# IRON DISORDERS GENE PANEL DG 2.17 ( 54 genes)

Releasedate: 06-12-2019

| *Gene* | *Median Coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|
| ABCB10 | 63.2 | 87.5% | 73.7% | No OMIM Disease ID |
| ABCB7 | 125.6 | 99.9% | 98.8% | Anemia, sideroblastic, with ataxia, 301310 |
| ACVR1 | 139.8 | 100.0% | 100.0% | Fibrodysplasia ossificans progressiva, 135100 |
| ALAS2 | 77.2 | 99.1% | 95.5% | Protoporphyria, erythropoietic, X-linked, 300752<br>Anemia, sideroblastic, 1, 300751 |
| ATP4A | 150.0 | 100.0% | 99.9% | No OMIM Disease ID |
| BMP6 | 153.2 | 99.8% | 98.2% | No OMIM Disease ID |
| C15orf41 | 121.9 | 100.0% | 99.4% | Dyserythropoietic anemia, congenital, type Ib, 615631 |
| CALR | 118.8 | 98.9% | 92.5% | Myelofibrosis, somatic, 254450<br>Thrombocythemia, somatic, 187950 |
| CCL2 | 134.9 | 100.0% | 100.0% | No OMIM disease ID |
| CDAN1 | 123.7 | 100.0% | 99.8% | Dyserythropoietic anemia, congenital, type Ia, 224120 |
| CP | 99.2 | 92.9% | 87.3% | Hemosiderosis, systemic, due to aceruloplasminemia, 604290<br>Cerebellar ataxia, 604290 |
| CYBRD1 | 127.4 | 100.0% | 99.8% | No OMIM Disease ID |
| EXOC6 | 101.7 | 99.0% | 95.9% | No OMIM Disease ID |
| FECH | 107.9 | 100.0% | 99.6% | Protoporphyria, erythropoietic, 1, 177000 |
| FTH1 | 70.5 | 99.2% | 91.7% | ?Hemochromatosis, type 5, 615517 |
| FTL | 164.3 | 100.0% | 98.4% | Hyperferritinemia-cataract syndrome, 600886<br>Neurodegeneration with brain iron accumulation 3, 606159<br>L-ferritin deficiency, dominant and recessive, 615604 |
| FXN | 67.4 | 100.0% | 98.3% | Friedreich ataxia with retained reflexes, 229300<br>Friedreich ataxia, 229300 |
| GATA1 | 102.0 | 99.9% | 99.0% | Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685<br>Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835<br>Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367<br>Thrombocytopenia with beta-thalassemia, X-linked, 314050 |
| GLRX5 | 149.3 | 99.8% | 97.8% | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860<br>Spasticity, childhood-onset, with hyperglycinemia, 616859 |
| HAMP | 185.1 | 100.0% | 100.0% | Hemochromatosis, type 2B, 613313 |

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|---|---|---|---|---|
| HEPH | 70.9 | 98.3% | 90.7% | No OMIM Disease ID |
| HFE | 114.7 | 100.0% | 99.1% | Hemochromatosis, 235200 |
| HFE2 | 162.3 | 100.0% | 100.0% | Hemochromatosis, type 2A, 602390 |
| HMOX1 | 153.8 | 98.0% | 91.0% | Heme oxygenase-1 deficiency, 614034 |
| HSCB | 114.1 | 99.8% | 97.1% | No OMIM Disease ID |
| HSPA9 | 83.8 | 88.2% | 84.2% | Even-plus syndrome, 616854<br>Anemia, sideroblastic, 4, 182170 |
| JAK2 | 100.6 | 97.1% | 95.0% | Myelofibrosis, somatic, 254450<br>Thrombocythemia 3, 614521<br>Polycythemia vera, somatic, 263300<br>Leukemia, acute myeloid, somatic, 601626<br>Erythrocytosis, somatic, 133100 |
| KIF23 | 145.4 | 99.3% | 96.4% | No OMIM Disease ID |
| KLF1 | 132.7 | 100.0% | 100.0% | Blood group--Lutheran inhibitor, 111150<br>Dyserythropoietic anemia, congenital, type IV, 613673 |
| LARS2 | 128.3 | 100.0% | 100.0% | Perrault syndrome 4, 615300<br>?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 |
| LPIN2 | 101.1 | 100.0% | 99.7% | Majeed syndrome, 609628 |
| MPL | 134.4 | 100.0% | 99.9% | Thrombocythemia 2, 601977<br>Thrombocytopenia, congenital amegakaryocytic, 604498<br>Myelofibrosis with myeloid metaplasia, somatic, 254450 |
| NCOA4 | 107.5 | 95.8% | 91.6% | No OMIM Disease ID |
| NDUFB11 | 110.0 | 99.2% | 96.2% | Linear skin defects with multiple congenital anomalies 3, 300952<br>?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| PANK2 | 161.5 | 100.0% | 100.0% | HARP syndrome, 607236<br>Neurodegeneration with brain iron accumulation 1, 234200 |
| PUS1 | 125.7 | 99.8% | 98.1% | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462 |
| SEC23B | 132.1 | 99.7% | 98.2% | ?Cowden syndrome 7, 616858<br>Dyserythropoietic anemia, congenital, type II, 224100 |
| SF3B1 | 126.8 | 99.6% | 98.5% | Myelodysplastic syndrome, somatic, 614286 |
| SFXN4 | 126.1 | 100.0% | 99.7% | Combined oxidative phosphorylation deficiency 18, 615578 |
| SLC11A2 | 97.4 | 99.9% | 98.5% | Anemia, hypochromic microcytic, with iron overload 1, 206100 |
| SLC19A2 | 103.2 | 100.0% | 99.7% | Thiamine-responsive megaloblastic anemia syndrome, 249270 |
| SLC25A37 | 229.4 | 100.0% | 100.0% | No OMIM Disease ID |
| SLC25A38 | 98.5 | 99.0% | 95.5% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950 |
| SLC40A1 | 121.3 | 100.0% | 99.9% | Hemochromatosis, type 4, 606069 |
| SLC46A1 | 121.7 | 100.0% | 98.0% | Folate malabsorption, hereditary, 229050 |

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| STEAP3 | 185.9 | 100.0% | 99.6% | ?Anemia, hypochromic microcytic, with iron overload 2, 615234 |
| TF | 106.6 | 100.0% | 99.8% | Atransferrinemia, 209300 |
| TFR2 | 138.0 | 99.8% | 98.7% | Hemochromatosis, type 3, 604250 |
| TFRC | 133.1 | 99.9% | 99.0% | Immunodeficiency 46, 616740 |
| TMEM14C | 95.8 | 100.0% | 98.5% | No OMIM Disease ID |
| TMPRSS6 | 117.1 | 100.0% | 99.8% | Iron-refractory iron deficiency anemia, 206200 |
| TRNT1 | 100.7 | 99.2% | 95.3% | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084<br>Retinitis pigmentosa and erythrocytic microcytosis, 616959 |
| UROS | 104.6 | 100.0% | 99.9% | Porphyria, congenital erythropoietic, 263700 |
| YARS2 | 188.2 | 100.0% | 99.4% | Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561 |

*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 : December 11th , 2019.*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*