IRON DISORDERS GENE PANEL DG 2.18 (54 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ABCB10	77,40%	71,20%	99,40%	96,80%	No OMIM disease ID
ABCB7	99,50%	98,20%	99,80%	99,30%	Anemia, sideroblastic, with ataxia, 301310
ACVR1	100%	100%	100%	100%	Fibrodysplasia ossificans progressiva, 135100
ALAS2	98,90%	94,90%	100%	100%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ATP4A	99,90%	98,90%	100%	100%	No OMIM disease ID
BMP6	95,70%	93,60%	99,00%	95,80%	No OMIM disease ID
C15orf41	100%	99,80%	96,30%	96,30%	Dyserythropoietic anemia, congenital, type Ib, 615631
CALR	94,80%	89,10%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CCL2	100%	100%	100%	100%	No OMIM disease ID
CDAN1	100%	99,60%	100%	100%	Dyserythropoietic anemia, congenital, type Ia, 224120
CP	94,80%	88,90%	100%	100%	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CYBRD1	100%	99,90%	100%	100%	No OMIM disease ID
EXOC6	99,20%	96,30%	100%	100%	No OMIM disease ID
FECH	100%	100%	100%	100%	Protoporphyria, erythropoietic, 1, 177000
FTH1	94,00%	76,60%	100%	100%	?Hemochromatosis, type 5, 615517
FTL	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FXN	95,50%	80,10%	100%	100%	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GATA1	99,80%	98,40%	100%	100%	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GLRX5	97,30%	89,10%	99,60%	95,40%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859

HAMP	100%	100%	100%	100%	Hemochromatosis, type 2B, 613313
HEPH	98,80%	91,90%	100%	100%	No OMIM disease ID
HFE	100%	99,70%	100%	100%	Hemochromatosis, 235200
HFE2	100%	100%	100%	100%	Hemochromatosis, type 2A, 602390
HMOX1	98,40%	89,90%	100%	100%	Heme oxygenase-1 deficiency, 614034
HSCB	100%	98,70%	100%	100%	No OMIM disease ID
HSPA9	88,50%	84,50%	100%	100%	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
JAK2	98,10%	95,80%	100%	100%	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
KIF23	99,50%	96,30%	100%	100%	No OMIM disease ID
KLF1	100%	97,80%	100%	100%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673
LARS2	100%	100%	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LPIN2	100%	100%	100%	100%	Majeed syndrome, 609628
MPL	100%	99,50%	100%	100%	Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498 Myelofibrosis with myeloid metaplasia, somatic, 254450
NCOA4	96,40%	93,00%	100%	100%	No OMIM disease ID
NDUFB11	99,50%	96,50%	100%	99,50%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
PANK2	100%	99,30%	100%	100%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	100%	99,50%	99,60%	97,20%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
SEC23B	99,90%	99,30%	100%	100%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SF3B1	99,70%	98,60%	100%	100%	Myelodysplastic syndrome, somatic, 614286
SFXN4	99,90%	98,90%	100%	100%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	100%	99,90%	100%	100%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	100%	99,70%	100%	100%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	100%	100%	100%	100%	No OMIM disease ID
SLC25A38	99,70%	97,10%	100%	100%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	100%	99,50%	100%	100%	Hemochromatosis, type 4, 606069
SLC46A1	99,90%	98,50%	100%	100%	Folate malabsorption, hereditary, 229050

STEAP3	100%	99,70%	100%	100%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	100%	100%	100%	100%	Atransferrinemia, 209300
TFR2	99,10%	97,80%	100%	100%	Hemochromatosis, type 3, 604250
TFRC	100%	99,80%	100%	100%	Immunodeficiency 46, 616740
TMEM14C	100%	99,80%	100%	100%	No OMIM disease ID
TMPRSS6	99,90%	99,10%	100%	100%	Iron-refractory iron deficiency anemia, 206200
TRNT1	99,50%	96,50%	100%	100%	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
UROS	100%	99,90%	100%	100%	Porphyria, congenital erythropoietic, 263700
YARS2	100%	99,80%	100%	100%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors