WES IRON DISORDERS DG 3.7

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
ABCB10	100.0%	100.0%	100.0%	99.3%	
ABCB7	99.8%	99.3%	99.3%	77.3%	Anemia, sideroblastic, with ataxia, 301310
ACVR1	100.0%	99.9%	100.0%	99.7%	Fibrodysplasia ossificans progressiva, 135100
ALAS2	100.0%	99.8%	98.9%	77.2%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	100.0%	100.0%	100.0%	99.2%	
BMP6	100.0%	100.0%	100.0%	99.4%	
C15orf41	100.0%	99.9%	100.0%	99.9%	Dyserythropoietic anemia, congenital, type Ib, 615631
CALR	100.0%	100.0%	100.0%	99.8%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CCL2	100.0%	100.0%	100.0%	98.0%	
CDAN1	100.0%	100.0%	100.0%	99.4%	Dyserythropoietic anemia, congenital, type Ia, 224120
CP	100.0%	100.0%	100.0%	99.0%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CYBRD1	100.0%	100.0%	100.0%	99.2%	
EXOC6	100.0%	100.0%	100.0%	99.1%	
FECH	100.0%	100.0%	100.0%	99.6%	Protoporphyria, erythropoietic, 1, 177000
FTH1	100.0%	100.0%	100.0%	99.8%	?Hemochromatosis, type 5, 615517

FTL	100.0%	100.0%	100.0%	99.0%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	100.0%	100.0%	100.0%	99.0%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GATA1	100.0%	100.0%	98.2%	72.4%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 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 Hemolytic anemia due to elevated adenosine deaminase, 301083
GLRX5	100.0%	100.0%	100.0%	99.6%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
HAMP	100.0%	100.0%	100.0%	97.7%	Hemochromatosis, type 2B, 613313
HEPH	99.8%	99.3%	99.0%	76.0%	
HFE	100.0%	100.0%	100.0%	99.6%	Hemochromatosis, type 1, 235200
HJV	100.0%	100.0%	100.0%	99.6%	Hemochromatosis, type 2A, 602390
HMOX1	100.0%	100.0%	100.0%	99.8%	Heme oxygenase-1 deficiency, 614034
HSCB	100.0%	100.0%	100.0%	99.5%	?Anemia, sideroblastic, 5, 619523

HSPA9	100.0%	100.0%	100.0%	99.4%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
JAK2	100.0%	100.0%	100.0%	99.2%	Myelofibrosis, somatic, 254450 Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300
KIF23	100.0%	100.0%	100.0%	98.9%	Anemia, congenital dyserythropoietic, type IIIA, 105600
KLF1	100.0%	100.0%	100.0%	99.9%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
LARS2	100.0%	100.0%	100.0%	99.5%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LPIN2	100.0%	100.0%	100.0%	99.1%	Majeed syndrome, 609628
MPL	100.0%	100.0%	100.0%	99.5%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
NCOA4	100.0%	100.0%	100.0%	99.2%	
NDUFB11	99.7%	97.9%	93.6%	63.1%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
PANK2	100.0%	100.0%	100.0%	99.6%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200

PUS1	100.0%	100.0%	100.0%	99.8%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
SEC23B	100.0%	100.0%	100.0%	99.3%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SF3B1	100.0%	100.0%	100.0%	99.2%	Myelodysplastic syndrome, somatic, 614286
SFXN4	100.0%	100.0%	100.0%	97.9%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	100.0%	100.0%	100.0%	99.5%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	100.0%	100.0%	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	100.0%	100.0%	100.0%	99.9%	
SLC25A38	100.0%	100.0%	100.0%	99.4%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	100.0%	100.0%	100.0%	99.7%	Hemochromatosis, type 4, 606069
SLC46A1	100.0%	100.0%	100.0%	99.8%	Folate malabsorption, hereditary, 229050
STEAP3	100.0%	100.0%	100.0%	99.8%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	100.0%	100.0%	100.0%	99.7%	Atransferrinemia, 209300
TFR2	100.0%	100.0%	100.0%	99.4%	Hemochromatosis, type 3, 604250
TFRC	100.0%	100.0%	99.9%	98.9%	Immunodeficiency 46, 616740
TMEM14C	100.0%	100.0%	100.0%	99.7%	
TMPRSS6	100.0%	100.0%	100.0%	99.4%	Iron-refractory iron deficiency anemia, 206200

TRNT1	100.0%	100.0%	100.0%	99.2%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
UROS	100.0%	100.0%	100.0%	98.9%	Porphyria, congenital erythropoietic, 263700
YARS2	100.0%	100.0%	100.0%	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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 3.7.0.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors