

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

Profiles	Test Description	Useful For
Protein/Functional Testing Panels		
EEEV1	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 A2, 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
REVE2	Erythrocytosis Evaluation	Longstanding JAK2 negative erythrocytosis (elevated Hgb/Hct)
THEV1	Thalassemia and Hemoglobinopathy Evaluation	Complex diagnostic hemoglobin testing with comprehensive diagnosis
Molecular Testing Panels		
NHHA	Hereditary Hemolytic Anemia Gene Panel	Comprehensive genotyping for hemolytic anemia, neonatal anemia
NMEM	Red Blood Cell Membrane Disorders Gene Panel	Focused genotyping of presumed/known RBC membrane disorder
NENZ	Red Blood Cell Enzyme Disorders Gene Panel	Focused genotyping of presumed/known RBC enzyme disorder
NCDA	Congenital Dyserythropoietic Anemia Gene Panel	Focused genotyping of presumed/known CDA disorder
NHEP	Hereditary Erythrocytosis Full Gene Panel	Comprehensive genotyping for genes commonly associated with hereditary erythrocytosis and select erythropoiesis genes
NHEM	Hereditary Erythrocytosis Focused Gene Panel	Focused genotyping of genes commonly associated with hereditary erythrocytosis
NCYB	CYB5 and CYB5 Reductase Genetic Analysis	Focused genotyping of presumed/known recessive congenital methemoglobinemia

Test Description	Test IDs	HBEL1	THEV1	HAEV1	REVE2	MEV1	RBCME	EEEV1	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	FERR1		✓*						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)

Benign Hematology Evaluation Comparison (continued)

Test Description	Test IDs	HBEL1	THEV1	HAEV1	REVE2	MEV1	RBCME	EEEV1	Available as a stand-alone (Test ID)
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	WAGDR	R	R	R	R	R			Y (AGDD)
DNA seq, beta globin (<i>HBB</i>)	WBSQR	R	R	R	R	R			Y (WBSEQ)
MLPA beta globin cluster, beta del/dup	WBGDR	R	R	R	R	R			Y (WBGDD)
DNA seq, gamma globin (<i>HBG1</i> , <i>HBG2</i>)	WGSQR	R	R	R	R	R			Y (WGSEQ)
DNA seq, hereditary erythrocytosis (EPOR, HIF2a, PHD2 focused regions)	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>)	PKLRZ			S				S	Y
DNA seq, glucose-6-phosphate dehydrogenase (<i>G6PD</i>)	G6PDZ			S				S	Y
Hereditary Hemolytic Anemia NGS panel	NHHA			S			S	S	Y
RBC Membrane Disorders NGS panel	NMEM			S			S		Y
RBC Enzyme Disorders NGS panel	NENZ			S				S	Y
Congenital dyserythropoietic anemia NGS panel	NCDA			S					Y
Erythrocytosis Full NGS panel	NHEP				S				Y
Erythrocytosis Focused NGS panel	NHEM				S				Y
Recessive Congenital Methemoglobinemia (CYB5, CYB5 Reductase) NGS	NCYB					S			Y

✓ = always performed

NGS = next-generation sequencing

* = performed if serum sample received

Y = Yes (available)

R = possible reflex

** = performed if smear received

S = separate order required

*** = performed if normal shipping control received