MAYO CLINIC LABORATORIES

Hereditary Erythrocytosis Gene Panel and Subpanel Comparison

Gene		NHEP	NHEM	NCYB	Associated Disorder
ACO1 (IRP1)	Aconitase 1	X			Erythrocytosis/polycythemia
ANKRD26	Ankyrin repeat domain 26	X			Autosomal dominant thrombocytopenia 2
BHLHE41	Basic helix-loop-helix family member e41	X			Erythrocytosis/polycythemia
BPGM	Bisphosphoglycerate mutase	X	X		Erythrocytosis, familial, 8
CYB5A	Cytochrome b5 type A	X		X	Methemoglobinemia, type IV
CYB5R3	Cytochrome b5 reductase 3	X		X	Methemoglobinemia, type I and type II
EGLN1 (PHD2)	Egl-9 family hypoxia inducible factor 1	X	X		Erythrocytosis, familial, 3
EGLN2	Egl-9 family hypoxia inducible factor 2	X			Erythrocytosis/polycythemia
EGLN3	Egl-9 family hypoxia inducible factor 3	X			Erythrocytosis/polycythemia
EPAS1 (HIF2A)	Endothelial PAS domain protein 1	X	X		Erythrocytosis, familial, 4
EPO	Erythropoietin	X			Erythrocytosis, familial, 5
EPOR	Erythropoietin receptor	X	X		Erythrocytosis, familial, 1
GFI1B	Growth factor independent 1B transcriptional repressor	X			Erythrocytosis; bleeding disorder, platelet-type, 17
HIF1A	Hypoxia inducible factor 1 subunit alpha	X			Erythrocytosis/polycythemia
HIF1AN	Hypoxia inducible factor 1 subunit alpha inhibitor	X			Erythrocytosis/polycythemia
HIF3A	Hypoxia inducible factor 3 subunit alpha	X			Erythrocytosis/polycythemia
JAK2	Janus kinase 2	X			Erythrocytosis, somatic; thrombocythemia 3
KDM6A	Lysine demethylase 6A	X			Erythrocytosis/polycythemia; Kabuki syndrome 2
PFKM	Phosphofructokinase	X			Phosphofructokinase (PFK) deficiency (glycogen storage disease VII/Tarui disease)
PIEZO1	PIEZO ion channel component 1	X			Dehydrated hereditary stomatocytosis/ hereditary xerocytosis, perinatal edema
PKLR	Pyruvate kinase	X			Pyruvate kinase (PK) deficiency
SH2B3	SH2B adaptor protein 3	X			Erythrocytosis, somatic; thrombocythemia, somatic
SOCS3	Suppressor of cytokine signaling 3	X			Erythrocytosis/polycythemia
VHL	von Hippel-Lindau tumor suppressor	X	X		Erythrocytosis, familial, 2; pheochromocytoma; von Hippel-Lindau syndrome