

# IRON DISORDERS PANEL DG-5.0.0 (46 GENES)

| Gene   | Twist X2 covered 10x | Twist X2 covered 20x | srWGS covered 10x | srWGS covered 15x | srWGS covered 20x | Associated Phenotype description and OMIM disease ID                                      |
|--------|----------------------|----------------------|-------------------|-------------------|-------------------|-------------------------------------------------------------------------------------------|
| ABCB7  | 100%                 | 100%                 | 99.2%             | 90.7%             | 71.4%             | Anemia, sideroblastic, with ataxia, 301310                                                |
| ALAS2  | 100%                 | 100%                 | 98.4%             | 87.7%             | 67.8%             | Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752         |
| BMP6   | 100%                 | 100%                 | 100%              | 100%              | 99.2%             | {Iron overload, susceptibility to}, 620121                                                |
| CALR   | 100%                 | 100%                 | 100%              | 100%              | 99.6%             | Myelofibrosis, somatic, 254450;Thrombocythemia, somatic, 187950                           |
| CDAN1  | 100%                 | 100%                 | 100%              | 100%              | 99.4%             | Dyserythropoietic anemia, congenital, type Ia, 224120                                     |
| CDIN1  | 100%                 | 100%                 | 100%              | 100%              | 99.8%             | Dyserythropoietic anemia, congenital, type Ib, 615631                                     |
| CP     | 100%                 | 100%                 | 100%              | 100%              | 99.9%             | Aceruloplasminemia, 604290                                                                |
| CYBRD1 | 100%                 | 100%                 | 100%              | 100%              | 99.5%             |                                                                                           |
| FECH   | 100%                 | 100%                 | 100%              | 100%              | 99.7%             | Protoporphyria, erythropoietic, 1, 177000                                                 |
| FTH1   | 100%                 | 100%                 | 100%              | 100%              | 99.6%             | Neurodegeneration with brain iron accumulation 9, 620669;?Hemochromatosis, type 5, 615517 |

|       |      |       |       |       |       |                                                                                                                                                                                                                                                                                                                                                           |
|-------|------|-------|-------|-------|-------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| FTL   | 100% | 100%  | 100%  | 99.9% | 99.1% | Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159                                                                                                                                                                                                |
| GATA1 | 100% | 100%  | 98.3% | 86.3% | 64.4% | 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 |
| GLRX5 | 100% | 100%  | 100%  | 99.6% | 99%   | Anemia, sideroblastic, 3, pyridoxine-refractory, 616860;Spasticity, childhood-onset, with hyperglycinemia, 616859                                                                                                                                                                                                                                         |
| HAMP  | 100% | 100%  | 100%  | 99.9% | 98.7% | Hemochromatosis, type 2B, 613313                                                                                                                                                                                                                                                                                                                          |
| HFE   | 100% | 99.9% | 100%  | 99.9% | 99.4% | Hemochromatosis, type 1, 235200                                                                                                                                                                                                                                                                                                                           |
| HJV   | 100% | 100%  | 100%  | 100%  | 99.6% | Hemochromatosis, type 2A, 602390                                                                                                                                                                                                                                                                                                                          |
| HMOX1 | 100% | 100%  | 100%  | 100%  | 99.8% | Heme oxygenase-1 deficiency, 614034;{Pulmonary disease, chronic obstructive, susceptibility to}, 606963                                                                                                                                                                                                                                                   |

|       |       |       |      |       |       |                                                                                                                                                                                                              |
|-------|-------|-------|------|-------|-------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| HSCB  | 85.5% | 85.5% | 100% | 100%  | 99.9% | ?Anemia, sideroblastic, 5, 619523                                                                                                                                                                            |
| HSPA9 | 100%  | 100%  | 100% | 99.9% | 99.6% | Even-plus syndrome, 616854;Anemia, sideroblastic, 4, 182170                                                                                                                                                  |
| JAK2  | 100%  | 100%  | 100% | 100%  | 99.5% | {Budd-Chiari syndrome, somatic}, 600880;Myelofibrosis, somatic, 254450;Erythrocytosis, somatic, 133100;Leukemia, acute myeloid, somatic, 601626;Thrombocythemia 3, 614521;Polycythemia vera, somatic, 263300 |
| KIF23 | 97.7% | 97.7% | 100% | 100%  | 99.8% | Anemia, congenital dyserythropoietic, type IIIA, 105600                                                                                                                                                      |
| KLF1  | 100%  | 100%  | 100% | 100%  | 99.9% | Blood group--Lutheran inhibitor, 111150;[Hereditary persistence of fetal hemoglobin], 613566;Anemia, dyserythropoietic congenital, type IVa, 613673;Anemia, congenital dyserythropoietic, type IVb, 620969   |
| LARS2 | 96.2% | 96.2% | 100% | 100%  | 99.8% | Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021                                                                                                                       |
| LPIN2 | 99.6% | 99.2% | 100% | 100%  | 99.6% | Majeed syndrome, 609628                                                                                                                                                                                      |
| MPL   | 100%  | 100%  | 100% | 100%  | 99.6% | Myelofibrosis with myeloid metaplasia, somatic, 254450;Amegakaryocytic thrombocytopenia, congenital, 1, 604498;Thrombocythemia 2, 601977                                                                     |

|          |       |       |       |       |       |                                                                                                                               |
|----------|-------|-------|-------|-------|-------|-------------------------------------------------------------------------------------------------------------------------------|
| NDUFB11  | 99.8% | 98.8% | 91.2% | 75.8% | 59%   | Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| PANK2    | 100%  | 100%  | 100%  | 100%  | 99.8% | Neurodegeneration with brain iron accumulation 1, 234200                                                                      |
| PUS1     | 100%  | 100%  | 100%  | 99.9% | 99.4% | Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462                                                                 |
| RACGAP1  | 97.7% | 94.7% | 100%  | 100%  | 99.6% | Anemia, congenital dyserythropoietic, type IIb, autosomal recessive, 619789                                                   |
| SEC23B   | 100%  | 100%  | 100%  | 100%  | 99.7% | ?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100                                              |
| SF3B1    | 100%  | 100%  | 100%  | 100%  | 99.9% | Myelodysplastic syndrome, somatic, 614286                                                                                     |
| SFXN4    | 100%  | 100%  | 100%  | 100%  | 99.8% | Combined oxidative phosphorylation deficiency 18, 615578                                                                      |
| SLC11A2  | 100%  | 100%  | 100%  | 100%  | 99.8% | Anemia, hypochromic microcytic, with iron overload 1, 206100                                                                  |
| SLC19A2  | 100%  | 100%  | 100%  | 100%  | 100%  | Thiamine-responsive megaloblastic anemia syndrome, 249270                                                                     |
| SLC25A38 | 90.6% | 90.6% | 100%  | 100%  | 99.5% | Anemia, sideroblastic, 2, pyridoxine-refractory, 205950                                                                       |
| SLC40A1  | 100%  | 100%  | 100%  | 100%  | 99.8% | Hemochromatosis, type 4, 606069                                                                                               |
| SLC46A1  | 100%  | 100%  | 100%  | 99.9% | 98.9% | Folate malabsorption, hereditary, 229050                                                                                      |

|         |       |       |      |       |       |                                                                                                                                                                |
|---------|-------|-------|------|-------|-------|----------------------------------------------------------------------------------------------------------------------------------------------------------------|
| STEAP3  | 100%  | 100%  | 100% | 100%  | 99.3% | ?Anemia, hypochromic microcytic, with iron overload 2, 615234                                                                                                  |
| TF      | 100%  | 100%  | 100% | 99.9% | 99.5% | Atransferrinemia, 209300                                                                                                                                       |
| TFR2    | 96.1% | 94.6% | 100% | 99.9% | 98.9% | Hemochromatosis, type 3, 604250                                                                                                                                |
| TFRC    | 94.7% | 94.7% | 100% | 99.8% | 99.1% | Immunodeficiency 46, 616740                                                                                                                                    |
| TMEM14C | 100%  | 100%  | 100% | 100%  | 99.8% |                                                                                                                                                                |
| TMPRSS6 | 100%  | 100%  | 100% | 100%  | 99.3% | Iron-refractory iron deficiency anemia, 206200                                                                                                                 |
| TRNT1   | 92%   | 91.9% | 100% | 100%  | 99.8% | Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084;Retinitis pigmentosa and erythrocytic microcytosis, 616959 |
| UROS    | 95.3% | 95.3% | 100% | 100%  | 99.8% | Porphyria, congenital erythropoietic, 263700                                                                                                                   |
| YARS2   | 100%  | 100%  | 100% | 100%  | 99.6% | 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.

*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 5.0.0*

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