IRON DISORDERS GENE PANEL DG 2.4.x

gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM ID
ABCB10	31.3	63%	54%	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014))
ABCB7	125.2	100%	100%	Anemia, sideroblastic, with ataxia, 301310
ALAS2	84.4	92%	88%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	92.7	98%	92%	No OMIM-phenotype ?iron-deficiency anemia (Krieg et al. (2011)
BMP6	76.9	95%	88%	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008))
C15orf41	90.9	94%	88%	Dyserythropoietic anemia, congenital, type Ib, 615631
CCL2	129.4	100%	100%	No OMIM phenotype ?Hemochromatosis, based on literature (Muckenthaler (submitted 2015))
CDAN1	89.4	100%	97%	Dyserythropoietic anemia, congenital, type Ia,224120
CP	85.2	98%	93%	[Hypoceruloplasminemia, hereditary], 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CYBRD1	72.9	100%	100%	No OMIM phenotype ?anemia (Iolascon et al. (2009))
EXOC6	112.8	100%	99%	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	104.2	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FTH1	54.9	98%	76%	?Hematochromatosis,type 5,615517
FTL	75.6	96%	91%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FXN	83.2	88%	76%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300
GATA1	123.4	99%	97%	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367 Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 Thrombocytopenia with beta-thalassemia, X-linked, 314050
GLRX5	39.1	81%	62%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950

HAMP	112	100%	99%	Hemochromatosis, type 2B, 613313
HEPH	99.4	99%	98%	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004))
HFE	95.6	100%	94%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300
HFE2	103.8	97%	93%	Hemochromatosis type 2A
HMOX1	64	100%	93%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HSCB	92.4	100%	100%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation))
HSPA9	68.8	92%	85%	No OMIM phenotype ?CSA (M.D. Fleming (manuscript in preparation)).5' untranslated region
KIF23	108.7	100%	99%	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013))
KLF1	49.9	100%	95%	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Anemia, dyserythropoietic congenital, type IV, 613673
PANK2	113.2	99%	93%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PUS1	65	99%	96%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
SEC23B	109	100%	100%	Anemia dyserythropoietic congenital type II,224100
SFXN4	87.3	100%	98%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	85.8	100%	98%	Anemia, hypochromic microcytic,206100
SLC19A2	89.2	100%	99%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	146	100%	100%	No OMIM phenotype ?anemia and disruptions in ISC biogenesis, inhibition protoporphyrin biosynthesis (Shaw et al. (2006), Wang et al. (2011); erythropoietic protophyria)
SLC25A38	69.5	100%	95%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC40A1	117.6	99%	96%	Hemochromatosis, type 4, 606069
SLC46A1	84	100%	98%	Folate malabsorption, hereditary, 229050
STEAP3	86.9	95%	90%	?Anemia,hypochromic microcytic,with iron overload 2,615234

TF	99.8	98%	95%	Atransferrinemia, 209300
TFR2	74.2	96%	86%	Hemochromatosis, type 3, 604250
TFRC	111.2	100%	99%	No OMIM phenotype ?microcytic anemia (Wingert et al. (2004), Isolascon et al. (2009)).
TMEM14C	105	100%	100%	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	70.6	98%	92%	Iron-refractory iron deficiency anemia, 206200
UROS	81.4	95%	83%	Porphyria, congenital erythropoietic, 263700
YARS2	99.4	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.
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
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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