IRON DISORDERS GENE PANEL DGD09072015

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ABCB10	26,8	64%	49%	No OMIM phenotype ?anemia with protoporphyrin IX (PPIX) accumulation (Chen et al. (2009), Yamamoto et al. (2014)).
ABCB7	60,8	100%	97%	Anemia, sideroblastic, with ataxia, 301310
ALAS2	44,4	84%	77%	Anemia, sideroblastic, X-linked, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ATP4A	92,7	100%	93%	No OMIM-phenotype ? iron-deficiency anemia (Krieg et al. (2011) (mice studies)).
BMP6	72	92%	87%	No OMIM phenotype ?hemochromatosis (Babitt et al. (2007), Kautz et al. (2008)).
C15orf41	86,3	94%	91%	Dyserythropoietic anemia, congenital, type Ib, 615631
CCL2	126,4	100%	100%	No OMIM phenotype ?Hemochromatosis (Muckenthaler (submitted 2015).
CDAN1	85,5	99%	96%	Dyserythropoietic anemia, congenital, type Ia,224120
CP	86,8	99%	93%	[Hypoceruloplasminemia, hereditary], 604290 Cerebellar ataxia, 604290 Hemosiderosis, systemic, due to aceruloplasminemia, 604290
CYBRD1	81,2	100%	100%	No OMIM phenotype ?anemia (Iolascon et al. (2009)).
EXOC6	122,7	100%	99%	No OMIM ?Hemoglobin deficit (hypochromic anemia) (Lim et al. (2005), Fleming et al. (2005))
FECH	100,2	100%	100%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FTH1	53,1	81%	67%	No OMIM phenotype ?Hemochromatosis type 5 (3). Kato et al (2001), Arosio et al. (2010).
FTL	84,2	100%	95%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159
FXN	82,9	92%	87%	Friedreich ataxia, 229300 Friedreich ataxia with retained reflexes, 229300

GATA1	54,1	97%	84%	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	40,5	91%	64%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
HAMP	100,4	96%	85%	Hemochromatosis, type 2B, 613313
HEPH	48	97%	83%	No OMIM phenotype ? anemia (Vulpe et al. (1999), Anderson et al. (2002), Chen et al. (2004)).
HFE	91,7	99%	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	110,4	99%	93%	Hemochromatosis type 2A
HMOX1	60,1	100%	92%	Heme oxygenase-1 deficiency, 614034 Pulmonary disease, chronic obstructive, susceptibility to, 606963
HSCB	103,4	100%	100%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
HSPA9	72,5	93%	83%	No OMIM phenotype ?non-syndromic CSA (M.D. Fleming (manuscript in preparation)).
KIF23	109,4	100%	98%	No OMIM phenotype ?Congenital dyserythropoietic anemia type III (CDAIII, Liljeholm et al. (2013)).
KLF1	48,3	98%	88%	Blood group--Lutheran inhibitor, 111150 [Hereditary persistence of fetal hemoglobin], 613566 Anemia, dyserythropoietic congenital, type IV, 613673
PANK2	106,5	100%	94%	Neurodegeneration with brain iron accumulation 1, 234200 HARP syndrome, 607236
PUS1	65,2	99%	96%	Mitochondrial myopathy and sideroblastic anemia 1, 600462
SEC23B	121,1	100%	100%	Anemia dyserythropoietic congenital type II,224100
SFXN4	91	100%	99%	Combined oxidative phosphorylation deficiency 18, 615578
SLC11A2	78,9	99%	98%	Anemia hypochromic microcytic
SLC19A2	87,4	100%	99%	Thiamine-responsive megaloblastic anemia syndrome, 249270

SLC25A37	141,2	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	72,6	100%	96%	Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive, 205950
SLC40A1	112,9	100%	98%	Hemochromatosis, type 4, 606069
SLC46A1	83,4	100%	94%	Folate malabsorption, hereditary, 229050
STEAP3	75,5	96%	90%	No OMIM phenotype
TF	98,8	98%	96%	Atransferrinemia, 209300
TFR2	71,1	96%	86%	Hemochromatosis, type 3, 604250
TFRC	111,9	100%	100%	No OMIM phenotype ?microcytic anemia (Wingert et al. (2004), Isolascon et al. (2009)).
TMEM14C	111,2	100%	100%	No OMIM phenotype ?combined porphyria and anemia (Paw et al. (2013), Yien et al. (2014)).
TMPRSS6	76,9	99%	96%	Iron-refractory iron deficiency anemia, 206200
UROS	72,7	93%	86%	Porphyria, congenital erythropoietic, 263700
YARS2	92,4	100%	100%	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : June 30th, 2015
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors