MAYO CLINIC LABORATORIES

Erythrocytosis Genotyping Comparison Chart

Gene		**Comprehensive hereditary erythrocytosis NGS panel**	**Focused hereditary erythrocytosis NGS panel** (more commonly involved genes)	**Congenital methemoglobinemia NGS panel**	**Focused hereditary erythrocytosis Sanger sequencing panel** (more commonly involved genes/exons)	**Erythrocytosis protein studies including assessment for high oxygen hemoglobin variants with molecular reflex to HEMP**	**Associated Disorder**
		NHEP	**NHEM**	**NCYB**	**HEMP**	**REVE2**	
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		X	Reflex	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		X	Reflex	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		Exon 9 and 12	Reflex exon 9 and 12	Erythrocytosis, familial, 4
EPO	Erythropoietin	X					Erythrocytosis, familial, 5
EPOR	Erythropoietin receptor	X	X		Exon 8	Reflex exon 8	Erythrocytosis, familial, 1
GFI1B	Growth factor independent 1B transcriptional repressor	X					Erythrocytosis; bleeding disorder, platelet-type, 17
HBA1/HBA2	Alpha globin genes					Reflex	
HBB	Beta globin					Reflex	

Erythrocytosis Genotyping Comparison Chart (continued)

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Hemoglobin electrophoresis including mass spectrometry						X	
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		X	Reflex	Erythrocytosis, familial, 2; pheochromocytoma; von Hippel-Lindau syndrome