MAYO CLINIC LABORATORIES

Hereditary Hemolytic Anemia Gene Panel and Subpanel Comparison

Gene		NHHA	NENZ	NMEM	NCDA	Associated Disorders
ABCB6	ATP binding cassette subfamily B member 6	X		X		Pseudohyperkalemia, familial, 2
AHSP	α-hemoglobin stabilizing protein	X				Thalassemia
AK1	Adenylate kinase	X	X			Adenylate kinase deficiency
ALDOA	Aldolase	X	X			Aldolase deficiency
ANK1	Ankyrin	X		X		Hereditary spherocytosis
BCL11A	BAF chromatin remodeling complex subunit BCL11A	X				Fetal hemoglobin (Hb F) levels; Dias-Logan syndrome
CDIN1 (C15ORF41)	CDAN1 interacting nuclease 1	X			X	Congenital dyserythropoietic anemia (CDA) type Ib
CD59	CD59 glycoprotein	X				*CD59*-mediated hemolytic anemia with immune-mediated polyneuropathy)/neonatal anemia
CDAN1	Codanin1	X			X	Congenital dyserythropoietic anemia (CDA) type Ia
EPB41	Protein 4.1	X		X		Hereditary elliptocytosis, pyropoikilocytosis
EPB42	Protein 4.2	X		X		Hereditary spherocytosis
G6PD	Glucose-6 phosphate dehydrogenase	X	X			G6PD deficiency
GATA1	GATA binding protein 1	X			X	X-linked thrombocytopenia with thalassemia/ Congenital dyserythropoietic anemia (CDA) variant
GCLC	Gamma Glutamate Cysteine Ligase	X	X			Glutathione deficiency
GPI	Glucose-6 phosphate isomerase	X	X			Glucose phosphate isomerase (GPI) deficiency
GSR	Glutathione reductase	X	X			Glutathione deficiency
GSS	Glutathione synthetase	X	X			Glutathione deficiency
GYPC	Glycophorin C and D	X		X		Hereditary elliptocytosis
HK1	Hexokinase	X	X			Hexokinase (HK) deficiency
HMOX1	Heme oxygenase-1	X	X			HMOX1 deficiency (hemolysis, Fe accumulation, growth retardation), increased Hb F
KCNN4	Potassium calcium-activated channel subfamily N member 4	X		X		Dehydrated hereditary stomatocytosis-2 (DHS2) (hereditary xerocytosis)
KIF23	Kinesin family member	X			X	Congenital dyserythropoietic anemia (CDA) type III
KLF1	Kruppel-like factor 1	X			X	Congenital dyserythropoietic anemia (CDA) type IV, increased Hb A2, non-deletional HPFH
NT5C3A	5-Nucleotidase cytosolic 3A	X	X			P5NT deficiency (Pyrimidine 5' nucleotidase/ uridine 5' monophosphate hydrolase UMPH1 deficiency)
PFKM	Phosphofructokinase	X	X			Phosphofructokinase (PFK) deficiency (glycogen storage disease VII/Tarui disease)
PGK1	Phosphoglycerate kinase	X	X			Phosphoglycerate kinase 1 (PGK1) deficiency
PGLS	6-Phosphogluconolactonase	X	X			6-phosphogluconolactonase deficiency

MC4091-126rev0225

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Gene		NHHA	NENZ	NMEM	NCDA	Associated Disorders
PIEZO1	PIEZO ion channel component 1	X		X		Dehydrated hereditary stomatocytosis/ hereditary xerocytosis, perinatal edema
PKLR	Pyruvate kinase	X	X			Pyruvate kinase (PK) deficiency
RHAG	Rhesus blood group-associated glycoprotein	X		X		Overhydrated hereditary stomatocytosis, Rh-null hemolytic anemia
SEC23B	SEC23 homolog B, coat complex II component	X			X	Congenital dyserythropoietic anemia (CDA) type II
SLC2A1	Solute carrier family 2 (GLUT1)	X		X		Stomatin-deficient cryohydrocytosis, GLUT1 deficiency
SLC4A1	Solute carrier family 4 (Band3)	X		X		Hereditary spherocytosis, SE Asian ovalocytosis, cryohydrocytosis, stomatocytosis, acanthocytosis, distal renal tubular acidosis with hemolysis
SPTA1	Spectrin, alpha	X		X		Hereditary spherocytosis, hereditary elliptocytosis, pyropoikilocytosis
SPTB	Spectrin, beta	X		X		Hereditary spherocytosis, hereditary elliptocytosis, pyropoikilocytosis
TMPRSS6	Transmembrane serine protease 6	X				Iron-refractory iron deficiency anaemia (IRIDA)
TPI1	Triosephosphate isomerase	X	X			Triosephosphate isomerase (TPI) deficiency