IRON DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ABCB10	70.3	78%	70%	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB7	156	99%	97%	Anemia, sideroblastic, with ataxia, 301310
ALAS2	107.4	99%	96%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	172.8	99%	98%	No OMIM-phenotype Gastric neuroendocrine tumor, type 1 (Calvete (2015) Hum Mol Genet 24,2914)
BMP6	111.4	92%	89%	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
C15orf41	144.5	100%	99%	Dyserythropoietic anemia, congenital, type Ib, 615631
CCL2	151.5	100%	100%	{Coronary artery disease, modifier of} {HIV-1, resistance to}, 609423 {Mycobacterium tuberculosis, susceptibility to}, 607948 {Spina bifida, susceptibility to}, 182940
CDAN1	113.1	98%	96%	Dyserythropoietic anemia, congenital, type Ia, 224120
CP	141	94%	90%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CYBRD1	149	100%	99%	No OMIM phenotype Iron overload (Zaahl (2004) Hum Genet 115,409 {Haemochromatosis,phenotype modifier,association with} (Constantine (2009) Br J Haematol 147,140)
EXOC6	101	96%	90%	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	142.4	99%	99%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FTH1	99.1	98%	88%	?Hemochromatosis, type 5, 615517
FTL	131.3	99%	92%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
FXN	86.1	86%	76%	Friedreich ataxia with retained reflexes, 229300

				Friedreich ataxia, 229300
GATA1	95	99%	97%	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	102.2	93%	86%	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860 Spasticity, childhood-onset, with hyperglycinemia, 616859
HAMP	192.7	100%	100%	Hemochromatosis, type 2B, 613313
HEPH	100.9	99%	95%	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HFE	155	99%	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	133.3	99%	99%	Hemochromatosis type 2A,602390
HMOX1	142.1	97%	90%	Heme oxygenase-1 deficiency, 614034 {Pulmonary disease, chronic obstructive, susceptibility to}, 606963
HSCB	96.6	98%	94%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSPA9	97.5	89%	85%	Anemia, sideroblastic, 4, 182170 Even-plus syndrome, 616854
KIF23	187.3	95%	93%	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KLF1	60.9	92%	85%	Blood group--Lutheran inhibitor, 111150 Dyserythropoietic anemia, congenital, type IV, 613673 [Hereditary persistence of fetal hemoglobin], 613566
NCOA4	118.3	94%	90%	?Thyroid cancer,nonmedullary,1},188550
NDUFB11	101.4	94%	84%	Linear skin defects with multiple congenital anomalies 3, 300952
PANK2	177.5	99%	96%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	150.8	99%	96%	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462
SEC23B	185.2	97%	96%	Cowden syndrome 7, 616858

				Dyserythropoietic anemia, congenital, type II, 224100
SFXN4	155.3	99%	98%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	146.5	100%	99%	Anemia, hypochromic microcytic, with iron overload 1, 206100
SLC19A2	128.5	99%	98%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	191.5	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	117.7	99%	96%	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950
SLC40A1	164	99%	99%	Hemochromatosis, type 4, 606069
SLC46A1	105.4	98%	94%	Folate malabsorption, hereditary, 229050
STEAP3	186.7	100%	99%	?Anemia, hypochromic microcytic, with iron overload 2, 615234
TF	143.2	100%	100%	Atransferrinemia, 209300
TFR2	105.9	99%	95%	Hemochromatosis, type 3, 604250
TFRC	177.7	99%	99%	Immunodeficiency 46, 616740
TMEM14C	140	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	115.4	99%	99%	Iron-refractory iron deficiency anemia, 206200
UROS	119.9	100%	100%	Porphyria, congenital erythropoietic, 263700
YARS2	186.8	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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