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 IIIb, 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.
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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