IRON DISORDERS GENE PANEL DG 2.5/2.6

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
ABCB10	61.4	72%	67%	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB7	85.3	96%	93%	Anemia, sideroblastic, with ataxia, 301310
ALAS2	63.7	97%	91%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	142.6	99%	98%	No OMIM-phenotype ?iron-deficiency anemia (Krieg et al. (2011) (mice studies)).
BMP6	93.6	93%	89%	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
C15orf41	124.3	100%	100%	Dyserythropoietic anemia, congenital, type Ib, 615631
CCL2	123.8	100%	100%	{Coronary artery disease,modifier of} {HIV-1,resistance to},609423 {Mycobacterium tuberculosis,susceptibility to},607948 {Spina bifida,susceptibility to},192940
CDAN1	87.3	97%	95%	Dyserythropoietic anemia, congenital, type Ia,224120
CP	127.7	94%	91%	Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290 [Hypoceruloplasminemia, hereditary], 604290
CYBRD1	138.6	100%	96%	No OMIM phenotype Iron overload (Zaahl (2004) Hum Genet 115,409 Haemochromatosis,phenotype modifier,association with (Constantine (2009) Br J Haematol 147,140)
EXOC6	86.8	94%	85%	No OMIM phenotype ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	122.4	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FTH1	69.8	97%	83%	?Hematochromatosis,type 5,615517
FTL	104.5	99%	82%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FXN	82	75%	75%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300

GATA1	49.6	97%	90%	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	99.2	90%	84%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
HAMP	162	100%	100%	Hemochromatosis, type 2B, 613313
HEPH	54.4	97%	84%	No OMIM phenotype ?anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HFE	126.5	100%	97%	Hemochromatosis, 235200 {Microvascular complications of diabetes 7}, 612635 {Porphyria variegata, susceptibility to}, 176200 {Porphyria cutanea tarda, susceptibility to}, 176100 {Alzheimer disease, susceptibility to}, 104300 [Transferrin serum level QTL2],614193
HFE2	106.1	100%	100%	Hemochromatosis type 2A
HMOX1	123.9	98%	88%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HSCB	80.6	98%	92%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSPA9	81.8	88%	84%	Anemia,sideroblastic,4,182170 Even-plus syndrome,616854
KIF23	168.2	96%	94%	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KLF1	50.7	90%	83%	Blood group--Lutheran inhibitor, 111150 Dyserthropoietic anemia,congenital,type IV,613673 [Hereditary persistence of fetal hemoglobin], 613566
PANK2	142.9	95%	86%	HARP syndrome, 607236 Neurodegeneration with brain iron accumulation 1, 234200
PUS1	130	99%	97%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
SEC23B	157.3	97%	96%	Anemia dyserythropoietic congenital type II,224100
SFXN4	132	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	130.1	100%	100%	Anemia, hypochromic microcytic,206100
SLC19A2	115.7	97%	92%	Thiamine-responsive megaloblastic anemia syndrome, 249270
SLC25A37	169.8	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	109.8	100%	98%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC40A1	137.7	99%	98%	Hemochromatosis, type 4, 606069
SLC46A1	90.7	97%	88%	Folate malabsorption, hereditary, 229050
STEAP3	144.2	100%	99%	?Anemia,hypochromic microcytic,with iron overload 2,615234
TF	125.8	100%	99%	Atransferrinemia, 209300
TFR2	86.8	98%	95%	Hemochromatosis, type 3, 604250
TFRC	153.5	98%	97%	Immunodeficiency 46,616740
TMEM14C	120.2	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	93.9	100%	98%	Iron-refractory iron deficiency anemia, 206200
UROS	97.3	100%	100%	Porphyria, congenital erythropoietic, 263700
YARS2	157.3	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 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 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