IRON DISORDERS GENE PANEL DG 3.00 (54 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ABCB10	77,4	71,2	99,4	96,8	No OMIM disease ID
ABCB7	99,5	98,2	99,3	98,8	Anemia, sideroblastic, with ataxia, 301310
ACVR1	100	100	100	100	Fibrodysplasia ossificans progressiva, 135100
ALAS2	98,9	94,9	100	100	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ATP4A	99,9	98,9	100	100	No OMIM disease ID
BMP6	95,7	93,6	99	95,8	No OMIM disease ID
CALR	94,8	89,1	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CCL2	100	100	100	100	{HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940 {Coronary artery disease, modifier of}, 0
CDAN1	100	99,6	100	100	Dyserythropoietic anemia, congenital, type Ia, 224120
C15orf41	85,9	85,6	100	100	Dyserythropoietic anemia, congenital, type Ib, 615631
CP	94,8	88,9	100	100	[Hypoceruloplasminemia, hereditary], 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 Cerebellar ataxia, 604290
CYBRD1	100	99,9	100	100	No OMIM disease ID
EXOC6	99,2	96,3	100	100	No OMIM disease ID
FECH	100	100	100	100	Protoporphyria, erythropoietic, 1, 177000
FTH1	94	76,6	100	100	?Hemochromatosis, type 5, 615517

FTL	98,5	89,4	100	100	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
FXN	95,5	80,1	100	100	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GATA1	99,8	98,4	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	97,3	89,1	99,6	95,4	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
HAMP	100	100	100	100	Hemochromatosis, type 2B, 613313
HEPH	98,8	91,9	100	100	No OMIM disease ID
HFE	100	99,7	100	100	{Porphyria variegata, susceptibility to}, 176200 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 Hemochromatosis, 235200
HJV	100	100	100	100	Hemochromatosis, type 2A, 602390
HMOX1	98,4	89,9	100	100	{Pulmonary disease, chronic obstructive, susceptibility to}, 606963 Heme oxygenase-1 deficiency, 614034
HSCB	100	98,7	100	100	No OMIM disease ID
HSPA9	88,5	84,5	100	100	Even-plus syndrome, 616854 Anemia, sideroblastic, 4, 182170
JAK2	98,1	95,8	100	100	Myelofibrosis, somatic, 254450 Thrombocythemia 3, 614521 Polycythemia vera, somatic, 263300 {Budd-Chiari syndrome, somatic}, 600880 Leukemia, acute myeloid, somatic, 601626 Erythrocytosis, somatic, 133100
KIF23	99,5	96,3	100	100	No OMIM disease ID
KLF1	100	97,8	100	100	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 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	99,5	100	100	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocytopenia, congenital amegakaryocytic, 604498 Thrombocythemia 2, 601977
NCOA4	96,4	93	100	100	No OMIM disease ID
NDUFB11	99,5	96,5	100	99,5	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
PANK2	100	99,3	100	100	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	100	99,5	99,6	97,2	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
SEC23B	99,9	99,3	100	100	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SF3B1	99,7	98,6	100	100	Myelodysplastic syndrome, somatic, 614286
SFXN4	99,9	98,9	100	100	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	98,2	98	100	100	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	100	99,7	100	100	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	100	100	100	100	No OMIM disease ID
SLC25A38	97,9	95,3	100	100	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	100	99,5	100	100	Hemochromatosis, type 4, 606069
SLC46A1	99,9	98,5	100	100	Folate malabsorption, hereditary, 229050
STEAP3	100	99,7	100	100	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	100	100	100	100	Atransferrinemia, 209300
TFR2	99,1	97,8	100	100	Hemochromatosis, type 3, 604250
TFRC	100	99,8	100	100	Immunodeficiency 46, 616740
TMEM14C	100	99,8	100	100	No OMIM disease ID
TMPRSS6	99,9	99,1	100	100	Iron-refractory iron deficiency anemia, 206200

TRNT1	99,5	96,5	100	100	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 Retinitis pigmentosa and erythrocytic microcytosis, 616959
UROS	100	99,9	100	100	Porphyria, congenital erythropoietic, 263700
YARS2	100	99,8	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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors