastic anemia, 609135;Nijmegen breakage syndrome, 251260                                                                                         |
| NF1    | 99.4% | 99.4% | 100% | 99.9% | 99.3% | Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321 |
| NFE2   | 100%  | 100%  | 100% | 99.9% | 98.8% |                                                                                                                                                                                          |
| NHP2   | 100%  | 100%  | 100% | 100%  | 98.9% | Dyskeratosis congenita, autosomal recessive 2, 613987                                                                                                                                    |

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| NOP10 | 92.5% | 92.5% | 100% | 100%  | 99.3% | ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230 |
| NPAT  | 100%  | 100%  | 100% | 100%  | 99.7% |                                                                                                                                                                                                                             |
| NPM1  | 87.6% | 87.6% | 100% | 99.9% | 98.8% | Leukemia, acute myeloid, somatic, 601626                                                                                                                                                                                    |

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| NRAS  | 100%  | 100%  | 100% | 99.9% | 99.2% | Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpennin g-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500 |
| OSTM1 | 99.9% | 98.9% | 100% | 100%  | 99.7% | Osteopetrosis, autosomal recessive 5, 259720                                                                                                                                                                                                                                                                                                                                                      |
| PALB2 | 100%  | 100%  | 100% | 100%  | 99.4% | {Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832                                                                                                                                                                                                                                 |

|         |       |       |      |       |       |                                                                                                                                                                                                      |
|---------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| PARN    | 97%   | 95.3% | 100% | 100%  | 99.5% | Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371                                                            |
| PARP4   | 100%  | 100%  | 100% | 99.9% | 99.4% |                                                                                                                                                                                                      |
| PAX5    | 100%  | 100%  | 100% | 100%  | 99.1% | {Leukemia, acute lymphoblastic, susceptibility to, 3}, 615545                                                                                                                                        |
| PLEKHM1 | 100%  | 100%  | 100% | 99.7% | 98.5% | ?Osteopetrosis, autosomal recessive 6, 611497;Osteopetrosis, autosomal dominant 3, 618107                                                                                                            |
| PMS2    | 93.7% | 93.4% | 100% | 99.9% | 99.2% | Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101                                                                                                                                   |
| POT1    | 100%  | 100%  | 100% | 100%  | 99.5% | Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 |
| PRDX2   | 100%  | 99.7% | 100% | 99.8% | 97.7% |                                                                                                                                                                                                      |

|        |       |       |      |       |       |                                                                                                                                    |
|--------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------|
| PRF1   | 100%  | 100%  | 100% | 100%  | 99.6% | Hemophagocytic lymphohistiocytosis, familial, 2, 603553;Aplastic anemia, 609135;Lymphoma, non-Hodgkin, 605027                      |
| PTPN11 | 89.4% | 89.2% | 100% | 100%  | 99.5% | Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785 |
| RAD51  | 89.3% | 89.3% | 100% | 100%  | 99.1% | Mirror movements 2, 614508;{Breast cancer, susceptibility to}, 114480;Fanconi anemia, complementation group R, 617244              |
| RAD51C | 90.3% | 90.3% | 100% | 99.9% | 98.7% | {Breast-ovarian cancer, familial, susceptibility to, 3}, 613399;Fanconi anemia, complementation group O, 613390                    |
| RBBP6  | 100%  | 100%  | 100% | 100%  | 99.2% |                                                                                                                                    |
| RBM8A  | 100%  | 100%  | 100% | 100%  | 99%   | Thrombocytopenia-absent radius syndrome, 274000                                                                                    |

|        |       |       |      |       |       |                                                                                                                     |
|--------|-------|-------|------|-------|-------|---------------------------------------------------------------------------------------------------------------------|
| RFWD3  | 100%  | 100%  | 100% | 99.9% | 99.2% | ?Fanconi anemia, complementation group W, 617784                                                                    |
| RMRP   |       |       |      |       |       | Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250 |
| RPA1   | 100%  | 100%  | 100% | 99.9% | 99.3% | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767                                 |
| RPL11  | 100%  | 100%  | 100% | 100%  | 99.1% | Diamond-Blackfan anemia 7, 612562                                                                                   |
| RPL15  | 99.2% | 93.4% | 100% | 100%  | 99.4% | Diamond-Blackfan anemia 12, 615550                                                                                  |
| RPL18  | 100%  | 100%  | 100% | 100%  | 99.5% | ?Diamond-Blackfan anemia 18, 618310                                                                                 |
| RPL26  | 100%  | 100%  | 100% | 100%  | 99.6% | ?Diamond-Blackfan anemia 11, 614900                                                                                 |
| RPL27  | 100%  | 99.9% | 100% | 100%  | 99.7% | ?Diamond-Blackfan anemia 16, 617408                                                                                 |
| RPL31  | 100%  | 99.9% | 100% | 100%  | 99%   |                                                                                                                     |
| RPL35  | 100%  | 100%  | 100% | 100%  | 99.6% | ?Diamond-Blackfan anemia 19, 618312                                                                                 |
| RPL35A | 100%  | 100%  | 100% | 100%  | 99.7% | Diamond-Blackfan anemia 5, 612528                                                                                   |
| RPL4   | 100%  | 100%  | 100% | 100%  | 99.4% |                                                                                                                     |

|        |       |       |      |      |       |                                                                    |
|--------|-------|-------|------|------|-------|--------------------------------------------------------------------|
| RPL5   | 100%  | 100%  | 100% | 100% | 99.8% | Diamond-Blackfan anemia 6, 612561                                  |
| RPL9   | 100%  | 100%  | 100% | 100% | 99.6% |                                                                    |
| RPS10  | 100%  | 100%  | 100% | 100% | 99.4% | Diamond-Blackfan anemia 9, 613308                                  |
| RPS15A | 79.7% | 79.7% | 100% | 100% | 99.5% | ?Diamond-Blackfan anemia 20, 618313                                |
| RPS17  | 100%  | 100%  | 100% | 100% | 99.4% | Diamond-Blackfan anemia 4, 612527                                  |
| RPS19  | 100%  | 99.8% | 100% | 100% | 99.1% | Diamond-Blackfan anemia 1, 105650                                  |
| RPS24  | 100%  | 100%  | 100% | 100% | 99.6% | Diamond-blackfan anemia 3, 610629                                  |
| RPS26  | 100%  | 97.7% | 100% | 100% | 99.2% | Diamond-Blackfan anemia 10, 613309                                 |
| RPS27  | 100%  | 100%  | 100% | 100% | 99.5% | ?Diamond-Blackfan anemia 17, 617409                                |
| RPS28  | 100%  | 100%  | 100% | 100% | 97.9% | Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164 |
| RPS29  | 100%  | 100%  | 100% | 100% | 99.8% | Diamond-Blackfan anemia 13, 615909                                 |
| RPS7   | 100%  | 100%  | 100% | 100% | 99.5% | Diamond-Blackfan anemia 8, 612563                                  |

|        |      |      |      |       |       |                                                                                                                                                                                                |
|--------|------|------|------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| RTEL1  | 100% | 100% | 100% | 100%  | 99%   | Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 |
| RUNX1  | 100% | 100% | 100% | 99.4% | 95.9% | Platelet disorder, familial, with associated myeloid malignancy, 601399;Leukemia, acute myeloid, 601626                                                                                        |
| SAMD9  | 100% | 100% | 100% | 99.9% | 99.5% | Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053                                                      |
| SAMD9L | 100% | 100% | 100% | 100%  | 99.8% | Ataxia-pancytopenia syndrome, 159550;?Spinocerebellar ataxia 49, 619806;Monosomy 7 myelodysplasia and leukemia syndrome 1, 252270                                                              |

|          |      |       |       |       |       |                                                                                                                                |
|----------|------|-------|-------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------|
| SBDS     | 100% | 100%  | 100%  | 100%  | 99.7% | {Aplastic anemia, susceptibility to}, 609135;Shwachman-Diamond syndrome 1, 260400                                              |
| SEC23B   | 100% | 100%  | 100%  | 100%  | 99.6% | ?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100                                               |
| SH2B3    | 100% | 100%  | 100%  | 100%  | 99.3% | Thrombocythemia, somatic, 187950;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100                                |
| SH2D1A   | 100% | 100%  | 98.9% | 89.1% | 68.7% | Lymphoproliferative syndrome, X-linked, 1, 308240                                                                              |
| SLC19A2  | 100% | 100%  | 100%  | 100%  | 99.6% | Thiamine-responsive megaloblastic anemia syndrome, 249270                                                                      |
| SLC25A38 | 100% | 100%  | 100%  | 100%  | 99.2% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950                                                                        |
| SLC37A4  | 100% | 99.9% | 100%  | 100%  | 99.4% | Glycogen storage disease Ib, 232220;Congenital disorder of glycosylation, type IIw, 619525;Glycogen storage disease Ic, 232240 |

|         |       |       |      |       |       |                                                                                                |
|---------|-------|-------|------|-------|-------|------------------------------------------------------------------------------------------------|
| SLC46A1 | 100%  | 100%  | 100% | 99.9% | 98.6% | Folate malabsorption, hereditary, 229050                                                       |
| SLC4A2  | 100%  | 100%  | 100% | 99.9% | 98.8% | ?Osteopetrosis, autosomal recessive 9, 620366                                                  |
| SLX4    | 100%  | 100%  | 100% | 99.9% | 98.9% | Fanconi anemia, complementation group P, 613951                                                |
| SNX10   | 89.3% | 89.3% | 100% | 100%  | 99.7% | Osteopetrosis, autosomal recessive 8, 615085                                                   |
| SOS1    | 98.8% | 98.5% | 100% | 100%  | 99.4% | Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300                                    |
| SRP54   | 100%  | 100%  | 100% | 100%  | 99.8% | Neutropenia, severe congenital, 8, autosomal dominant, 618752                                  |
| SRP72   | 100%  | 100%  | 100% | 100%  | 99.6% | Bone marrow failure syndrome 1, 614675                                                         |
| STIM1   | 100%  | 99.7% | 100% | 100%  | 99.5% | Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783 |
| STN1    | 87.1% | 87%   | 100% | 100%  | 99.4% | Cerebroretinal microangiopathy with calcifications and cysts 2, 617341                         |

|         |       |       |       |       |       |                                                                                                                                                                                                                                                                                                                     |
|---------|-------|-------|-------|-------|-------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TFAZZIN | 100%  | 99.8% | 98.3% | 86.1% | 64.5% | Barth syndrome,<br>302060                                                                                                                                                                                                                                                                                           |
| TBXAS1  | 100%  | 100%  | 100%  | 100%  | 99.6% | Ghosal<br>hematodiaphyseal<br>syndrome, 231095                                                                                                                                                                                                                                                                      |
| TCIRG1  | 100%  | 100%  | 100%  | 100%  | 99%   | Osteopetrosis,<br>autosomal recessive 1,<br>259700                                                                                                                                                                                                                                                                  |
| TERC    |       |       |       |       |       | Pulmonary fibrosis<br>and/or bone marrow<br>failure syndrome,<br>telomere-related, 2,<br>614743;Dyskeratosis<br>congenita, autosomal<br>dominant 1, 127550                                                                                                                                                          |
| TERF2IP | 99.7% | 94.2% | 100%  | 100%  | 99.5% |                                                                                                                                                                                                                                                                                                                     |
| TERT    | 100%  | 100%  | 100%  | 100%  | 99%   | Dyskeratosis congenita,<br>autosomal dominant 2,<br>613989;Dyskeratosis<br>congenita, autosomal<br>recessive 4,<br>613989;Pulmonary<br>fibrosis and/or bone<br>marrow failure<br>syndrome, telomere-<br>related, 1,<br>614742;{Melanoma,<br>cutaneous malignant,<br>9}, 615134;{Leukemia,<br>acute myeloid}, 601626 |

|           |       |       |       |       |       |                                                                                                                                    |
|-----------|-------|-------|-------|-------|-------|------------------------------------------------------------------------------------------------------------------------------------|
| TET2      | 100%  | 100%  | 100%  | 100%  | 99.5% | Myelodysplastic syndrome, somatic, 614286;Immunodeficiency 75, 619126                                                              |
| THPO      | 100%  | 100%  | 100%  | 100%  | 99.6% | Thrombocythemia 1, 187950;Thrombocytopenia 9, 620478;Amegakaryocytic thrombocytopenia, congenital, 2, 620481                       |
| TINF2     | 100%  | 100%  | 100%  | 99.9% | 98.7% | Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130                                                       |
| TLR8      | 100%  | 100%  | 99.3% | 91.5% | 73.8% | Immunodeficiency 98 with autoinflammation, X-linked, 301078                                                                        |
| TNFRSF11A | 99.6% | 98.6% | 100%  | 99.9% | 99%   | Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810 |
| TNFSF11   | 100%  | 100%  | 100%  | 100%  | 99.4% | Osteopetrosis, autosomal recessive 2, 259710                                                                                       |

