



Gene		NHEP	NHEM	NCYB	Associated Disorder
<i>AC01 (IRP1)</i>	Aconitase 1	X			Erythrocytosis/polycythemia
<i>ANKRD26</i>	Ankyrin repeat domain 26	X			Autosomal dominant thrombocytopenia 2
<i>BHLHE41</i>	Basic helix-loop-helix family member e41	X			Erythrocytosis/polycythemia
<i>BPGM</i>	Bisphosphoglycerate mutase	X	X		Erythrocytosis, familial, 8
<i>CYB5A</i>	Cytochrome b5 type A	X		X	Methemoglobinemia, type IV
<i>CYB5R3</i>	Cytochrome b5 reductase 3	X		X	Methemoglobinemia, type I and type II
<i>EGLN1 (PHD2)</i>	Egl-9 family hypoxia inducible factor 1	X	X		Erythrocytosis, familial, 3
<i>EGLN2</i>	Egl-9 family hypoxia inducible factor 2	X			Erythrocytosis/polycythemia
<i>EGLN3</i>	Egl-9 family hypoxia inducible factor 3	X			Erythrocytosis/polycythemia
<i>EPAS1 (HIF2A)</i>	Endothelial PAS domain protein 1	X	X		Erythrocytosis, familial, 4
<i>EPO</i>	Erythropoietin	X			Erythrocytosis, familial, 5
<i>EPOR</i>	Erythropoietin receptor	X	X		Erythrocytosis, familial, 1
<i>GFI1B</i>	Growth factor independent 1B transcriptional repressor	X			Erythrocytosis; bleeding disorder, platelet-type, 17
<i>HIF1A</i>	Hypoxia inducible factor 1 subunit alpha	X			Erythrocytosis/polycythemia
<i>HIF1AN</i>	Hypoxia inducible factor 1 subunit alpha inhibitor	X			Erythrocytosis/polycythemia
<i>HIF3A</i>	Hypoxia inducible factor 3 subunit alpha	X			Erythrocytosis/polycythemia
<i>JAK2</i>	Janus kinase 2	X			Erythrocytosis, somatic; thrombocythemia 3
<i>KDM6A</i>	Lysine demethylase 6A	X			Erythrocytosis/polycythemia; Kabuki syndrome 2
<i>PFKM</i>	Phosphofructokinase	X			Phosphofructokinase (PFK) deficiency (glycogen storage disease VII/Tarui disease)
<i>PIEZO1</i>	PIEZO ion channel component 1	X			Dehydrated hereditary stomatocytosis/ hereditary xerocytosis, perinatal edema
<i>PKLR</i>	Pyruvate kinase	X			Pyruvate kinase (PK) deficiency
<i>SH2B3</i>	SH2B adaptor protein 3	X			Erythrocytosis, somatic; thrombocythemia, somatic
<i>SOCS3</i>	Suppressor of cytokine signaling 3	X			Erythrocytosis/polycythemia
<i>VHL</i>	von Hippel-Lindau tumor suppressor	X	X		Erythrocytosis, familial, 2; pheochromocytoma; von Hippel-Lindau syndrome