IRON DISORDERS GENE PANEL DG 2.16 (53 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
ABCB10	60,9	85.4%	71.8%	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB7	126,2	99.9%	98.6%	Anemia, sideroblastic, with ataxia, 301310
ALAS2	74,7	98.9%	94.7%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	136,3	100.0%	99.6%	No OMIM-phenotype Gastric neuroendocrine tumor, type 1 (Calvete (2015) Hum Mol Genet 24,2914)
BMP6	137,8	99.0%	96.6%	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
C15orf41	122	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ib, 615631
CALR	111,8	98.1%	91.7%	Myelofibrosis, somatic, 254450 Thrombocythemia, somatic, 187950
CCL2	133	100.0%	100.0%	{Coronary artery disease, modifier of}, 0 {HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940
CDAN1	112,4	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ia, 224120
CP	100,6	93.1%	87.4%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CYBRD1	122,5	100.0%	99.6%	No OMIM phenotype Iron overload (Zaahl (2004) Hum Genet 115,409 {Haemochromatosis,phenotype modifier,association with} (Constantine (2009) Br J Haematol 147,140)
EXOC6	104	99.0%	96.5%	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	104	100.0%	99.7%	Protoporphyria, erythropoietic, 1, 177000
FTH1	66,2	98.7%	87.9%	?Hemochromatosis, type 5, 615517
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159

FXN	64,9	99.7%	96.8%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GATA1	92,9	99.9%	98.2%	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050 Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367
GLRX5	137,6	99.6%	96.1%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
HAMP	169	100.0%	100.0%	Hemochromatosis, type 2B, 613313
HEPH	68,3	97.8%	89.2%	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HFE	108	100.0%	98.9%	Hemochromatosis, 235200 [Transferrin serum level QTL2], 614193 {Alzheimer disease, susceptibility to}, 104300 {Microvascular complications of diabetes 7}, 612635 {Porphyria cutanea tarda, susceptibility to}, 176100 {Porphyria variegata, susceptibility to}, 176200
HFE2	NC	NC	NC	Hemochromatosis, type 2A, 602390
HMOX1	137,4	96.5%	90.7%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HSCB	104,8	99.5%	97.2%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSPA9	82,6	89.5%	84.2%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
JAK2	103,5	97.6%	95.0%	Erythrocytosis, somatic, 133100 Leukemia, acute myeloid, somatic, 601626 Myelofibrosis, somatic, 254450 Polycythemia vera, somatic, 263300 Thrombocythemia 3, 614521 {Budd-Chiari syndrome, somatic}, 600880
KIF23	144,7	99.4%	96.8%	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KLF1	115,3	100.0%	99.9%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
LARS2	122,8	100.0%	100.0%	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 Perrault syndrome 4, 615300

LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
MPL	125,8	100.0%	99.8%	Myelofibrosis with myeloid metaplasia, somatic, 254450 Thrombocythemia 2, 601977 Thrombocytopenia, congenital amegakaryocytic, 604498
NCOA4	101,7	96.7%	92.2%	?Thyroid cancer,nonmedullary,1},188550
NDUFB11	103,3	98.6%	95.0%	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
PANK2	154,1	100.0%	100.0%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	113,3	99.8%	97.5%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
SEC23B	131	99.8%	99.0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SF3B1	130,9	99.6%	98.3%	Myelodysplastic syndrome, somatic, 614286
SFXN4	124	100.0%	99.7%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	96,2	99.9%	98.7%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	101,3	100.0%	99.6%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	212	100.0%	100.0%	No OMIM phenotype ?anemia and disruptions in ISC biogenesis, inhibition protoporphyrin biosynthesis (Shaw et al. (2006) erythropoietic protophyria (Wang et al. (2011))
SLC25A38	94,5	99.1%	95.2%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	120,8	100.0%	99.8%	Hemochromatosis, type 4, 606069
SLC46A1	111,1	99.9%	98.4%	Folate malabsorption, hereditary, 229050
STEAP3	166,9	100.0%	99.4%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	101,6	100.0%	99.7%	Atransferrinemia, 209300
TFR2	124	99.6%	98.3%	Hemochromatosis, type 3, 604250
TFRC	132	99.9%	99.0%	Immunodeficiency 46, 616740
TMEM14C	95,6	100.0%	97.8%	No OMIM phenotype ?combined porphyria and anemia, severe pathogenic effects are lethal but mild defects might modulate existing anemia and porphyria (Paw et al. (2013), Yien et al. (2014)).
TMPRSS6	107	100.0%	99.4%	Iron-refractory iron deficiency anemia, 206200
TRNT1	101,5	99.2%	96.5%	Retinitis pigmentosa and erythrocytic microcytosis, 616959 Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084
UROS	103,8	100.0%	99.7%	Porphyria, congenital erythropoietic, 263700
YARS2	175,2	99.9%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors