IRON DISORDERS GENE PANEL DG 2.11 (46 genes)

Gene	Median	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCB10	75.2	75	67	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB7	132.4	99	98	Anemia, sideroblastic, with ataxia, 301310
ALAS2	90.3	99	97	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	148.8	100	99	No OMIM-phenotype Gastric neuroendocrine tumor, type 1 (Calvete (2015) Hum Mol Genet 24,2914)
BMP6	108.1	92	90	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
C15orf41	124.8	99	97	Dyserythropoietic anemia, congenital, type Ib, 615631
CCL2	138.6	100	100	{Coronary artery disease, modifier of} {HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940
CDAN1	97.7	97	95	Dyserythropoietic anemia, congenital, type Ia, 224120
CP	120	93	89	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CYBRD1	131.9	100	100	No OMIM phenotype Iron overload (Zaahl (2004) Hum Genet 115,409 {Haemochromatosis,phenotype modifier,association with} (Constantine (2009) Br J Haematol 147,140)
EXOC6	87.1	96	90	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	122	99	99	Protoporphyria, erythropoietic, autosomal recessive, 177000
FTH1	96.3	95	84	?Hemochromatosis, type 5, 615517
FTL	148.1	98	93	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159

FXN	75.3	85	75	Friedreich ataxia with retained reflexes, 229300 Friedreich ataxia, 229300
GATA1	84.2	99	95	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	108.1	92	83	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
HAMP	175.8	100	100	Hemochromatosis, type 2B, 613313
HEPH	86.9	98	93	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HFE	142	100	99	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	117	100	100	Hemochromatosis type 2A,602390
HMOX1	129	95	89	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HSCB	90.2	99	95	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSPA9	91.6	91	85	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
KIF23	170	96	94	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KLF1	52	90	81	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
NCOA4	129.8	94	91	?Thyroid cancer,nonmedullary,1},188550
NDUFB11	110.4	94	88	Linear skin defects with multiple congenital anomalies 3, 300952
PANK2	146.6	99	93	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	127.4	98	94	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462

SEC23B	161.2	97	96	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SFXN4	131.8	100	99	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	135.3	100	99	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	119.3	99	97	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	194	100	100	No OMIM phenotype ?anemia and disruptions in ISC biogenesis, inhibition protoporphyrin biosynthesis (Shaw et al. (2006) erythropoietic protophyria (Wang et al. (2011))
SLC25A38	111.7	99	98	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	155.8	99	99	Hemochromatosis, type 4, 606069
SLC46A1	106.1	99	96	Folate malabsorption, hereditary, 229050
STEAP3	199.2	100	99	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	126	100	100	Atransferrinemia, 209300
TFR2	104.6	98	93	Hemochromatosis, type 3, 604250
TFRC	157.3	99	99	Immunodeficiency 46, 616740
TMEM14C	114.5	100	99	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	102	100	99	Iron-refractory iron deficiency anemia, 206200
UROS	108.2	100	99	Porphyria, congenital erythropoietic, 263700
YARS2	173.3	99	98	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 : April 14th, 2017.
This list is accurate for panel version DG 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors