

OPTIONS FOR THALASSEMIA DIAGNOSTICS: LABORATORIES AND ASSAYS*

Diagnostic Laboratory	Genetic Test	ID ¹	α-Thal	β-Thal	HbS (SCD)	DNA analysis	Additional comments
ARUP Laboratories	Hemoglobin Evaluation Reflexive Cascade	2005792	Yes	Yes	Yes	Yes ²	Multiple tests to identify hemoglobinopathy ² molecular genetics testing is included only if genetic disorder is suspected
	β-Globin (HBB) Sequencing	3004547	No	Yes	Yes	Yes	NGS
	α-Globin (HBA1 and HBA2) Sequencing and Deletion/Duplication	2011708	Yes	No	No	Yes	MLPA, PCR, Sequencing
Cincinnati Children's	α- (HBA1/2) and β- (HBB) Globin Gene Locus Analysis	5389004	Yes	Yes	Yes	Yes	MLPA and Sanger sequencing
Mayo Clinic Laboratories	Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum	THEV1	Yes	Yes	Yes	No ³	Hb electrophoresis, HPLC, ferritin ³ Additional molecular tests may be suggested to confirm genetic mutations for thalassemia and SCD interpretation that summarizes the testing performed as well as any pertinent clinical information
	β-Globin Gene Sequencing, Varies	WBSEQ	No	Yes	Yes	Yes	PCR/Sanger sequencing, performed after electrophoresis indicated beta-thalassemia
	β-Globin Gene Cluster, Deletion/Duplication, Varies	WBGDD	No	Yes	No	Yes	PCR/MLPA: This test can be used to identify a variety of conditions that involve large deletions of the beta-globin gene cluster, which will not be detected by DNA sequencing of the beta-globin gene

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<u>Quest Diagnostics</u>	α-Globin Gene Sequencing, Varies ⁴	WASEQ	Yes	No	No	Yes	PCR/Sanger sequencing performed after electrophoresis. ⁴ Can be ordered as a reflex testing only
	α-Globin Gene Sequencing, Blood ⁴	WASQR	Yes	No	No	Yes	PCR/Sanger sequencing: testing for non-deletional alpha thalassemia. ⁴ Can be ordered as a reflex testing only
	α-Globin Cluster Locus Deletion/Duplication, Blood	WAGDR	Yes	No	No	Yes	PCR/MLPA: this test is for genetic deletions and duplications only
	Thalassemia and Hemoglobinopathy Comprehensive Evaluation	17365	Yes	Yes	Yes	No ⁵	Isoelectric focusing, HPLC, electrophoresis. ⁵ Molecular tests are added at an additional charge, to further characterize thalassemia
	α-Globin Gene Sequencing	16116	Yes	No	No	Yes	DNA sequencing/PCR
	α-Globin Gene Deletion or Duplication	16124	Yes	No	No	Yes	Capillary electrophoresis and multiplex PCR
	α-Globin Common Mutation Analysis	11175	Yes	No	No	Yes	PCR
	β-Globin Complete™	14974	No	Yes	No	Yes	DNA sequencing, PCR
	α-Thalassemia Panel	6090	Yes	No	No	Yes	MLPA and Sanger sequencing
<u>Prevention Genetics</u>	β-Thalassemia and Hemoglobinopathy via the HBB Gene	7693	No	Yes	Yes	Yes	NGS

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Revvity	Sickle Cell & other Hemoglobinopathies	B0020	No	Yes	Yes	No	Isoelectric focusing
Labcorp	α-Thalassemia Analysis	484278	Yes	No	No	Yes	MLPA and PCR, additional testing available
	β-Thalassemia: HBB (Full Gene Sequencing)	252823	No	Yes	No	Yes	DNA sequencing. In cases in which a known mutation can be documented, the physician may prefer to order β-Thalassemia: HBB (Known Mutation) [252827]
	β-Thalassemia: HBB (Known Mutation)	252827	No	Yes	No	Yes	DNA sequencing. This test looks for presence or absence of familial mutations. The number of amplicons tested depends on the number and location of the familial mutations

1- ID Code refers to an identification number or a reference number of respective assay.

Abbreviations: Hb hemoglobin; DNA deoxyribonucleic acid; PCR polymerase chain reaction; GAP-PCR Gap single polymerase chain reaction; MLPA Multiplex Ligation-dependent Probe Amplification; NGS – next-generation sequencing; HPLC – high-performance liquid chromatography; SCD – sickle cell disease; Thal - thalassemia

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