

Notification Date: November 19, 2024 Effective Date: December 19, 2024

Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum

Test ID: THEV1

Explanation: On the effective date, ATHAL will be replaced by AGDD for standalone testing and WBDD will be replaced with WBGDD for standalone testing. Alpha-Globin Gene Analysis and Beta Globin Cluster Locus Deletion/Duplication ordered through a blood-only profile (THEV1, REVE2, MEV1, HAEV1, HBEL1, HBELP) will use profile-specific codes, WAGDR and WBGDR.

Current Reflex Tests				
Test ID	Reporting Name	Available Separately	Always performed	
HPFH	Hb F Distribution, B	No	No	
SDEX	Sickle Solubility, B	Yes	No	
IEF	Isoelectric Focusing, B	No	No	
UNHB	Hb Stability, B	No	No	
MASS	Hb Variant by Mass Spec, B	No	No	
ATHAL	Alpha-Globin Gene Analysis	Yes	No	
WASQR	Alpha Globin Gene Sequencing, B	Yes (Order WASEQ)	No	
WBSQR	Beta Globin Gene Sequencing, B	Yes (Order WBSEQ)	No	
WBDDR	Beta Globin Cluster Locus Del/Dup,B	Yes (Order WBDD)	No	
WGSQR	Gamma Globin Full Gene Sequencing	Yes (Order WGSEQ)	No	
THEV0	Thalassemia Summary Interpretation	No	No	

New Reflex Tests				
Test ID	Reporting Name	Available Separately	Always performed	
HPFH	Hb F Distribution, B	No	No	
SDEX	Sickle Solubility, B	Yes	No	
IEF	Isoelectric Focusing, B	No	No	
UNHB	Hb Stability, B	No	No	
MASS	Hb Variant by Mass Spec, B	No	No	
WAGDR	Alpha Globin Clustr Locus Del/Dup,B	Yes (Order AGDD)	No	
WASQR	Alpha Globin Gene Sequencing, B	Yes (Order WASEQ)	No	
WBSQR	Beta Globin Gene Sequencing, B	Yes (Order WBSEQ)	No	
WBGDR	Beta Globin Gene Cluster, Del/Dup,B	Yes (Order WBGDD)	No	
WGSQR	Gamma Globin Full Gene Sequencing	Yes (Order WGSEQ)	No	
THEV0	Thalassemia Summary Interpretation	No	No	

Current Test Algorithm

An additional consultative interpretation that summarizes all testing will be provided after test completion to incorporate subsequent results into overall evaluation if any of the following molecular tests are reflexed on this test.

- -ATHAL / Alpha-Globin Gene Analysis, Varies
- -WASQR / Alpha-Globin Gene Sequencing, Blood
- -WBSQR / Beta-Globin Gene Sequencing, Blood
- -WBDDR / Beta-Globin Cluster Locus

Deletion/Duplication, Blood

-WGSQR / Gamma-Globin Full Gene Sequencing, Varies

The results of the individual protein and molecular tests will be released as they are completed; with a final summary interpretation report correlating all performed testing with any clinical information or complete blood cell count results received.

New Testing Algorithm

An additional consultative interpretation that summarizes all testing will be provided after test completion to incorporate subsequent results into overall evaluation if any of the following molecular tests are reflexed on this test.

- -WAGDR / Alpha Globin Cluster Locus Deletion/Duplication, Blood
- -WASQR / Alpha-Globin Gene Sequencing, Blood
- -WBSQR / Beta-Globin Gene Sequencing, Blood
- -WBGDR / Beta-Globin Gene Cluster Deletion/Duplication, Blood
- -WGSQR / Gamma-Globin Full Gene Sequencing, Varies

The results of the individual protein and molecular tests will be released as they are completed; with a final summary interpretation report correlating all performed testing with any clinical information or complete blood cell count results received.