



Red Blood Cell (RBC) Disorder Evaluation Profiles at Mayo Clinic

Profiles	Test Description	Useful For
Protein/Functional Testing Panels		
EEEE1	Red Blood Cell Enzyme Evaluation	Comparative RBC Enzymatic activities/function
HAEV1	Hemolytic Anemia Evaluation	Protein/functional testing for Coombs negative hereditary hemolytic anemia, neonatal anemia
HBEL1	Hemoglobin Electrophoresis Evaluation	Diagnostic hemoglobin testing – General
HGBCE	Hemoglobin Variant, A2 and F Quantitation	Monitoring of previously confirmed Hb variant, Hb A ₂ , Hb A, and Hb F percentages (not diagnostic)
MEV1	Methemoglobinemia Evaluation	Cyanosis, causes of methemoglobin/sulfhemoglobinemia
RBCME	Red Blood Cell Membrane Evaluation	Hereditary spherocytosis, hereditary pyropoikilocytosis
REVE1	Erythrocytosis Evaluation	Longstanding <i>JAK2</i> negative erythrocytosis (elevated Hgb/Hct)
THEV1	Thalassemia and Hemoglobinopathy Evaluation	Complex diagnostic hemoglobin testing with comprehensive diagnosis
Molecular Testing Panels		
NGHHA	Hereditary Hemolytic Anemia Comprehensive Sequencing	Comprehensive genotyping for hemolytic anemia, neonatal anemia
NGMEM	Red Blood Cell Membrane Sequencing	Focused genotyping of presumed/known RBC membrane disorder
NGENZ	Red Blood Cell Enzyme Sequencing	Focused genotyping of presumed/known RBC enzyme disorder
NGCDA	Congenital Dyserythropoietic Anemia Sequencing	Focused genotyping of presumed/known CDA disorder

Available as
a stand-alone
(Test ID)

Test Description	Test IDs	HBEL1	THEV1	HAEV1	REVE1	MEV1	RBCME	EEEE1	Available as a stand-alone (Test ID)
Capillary Electrophoresis	HGBCE	✓	✓	✓	✓	✓			Y
Cation exchange HPLC	HPLC	✓	✓	✓	✓	✓			
Mass Spectrometry, Intact Hb variant	MASS	R	R	R	✓	R			
Isoelectric Focusing (IEF)	IEF	R	R	R	R	R			
Sickle Solubility	SDEX	R	R	R	R	R			Y
Hb Stability (Heat and Isopropanol)	UNHB	R	R	✓	R	R			
Hb F RBC distribution, flow cytometry	HPFH	R	R	R	R	R			
Serum ferritin	FERR		✓*						Y
PB Smear Morphology Review	PBSM/SMPB			✓**			✓**		
Osmotic Fragility, Incubated	FRAG			✓***			✓***		Y
EMA binding/band3, flow cytometry	BND3			✓***			✓***		
G6PD enzyme activity	G6PDC			✓				✓	Y (G6PD1)
PK enzyme activity	PKC			✓				✓	Y (PK1)
Glucose phosphate isomerase activity	GPIC			✓				✓	Y (GPI1)
Hexokinase activity	HKC			✓				✓	Y (HK1)
Adenylate Kinase activity	AKC			✓				✓	Y (AK1)
Phosphofructokinase activity	PFKC			✓				✓	Y (PFK1)
Phosphoglycerate Kinase activity	PGKC			✓				✓	Y (PGK1)
Triosephosphate Isomerase activity	TPIC			✓				✓	Y (TPI1)
Pyrimidine 5' Nucleotidase activity	P5NT			✓				✓	Y
Reduced Glutathione level	GSH			✓				✓	Y (GSH)
Oxygen dissociation (p50), spectrophotometry	P50P				✓***				Y (P50B)
Methemoglobin, spectrophotometry	METH					✓			Y (MET)
Sulfhemoglobin, spectrophotometry	SULF					✓			Y (MET)
Cytochrome b5 reductase (cb5r), spectrophotometry	METR1					✓			Y
DNA seq, alpha globin (<i>HBA1</i> , <i>HBA2</i>)	WASQR	R	R	R	R	R			Y (WASEQ)
MLPA alpha globin cluster, alpha del/dup	ATHAL	R	R	R	R	R			Y
DNA seq, beta globin (<i>HBB</i>)	WBSQR	R	R	R	R	R			Y (WBSEQ)
MLPA beta globin cluster, beta del/dup	WBDDR	R	R	R	R	R			Y (WBDD)
DNA seq, gamma globin (<i>HBG1</i> , <i>HBG2</i>)	WGSQR	R	R	R	R	R			Y (WGSEQ)

Benign Hematology Evaluation Comparison (continued)

Test Description	Test IDs	HBEL1	THEV1	HAEV1	REV1	MEV1	RBCME	EEEV1	Available as a stand-alone (Test ID)
DNA seq, hereditary erythrocytosis (<i>EPOR, HIF2a, PHD2</i>)	HEMP				R				Y
DNA seq, 2,3-BPG mutase (<i>BPGM</i>)	BPGMM				R				Y
DNA seq, von Hippel Lindau (<i>VHL</i>)	VHLE				R				Y
DNA seq and del/dup, pyruvate kinase (<i>PKLR</i>)	PKLR			S				S	Y
DNA seq, glucose-6-phosphate dehydrogenase (<i>G6PD</i>)	G6PDB			S				S	Y
KCNN4 testing	KCNN4								Y
Hereditary Hemolytic Anemia seq panel	NGHHA			S			S	S	Y
RBC Membrane seq panel	NGMEM			S			S		Y
RBC Enzyme seq panel	NGENZ			S				S	Y
Congenital dyserythropoietic anemia seq panel	NGCDA			S					Y

✓ = always performed

R = possible reflex

* = performed if serum sample received

Y = Yes (available)

S = separate order required

** = performed if smear received

*** = Performed if normal shipping control received