

Test Bulletin

ACL Adds *6 Allele Detection to UGT1A1 Genotyping (Test Order Code LAB12277)

Effective Tuesday, March 18, 2025, ACL Laboratories has added *6 (p.G71R) allele detection to the current testing for the UGT1A1 Genotyping test (Test Order Code LAB12277).

Clinical Indication: This test is designed to detect variants in the UGT1A1 gene, which may cause decreased production or function of the UDP-glucuronosyl transferase enzyme, responsible for bilirubin metabolism, leading to hyperbilirubinemia. The severity of the functional deficiency of hepatic UDP-glucuronosyl transferase is determined by the genetic variant(s); thus, the resulting phenotypic spectrum is variable.

Test Method: This test will be performed by ACL Laboratories using a laboratory developed test method based on PCR and fluorescent fragment size analysis.

Specimen Requirements: One pink (K2EDTA) 6.0 mL OR Two lavender (K2EDTA) 3.0 mL OR One ORAcollect® Dx Buccal Swab kit

Patient preparation for ORAcollect® Dx Buccal Swab kit - Collect One buccal swab using the ORAcollect® collection kit ensuring the sponge tip does not come into contact with any surface prior to collection. Patient should not eat, drink, smoke, or chew gum for 30 minutes before collecting oral sample.

Buccal swab will be orderable for clients using Workday: Item # 3029385 Container Collection Liquid Sample Painless Noninvasive Bacteriostatic ORAcollect[®].

Transport: 5.0 mL (minimum 1.0 mL) whole blood, refrigerated

Stability: Ambient: 3 days whole blood, 1 week ORAcollect[®] swab; Refrigerated: 2 weeks whole blood, 2 weeks ORAcollect[®] swab; Frozen: Unacceptable

Unacceptable Conditions: Plasma, serum, FFPE tissue blocks/slides, decalcified or frozen tissue. Specimens collected in anticoagulants other than K2EDTA. Clotted or grossly hemolyzed specimens.

Performed: Weekdays

Performing Sites: ACL Illinois Central Laboratory

Reporting Time: Final within 7 days

This new ACL assay detects the same variants as the previous assay, with the addition of detection of the *6 (p.G71R) allele. Based on the combination of UGT1A1 alleles present, there are three possible results: Normal Metabolizer, Intermediate Metabolizer, and Poor Metabolizer.

This test analyzes the *1, *6, *28, *36, and *37 UGT1A1 alleles. Assigning an allele a *1 or "normal" status is based upon negative results for the *6, *28, *36, and *37 alleles only. Other UGT1A1 alleles are not detected. Genetic and environmental factors other than UGT1A1 status may contribute to drug toxicity and efficacy.

If you have any questions, please contact:

ACL Molecular Pathology Laboratory at Rosemont (ph. 847.349.7182), or ACL Client Services (ph. 800.877.7016)

For additional information regarding these tests, as well as specimen collection requirements, please contact ACL Client Services at 1.800.877.7016 or visit our website at <u>https://www.acllaboratories.com/providers/test-directory</u>/.

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Referrals Update

Allergen: Sole IgE

Allergen: Sole IgE (Test Order Code LAB9298) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday, March 18, 2025.** Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Basil IgE

Allergen: Basil IgE (Test Order Code LAB9113) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday, March 18, 2025**. Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Goldenrod IgE

Allergen: Goldenrod IgE (Test Order Code LAB9190) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday**, **March 18**, **2025**. Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Malt IgE

Allergen: Malt IgE (Test Order Code LAB9224) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday, March 18, 2025.** Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Buckwheat IgE

Allergen: Buckwheat IgE (Test Order Code LAB9128) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday**, **March 18**, **2025**. Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Cheese Mold IgE

Allergen: Cheese Mold IgE (Test Order Code LAB9143) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday**, **March 18**, **2025**. Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

Allergen: Parsley IgE

Allergen: Parsley IgE (Test Order Code LAB9249) which was previously sent to Cleveland Clinic Laboratory, has been inactivated **effective Tuesday**, **March 18**, **2025**. Providers will need to utilize a Miscellaneous test code to order testing to be sent to ARUP.

New Orderable Code Beta-Amyloid 42/40 Ratio, Plasma

Effective Tuesday, March 18, 2025, Beta-Amyloid 42/40 Ratio, Plasma (Test Order Code LAB12921) is available as an orderable test with testing being performed at Quest Laboratories. Providers will no longer have to utilize a Miscellaneous test code for ordering. Test information is below.

Test Information	Beta-Amyloid 42/40 Ratio, Plasma (Test Order Code LAB12921)	
Specimen Requirement	1.2 mL (minimum 0.6 mL) plasma	
Collection Tube	Lavender	
Temperature	Frozen	
Stability	32 days	
Methodology	Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)	
TAT	Final within 7 days	
Performing Lab	Quest	

For additional information regarding these tests, as well as specimen collection requirements, please contact ACL Client Services at 1.800.877.7016 or visit our website at https://www.acllaboratories.com/providers/test-directory/.

New Orderable Code Neurofilament Light Chain (NfL), Plasma

Effective Tuesday, March 18, 2025, Neurofilament Light Chain (NfL), Plasma (Test Order Code LAB12923) is available as an orderable test with testing being performed at Quest Laboratories. Providers will no longer have to utilize a Miscellaneous test code for ordering. Test information is below.

Test Information	Neurofilament Light Chain (NfL), Plasma (Test Order Code LAB12923)	
Specimen Requirement	0.7 mL (minimum 0.5 mL) plasma	
Collection Tube	Lavender	
Temperature	Ambient	
Stability	35 days	
Methodology	Immunoassay (IA)	
TAT	Final within 7 days	
Performing Lab	Quest	

New Orderable Code Phosphorylated tau181(p-tau181), Plasma

Effective Tuesday, March 18, 2025, Phosphorylated tau181(p-tau181), Plasma (Test Order Code LAB12924) is available as an orderable test with testing being performed at Quest Laboratories. Providers will no longer have to utilize a Miscellaneous test code for ordering. Test information is below.

Test Information	Phosphorylated tau181(p-tau181), Plasma (Test Order Code LAB12924)	
Specimen Requirement	1.0 mL (minimum 0.5 mL) plasma	
Collection Tube	Lavender	
Temperature	Ambient	
Stability	14 days	
Methodology	Electrochemiluminescence (ECLIA)	
TAT	Final within 7 days	
Performing Lab	Quest	

New Orderable Code Chromatin Antibody IgG

Effective Tuesday, March 18, 2025, Chromatin Antibody IgG (Test Order Code LAB12902) is available as an orderable test with testing being performed at ARUP Laboratories. Testing was previously performed by ACL. Test information is below.

Test Information	Chromatin Antibody IgG (Test Order Code LAB12902)	
Specimen Requirement	1.0 mL (minimum 0.5 mL) serum	
Collection Tube	Gold Gel	
Temperature	Refrigerated	
Stability	2 weeks	
Methodology	Semi-Quantitative Enzyme-Linked Immunosorbent Assay	
TAT	Final within 6 days	
Performing Lab	ARUP	

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Updated Referral Testing Orderable Codes

Effective Tuesday, March 18, 2025, the following send-out assays were updated.

Allergen Duck Feathers IgE		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Allergen: Duck Feathers, IgE	Allergen, Animal, Duck Feathers IgE
Test Order Code	LAB9170	LAB12904
Performing Lab	CCL	ARUP
Specimen Type	0.5 mL serum	1.0 mL (minimum 0.5 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	30 days	2 weeks
Methodology	Fluorescent Enzyme Immunoassay (FEIA) by ImmunoCAP	Quantitative ImmunoCAP Fluorescent Enzyme Immunoassay
ТАТ	3 days	Final within 5 days
	Blastomyces Antibody, Immun	odiffusioo
	Current	
	(Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Blastomyces Antibody, Immunodiffusion	Blastomyces Dermatitidis Antibodies by Immunodiffusion
Test Order Code	LAB9393	LAB11014
Performing Lab	CCL	ARUP
Specimen Type	Serum	0.5 mL (minimum 0.15 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	1 week	2 weeks
Methodology	Immunodiffusion	Immunodiffusion
TAT	Final within 9 days	Final within 8 days
	HLA B5701	
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	HLA B5701 Genotype	HLA- B* 57:01 for Abacavir Sensitivity
Test Order Code	LAB9596	LAB12617
Performing Lab	CCL	ARUP
Specimen Type	Whole blood	Whole blood
Collection Tube	Lavender	Lavender
Temperature	Ambient	Refrigerated
Stability	1 week	1 week
Methodology	Polymerase Chain Reaction (PCR)	Polymerase Chain Reaction/Fluorescence Monitoring
TAT	Final within 10 days	Final within 12 days
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Fluconazole		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Fluconazole	Fluconazole, Quantitative by LC-MS/MS
Test Order Code	LAB9536	LAB12618
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.6 mL) serum
Collection Tube	Plain Red	Plain Red
Temperature	Refrigerated	Frozen
Stability	15 days	6 months
Methodology	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)	Quantitative High Performance Liquid Chromatography-Tandem Mass Spectrometry
TAT	Final within 6 days	Final within 8 days

Copper, Random Urine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Copper, Random Urine	Copper, Random Urine
Test Order Code	LAB9474	LAB12619
Performing Lab	CCL	ARUP
Specimen Type	Urine	8.0 mL (minimum 1.0 mL) Urine
Collection Tube	Sterile container	Sterile Container
Temperature	Ambient	Refrigerated
Stability	30 days	2 weeks
Methodology	Inductively Coupled Plasma / Mass Spectrometry (ICP-MS)	Inductively Coupled Plasma / Mass Spectrometry (ICP-MS)
TAT	Final within 9 days	Final within 7 days

Phenylalanine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Phenylalanine	Phenylalanine Monitoring, Plasma
Test Order Code	LAB9758	LAB12620
Performing Lab	CCL	ARUP
Specimen Type	Plasma	0.5mL (minimum 0.25 mL) plasma
Collection Tube	Green sodium heparin	Green lithium heparin, no gel
Temperature	Frozen	Frozen
Stability	2 weeks	1 month
Methodology	lon Exchange Chromatography	Liquid Chromatography-Tandem Mass Spectrometry
TAT	Final within 12 days	Final within 7 days

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Phenylalanine and Tyrosine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Phenylalanine and Tyrosine	Phenylalanine and Tyrosine
Test Order Code	LAB9759	LAB12621
Performing Lab	CCL	ARUP
Specimen Type	Plasma	0.5 mL (minimum 0.25 mL) plasma
Collection Tube	Green sodium heparin	Green lithium heparin, no gel
Temperature	Frozen	Frozen
Stability	2 weeks	1 month
Methodology	Ion Exchange Chromatography	Quantitative Liquid Chromatography-Tandem Mass Spectrometry
ТАТ	Final within 12 days	Final within 7 days

Manganese, Blood		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Manganese, Blood	Manganese, Whole Blood
Test Order Code	LAB9665	LAB12622
Performing Lab	CCL	ARUP
Specimen Type	Whole blood	6.0 mL (minimum 0.5ml) Whole blood
Collection Tube	Royal Blue EDTA	Royal Blue K2EDTA
Temperature	Refrigerated	Ambient
Stability	1 month	Indefinitely
Methodology	Inductively Coupled Plasma / Mass Spectrometry (ICP-MS)	Quantitative Inductively Coupled Plasma-Mass Spectrometry (ICP-MS)
ТАТ	Final within 10 days	Final within 5 days

Oxalate, 24 Hour Urine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Oxalate, 24 Hour Urine	Oxalate, 24 Hour Urine
Test Order Code	LAB9735	LAB12623
Performing Lab	CCL	ARUP
Specimen Type	Urine	4.0 mL (minimum 1.5 mL) Urine
Collection Tube	24-hour urine container	24-hour urine container
Temperature	Refrigerated	Frozen
Stability	1 week	1 month
Methodology	Enzyme Immunoassay	Quantitative Spectrophotometry
ТАТ	Final within 6 days	Final within 6 days

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Aldosterone, 24 Hour Urine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Aldosterone, 24 Hour Urine	Aldosterone, 24 Hour Urine
Test Order Code	LAB9097	LAB12625
Performing Lab	CCL	ARUP
Specimen Type	Urine	4.0 mL (minimum 0.5 mL) Urine
Collection Tube	24-hour urine container	24-hour urine container
Temperature	Refrigerated	Frozen
Stability	1 week	1 month
Methodology	Chemiluminescence Immunoassay (CLIA)	Chemiluminescence Immunoassay (CLIA)
TAT	Final within 9 days	Final within 7 days

Carbamazepine and Metabolite Level		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Carbamazepine and Metabolite Level	Carbamazepine Epoxide and Total
Test Order Code	LAB9423	LAB12626
Performing Lab	CCL	ARUP
Specimen Type	Plasma	1.0 mL (minimum 0.5 mL) serum
Collection Tube	Green lithium heparin no gel	Plain Red
Temperature	Refrigerated	Frozen
Stability	1 week	1 month
Methodology	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)	Quantitative Liquid Chromatography-Tandem Mass Spectrometry
TAT	Final within 6 days	Final within 10 days

Copper, 24-hour urine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Copper, 24 Hour Urine	Copper, Urine
Test Order Code	LAB9473	LAB12627
Performing Lab	CCL	ARUP
Specimen Type	Urine	8.0 mL (minimum 1.0 mL) Urine
Collection Tube	24-hour urine container	24-hour urine container
Temperature	Refrigerated	Refrigerated
Stability	30 days	2 weeks
Methodology	Inductively Coupled Plasma, Mass Spectrometry	Quantitative Inductively Coupled Plasma, Mass Spectrometry
TAT	Final within 10 days	Final within 7 days

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Von Willebrand Factor Multimers		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Von Willebrand Factor Multimer	Von Willebrand Factor Multimers
Test Order Code	LAB9890	LAB12628
Performing Lab	CCL	ARUP
Specimen Type	Plasma	1.0 mL (minimum 0.5 mL) platelet poor plasma
Collection Tube	Light Blue	One white discard tube followed by one light blue
Temperature	Frozen	Frozen
Stability	6 months	3 months
Methodology	Gel Electrophoresis	Qualitative Electrophoresis
ТАТ	Final within 16 days	Final within 13 days

FSH Tanner Stages		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	FSH Tanner Stages	FSH Tanner Stages
Test Order Code	LAB9542	LAB12629
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.3 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	2 weeks	2 weeks
Methodology	Electro Chemiluminescence Immunoassay (ECLIA)	Quantitative Electrochemiluminescence Immunoassay
ТАТ	Final within 4 days	Final within 3 days

Luteinizing Hormone, Tanner Stages		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Luteinizing Hormone, Tanner Stages	Luteinizing Hormone, Tanner Stages
Test Order Code	LAB9659	LAB12630
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.3 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	2 weeks	2 weeks
Methodology	Electro Chemiluminescence Immunoassay (ECLIA)	Quantitative Electrochemiluminescence Immunoassay
TAT	Final within 4 days	Final within 3 days

Posaconazole Level		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Posaconazole Level	Posaconazole Quantitative by LC-MS/MS
Test Order Code	LAB9774	LAB12631
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0mL (minimum 0.6mL) serum
Collection Tube	Plain Red	Plain Red
Temperature	Refrigerated	Frozen
Stability	15 days	6 months
Methodology	Liquid Chromatography, Tandem Mass Spectrometry	Quantitative High Performance Liquid Chromatography-Tandem Mass Spectrometry
TAT	Final within 6 days	Final within 8 days

Itraconazole Level		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Itraconazole Level	Itraconazole Quantitative by LC-MS/MS
Test Order Code	LAB9635	LAB12632
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.6 mL) serum
Collection Tube	Plain Red	Plain Red
Temperature	Refrigerated	Frozen
Stability	15 days	6 months
Methodology	Liquid Chromatography, Tandem Mass Spectrometry	Quantitative High Performance Liquid Chromatography-Tandem Mass Spectrometry
TAT	Final within 6 days	Final within 8 days

Crithidia Lucillae		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Crithidia Lucillae	Double- Stranded DNA (dsDNA) Anitbody, IgG by IFA (using Crithidia Lucillae)
Test Order Code	LAB9482	LAB12633
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0mL (minimum 0.15 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	1 week	2 weeks
Methodology	Indirect Immunofluorescence Assay (IFA)	Semi-Quantitative Indirect Fluorescent Antibody (IFA)
TAT	Final within 9 days	Final within 5 days

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Mycophenolic Acid		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Mycophenolic Acid	Mycophenolic Acid and Metabolites
Test Order Code	LAB9691	LAB12634
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0mL (minimum 0.1mL) serum
Collection Tube	Plain Red	Plain Red
Temperature	Refrigerated	Refrigerated
Stability	4 days	6 weeks
Methodology	Enzymatic	Quantitative Liquid Chromatography-Tandem Mass Spectrometry
ТАТ	Final within 2 days	Final within 6 days

Lacosamide		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Lacosamide	Lacosamide, Serum
Test Order Code	LAB9641	LAB12635
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.5 mL) serum
Collection Tube	Red Top	Plain Red
Temperature	Refrigerated	Refrigerated
Stability	2 weeks	1 week
Methodology	Liquid Chromatography-Tandem Mass Spectrometry	Quantitative Enzyme Immunoassay (EIA)
TAT	Final within 6 days	Final within 4 days

Fructosamine		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Fructosamine	Fructosamine
Test Order Code	LAB9541	LAB12636
Performing Lab	CCL	ARUP
Specimen Type	Serum	0.5 mL (minimum 0.3 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Refrigerated	Refrigerated
Stability	2 weeks	2 weeks
Methodology	Colorimetric	Quantitative Spectrophotometry
TAT	Final within 5 days	Final within 3 days

Vitamin E Level		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	Vitamin E Level	Vitamin E, Serum or Plasma
Test Order Code	LAB9886	LAB12637
Performing Lab	CCL	ARUP
Specimen Type	Plasma	1.0mL (minimum 0.2mL) serum
Collection Tube	Lavender	Gold Gel
Temperature	Frozen	Refrigerated
Stability	1 week	1 month
Methodology	High Performance Liquid Chromatography (HPLC)	Quantitative High Performance Liquid Chromatography (HPLC)
TAT	Final within 8 days	Final within 6 days

John Cunningham Virus Antibody with reflex to Inhibition Assay		
	Current (Deactivated 3.18.2025)	Replacement (Activated 3.18.2025)
Test Name	John Cunningham Virus Antibody, Index Value with reflex to Inhibition Assay	John Cunningham Virus Antibody by Elisa, Serum with Reflex to Inhibition Assay
	Reflex test: John Cunningham Virus Antibody, Inhibition	Reflex test: JC Virus Inhibition Assay, Serum
Test Order Code	LAB9637 & LAB9638	LAB12638 & LAB12639
Performing Lab	CCL	ARUP
Specimen Type	Serum	1.0 mL (minimum 0.5 mL) serum
Collection Tube	Gold Gel	Gold Gel
Temperature	Frozen	Refrigerated
Stability	2 weeks	2 weeks
Methodology	Immunoassay	Enzyme-Linked Immunosorbent Assay (ELISA)
TAT	Final within 7 days	Final within 11 days

For additional information regarding these tests, as well as specimen collection requirements, please contact ACL Client Services at 1.800.877.7016 or visit our website at <u>https://www.acllaboratories.com/providers/test-directory/</u>.

ACL Laboratories Tests for Genotyping (Test Order Code UGT1A1)

Biology of the Process

The UGT1A1 gene encodes the bilirubin UDP-glucuronosyltransferase (UGT) enzyme, which glucuronidates bilirubin, leading to its elimination. Reduced UGT1A1 activity leads to the accumulation of unconjugated bilirubin. Drugs that inhibit UGT1A1 activity can also increase levels of plasma unconjugated bilirubin, increasing the risk of side effects such as jaundice, neutropenia, and diarrhea. Differences in the number of TA repeats in the UGT1A1 promoter affect transcription levels of the gene. The wild type allele (*1) has six TA repeats and has normal transcription levels. Alleles with seven (*28) or eight (*37) repeats result in decreased transcriptional activity, while alleles with five repeats (*36) result in increased transcription levels. The *6 allele (p.G71R), common in individuals of east Asian descent, also reduces enzyme activity. Homozygosity for alleles with decreased activity has been associated with Gilbert syndrome (OMIM #143500), a common benign autosomal recessive disorder characterized by elevated levels of bilirubin in the blood.

Clinical Indication

Testing of UGT1A1 metabolizer status is used in patients undergoing itinotecan-based anticancer therapy to determine if drug dosage should be adjusted to reduce the possibility of side effects such as severe neutropenia and diarrhea. In addition, the Clinical Pharmacogenetics Implementation Consortium (CPIC) recommends UGT1A1 testing in patients undergoing atazanavir-based antiretoviral protease inhibitor therapy.

Limitation

This test analyzes the *1, *6, *28, *36, and *37 UGT1A1 alleles. Assigning an allele a *1 or "normal" status is based upon negative results for the *6, *28, *36, and *37 alleles only. Other UGT1A1 alleles are not detected. Genetic and environmental factors other than UGT1A1 status may contribute to drug toxicity and efficacy.

Testing Method

Laboratory developed test method based on PCR and fluorescent fragment size analysis

Reporting

Based on the combination of UGT1A1 alleles present, there are three possible results: Normal Metabolizer, Intermediate Metabolizer, and Poor Metabolizer.

Normal Range

Normal Metabolizer

References

Pratt VM, Scott SA, Pirmohamed M, et al., editors. Medical Genetics Summaries [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 2012–PharmGKB–UGT1A1 <u>https://www.pharmgkb.org/gene/PA420</u>

UGT1A1 and common exon allele nomenclature <u>https://www.pharmacogenomics.pha.ulaval.ca/wp-content/uploads/2015/04/</u> <u>UGT1A1-allele-nomenclature.html</u>

ACL Implements Changes to Myeloproliferative Neoplasm Panel (LAB12257) Billing

Effective Monday March 24, 2025, ACL Laboratories implemented changes to Myeloproliferative Neoplasms (MPN) (Test Order Code LAB12257) billing.

Providers will need to select the appropriate testing pathway for **Polycythemia Vera (PV)** OR **Essential Thrombocythemia (ET)/Primary Myelofibrosis (PMF)** at the time of order entry. This step is essential to ensure the correct CPT charges are applied and prevents insurance denials. A detailed workflow layout is provided below.

- Primary charge: JAK2 (V617F) Mutation (CPT 81270) will be used for prior authorization workflow
- Polycythemia Vera (PV) pathway: JAK2 (V617F) Mutation (CPT 81270) with reflex to JAK2 Exon 12-15 Mutations Detection (CPT 81279)
- Essential Thrombocythemia (ET)/Primary Myelofibrosis (PMF) pathway: JAK2 (V617F) Mutation (CPT 81270) with reflex to CALR (Calreticulin) Exon 9 Mutations (CPT 81219), and MPL Exon 10 Mutations Detection (CPT 81339)

JAK2 (V617F) Mutation results will include quantitative results (VAF%) when detected. This change is intended to eliminate insurance denials. Previously used 81479 CPT will be discontinued.

If you have any questions or need additional information, please contact ACL Client Services at 800-877-7016.