|       |       |       |       |       |       |                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
|-------|-------|-------|-------|-------|-------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TP53  | 94.7% | 94.7% | 100%  | 100%  | 99%   | {Basal cell carcinoma 7},<br>614740;{Adrenocortical carcinoma, pediatric},<br>202300;Hepatocellular carcinoma, somatic,<br>114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome,<br>151623;Pancreatic cancer, somatic,<br>260350;Nasopharyngeal carcinoma, somatic,<br>607107;{Osteosarcoma }, 259500;{Choroid plexus papilloma},<br>260500;{Colorectal cancer},<br>114500;{Glioma susceptibility 1},<br>137800;Bone marrow failure syndrome 5,<br>618165 |
| TPM4  | 100%  | 99.8% | 100%  | 99.8% | 98.5% | Bleeding disorder, platelet-type, 25,<br>620486                                                                                                                                                                                                                                                                                                                                                                                                                 |
| TSR2  | 100%  | 99.9% | 98.4% | 87.8% | 69.2% | ?Diamond-Blackfan anemia 14 with<br>mandibulofacial dysostosis, 300946                                                                                                                                                                                                                                                                                                                                                                                          |
| TUBB1 | 100%  | 100%  | 100%  | 99.9% | 99.5% | Macrothrombocytopenia, isolated, 1,<br>autosomal dominant,<br>613112                                                                                                                                                                                                                                                                                                                                                                                            |

|        |       |       |       |       |       |                                                                                                                                                                                                     |
|--------|-------|-------|-------|-------|-------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| TYK2   | 100%  | 100%  | 100%  | 99.9% | 98.4% | Immunodeficiency 35,<br>611521                                                                                                                                                                      |
| UBA1   | 100%  | 99.7% | 98.9% | 87.8% | 68%   | Spinal muscular<br>atrophy, X-linked 2,<br>infantile,<br>301830;VEXAS<br>syndrome, somatic,<br>301054                                                                                               |
| UBE2T  | 100%  | 100%  | 100%  | 99.8% | 98.7% | Fanconi anemia,<br>complementation group<br>T, 616435                                                                                                                                               |
| USB1   | 95%   | 93.2% | 100%  | 100%  | 99%   | Poikiloderma with<br>neutropenia, 604173                                                                                                                                                            |
| VPS13B | 100%  | 100%  | 100%  | 100%  | 99.5% | Cohen syndrome,<br>216550                                                                                                                                                                           |
| VPS45  | 87.7% | 86.8% | 100%  | 100%  | 99.2% | Neutropenia, severe<br>congenital, 5,<br>autosomal recessive,<br>615285                                                                                                                             |
| VPS4A  | 100%  | 100%  | 100%  | 99.8% | 97.6% | CIMDAG syndrome,<br>619273                                                                                                                                                                          |
| WAS    | 98.7% | 91.4% | 97.2% | 84.1% | 63.5% | Wiskott-Aldrich<br>syndrome,<br>301000;Neutropenia,<br>severe congenital, X-<br>linked,<br>300299;Thrombocytope<br>nia, X-linked,<br>intermittent,<br>313900;Thrombocytope<br>nia, X-linked, 313900 |

|        |      |      |      |      |       |                                                                                                                         |
|--------|------|------|------|------|-------|-------------------------------------------------------------------------------------------------------------------------|
| WRAP53 | 100% | 100% | 100% | 100% | 98.7% | Dyskeratosis congenita, autosomal recessive 3, 613988                                                                   |
| XRCC2  | 100% | 100% | 100% | 100% | 99.6% | Spermatogenic failure 50, 619145;?Premature ovarian failure 17, 619146;?Fanconi anemia, complementation group U, 617247 |
| YARS2  | 100% | 100% | 100% | 100% | 99.5% | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561                                                           |
| ZCCHC8 | 100% | 100% | 100% | 100% | 99.4% | ?Pulmonary fibrosis and/or bone marrow failure syndrome, 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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

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 : November 25th, 2024.

This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors