MEDICARE COVERAGE OF LABORATORY TESTING

Please remember when ordering laboratory tests that are billed to Medicare/Medicaid or other federally funded programs, the following requirements apply:

- Only tests that are medically necessary for the diagnosis or treatment of the patient should be ordered.
 Medicare does not pay for screening tests except for certain specifically approved procedures and may not pay for non-FDA approved tests or those tests considered experimental.
- If there is reason to believe that Medicare will not pay for a test, the patient should be informed. The patient should then sign an Advance Beneficiary Notice (ABN) to indicate that he or she is responsible for the cost of the test if Medicare denies payment.
- The ordering physician must provide an ICD-10 diagnosis code or narrative description, if required by the fiscal intermediary or carrier.
- Organ- or disease-related panels should be billed only when all components of the panel are medically necessary.
- Both ARUP- and client-customized panels should be billed to Medicare only when every component of the customized panel is medically necessary.
- Medicare National Limitation Amounts for CPT codes are available through the Centers for Medicare & Medicaid Services (CMS) or its intermediaries. Medicaid reimbursement will be equal to or less than the amount of Medicare reimbursement.

The CPT Code(s) for test(s) profiled in this bulletin are for informational purposes only. The codes reflect our interpretation of CPT coding requirements, based upon AMA guidelines published annually. CPT codes are provided only as guidance to assist you in billing. ARUP strongly recommends that clients reconfirm CPT code information with their local intermediary or carrier. CPT coding is the sole responsibility of the billing party.

The regulations described above are only guidelines. Additional procedures may be required by your fiscal intermediary or carrier.

Hotline Page #	Test Number	Summary of Changes by Test Name	Name Change	Methodology	Performed/Reported Schedule	Specimen Requirements	Reference Interval	Interpretive Data	Note	CPT Code	Component Change	Other Interface Change	New Test	Inactive
8	<u>2013725</u>	ABCC8-Related Hyperinsulinism, 3 Variants				X								
8	0051266	Achondroplasia (FGFR3) 2 Mutations				X								
8	0051265	Achondroplasia (FGFR3) 2 Mutations, Fetal				X								
51	<u>2011902</u>	Adrenoleukodystrophy, X- Linked (ABCD1) Sequencing												X
9	<u>2011708</u>	Alpha Globin (<i>HBA1</i> and <i>HBA2</i>) Sequencing and Deletion/Duplication (Extended TAT as of 1/11/21-no referral available)						X						
51	0051495	Alpha Thalassemia (HBA1 and HBA2) 7 Deletions												X



Hotline Page #	Test Number	Summary of Changes by Test Name	Name Change	Methodology	Performed/Reported Schedule	Specimen Requirements	Reference Interval	Interpretive Data	Note	CPT Code	Component Change	Other Interface Change	New Test	Inactive
10	3003651	Alpha Thalassemia (<i>HBA1</i> and <i>HBA2</i>) Deletion/Duplication with reflex to Hb Constant Spring											x	
11	3003656	Alpha Thalassemia (<i>HBA1</i> and <i>HBA2</i>) Deletion/Duplication with reflex to Hb Constant Spring, Fetal											X	
12	0050002	Alpha-1-Acid Glycoprotein				X								
12	0051256	Alpha-1-Antitrypsin (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to Alpha- 1-Antitrypsin Phenotype				х								
51	0050043	Alpha-1-Microglobulin, Urine												x
12	<u>2012209</u>	Amphetamines Urine Screen with Reflex to Quantitation				X								
12	<u>2005419</u>	Androstanediol Glucuronide Quantitative, Serum or Plasma			X	X								
12	2005077	Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR				X								
13	2012232	Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal				X								
13	0050392	Ankylosing Spondylitis (HLA-B27) Genotyping				Х								
13	3000601	Antinuclear Antibody (ANA) with HEp-2 Substrate, IgG by IFA with Reflex by Pattern				х								
13	0030192	APC Resistance Profile with Reflex to Factor V Leiden				х								
13	0055654	Apolipoprotein B (APOB) Mutation Detection				X								
14	2013341	Apolipoprotein E (<i>APOE</i>) Genotyping, Alzheimer Disease Risk				Х								
14	2013337	Apolipoprotein E (<i>APOE</i>) Genotyping, Cardiovascular Risk				X								
51	2011144	Arginine:Glycine Amidinotransferase (GATM) Deficiency Sequencing												X
14	0051415	Ashkenazi Jewish Diseases, 16 Genes				Х								
51	2008471	ATP7A-Related Copper Transport Disorders (ATP7A) Sequencing and Deletion/Duplication												х
14	3000724	B-Lymphoblastic Leukemia (B-ALL) Minimum Residual Disease Detection by Flow Cytometry	х					х	X			х		
15	3002528	Bacterial Strain Typing by Next Generation Sequencing											X	
16	3002582	Bacterial Vaginosis by TMA											х	
16	2012211	Barbiturates, Urine Screen with Reflex to Quantitation				X								



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16	<u>2012225</u>	Benzodiazepines, Urine Screen with Reflex to Quantitation				X								
17	0051433	Bloom Syndrome (BLM), 1 Variant				X								
17	<u>2012273</u>	Buprenorphine, Urine Screen with Reflex to Quantitation				X								
17	0051453	Canavan Disease (ASPA), 4 Variants				X								
18	<u>3002583</u>	Candida glabrata, Candida species, and Trichomonas vaginalis by TMA											x	
19	3003634	Capillary Malformation-Arteriovenous Malformation (CM-AVM) Panel, Sequencing and Deletion/Duplication											X	
19	<u>2012278</u>	Carisoprodol Urine with Reflex to Quantitation				X								
20	<u>3003704</u>	CD71 by Immunohistochemistry											X	
20	2005018	Celiac Disease (<i>HLA-DQ2</i> , and <i>HLA-DQ8</i>) Genotyping				X								
20	<u>2012155</u>	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, <i>PMP22</i> Deletion/Duplication with Reflex to Sequencing Panel				X								
51	2008652	c-MET by Immunohistochemistry												X
20	<u>2012231</u>	Cocaine, Urine Screen with Reflex to Quantitation				X								
21	3002463	Connective Tissue Disease First Line Panel with Reflex					X					x		
22	<u>3003756</u>	Copper, Red Blood Cells											X	
22	0050180	C-Reactive Protein				X								
22	0092572	Cutaneous Direct Immunofluorescence, Biopsy				X								
23	<u>3001508</u>	CYP2C19				X								
23	<u>3001501</u>	CYP2C8 and CYP2C9				X								
23	<u>3001513</u>	CYP2D6				X								
23	<u>3001518</u>	CYP3A4 and CYP3A5				X								
23	<u>2013661</u>	Cystic Fibrosis (CFTR) 165 Pathogenic Variants				X								
23	2013663	Cystic Fibrosis (<i>CFTR</i>) 165 Pathogenic Variants with Reflex to Sequencing				X								
24	2013664	Cystic Fibrosis (<i>CFTR</i>) 165 Pathogenic Variants with Reflex to Sequencing and Reflex to Deletion/Duplication				X								
24	2013662	Cystic Fibrosis (<i>CFTR</i>) 165 Pathogenic Variants, Fetal				X								
24	<u>3001524</u>	Cytochrome P450 Genotyping Panel				Х								
24	0030057	D-Dimer			X	X		X						-



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25	<u>2012166</u>	Dihydropyrimidine Dehydrogenase (<i>DPYD</i>), 3 Variants				X								
25	<u>3001581</u>	Dilated Cardiomyopathy Panel, Sequencing											X	1
25	0050215	Double-Stranded DNA (dsDNA) Antibody, IgG by ELISA with Reflex to dsDNA Antibody, IgG by IFA					х					X		
26	0092184	Drug Panel 7, Urine - Screen with Reflex to Confirmation/Quantitation				X								
26	0092185	Drug Panel 7A, Urine - Screen with Reflex to Confirmation/Quantitation				X								
26	0092186	Drug Panel 9, Urine - Screen with Reflex to Confirmation/Quantitation				X						X		
26	0092187	Drug Panel 9A, Urine - Screen with Reflex to Confirmation/Quantitation				X						X		
27	2012312	Drug Profile, Screen with Reflex to Quantitation	X			X	X					X		
27	2007479	Drug Profile, Targeted by Tandem Mass Spectrometry and Enzyme Immunoassay, Urine										X		
27	2009288	Drug Profile, Targeted with Interpretation by Tandem Mass Spectrometry and Enzyme Immunoassay, Urine										X		
27	0090448	Drugs of Abuse 7 Panel, Urine - Screen Only				X								
28	0090449	Drugs of Abuse 7A Panel, Urine - Screen Only				Х								
28	0090453	Drugs of Abuse 9 Panel, Urine - Screen Only				Х								
28	0090454	Drugs of Abuse 9A Panel, Urine - Screen Only				X								
28	0092280	Drugs of Abuse Test, Alcohol, Urine - Screen with Reflex to Confirmation/Quantitation				X								
28	2011241	Duchenne/Becker Muscular Dystrophy (<i>DMD</i>) Deletion/Duplication with Reflex to Sequencing				X								
28	0051463	Dysautonomia, Familial (IKBKAP), 2 Variants				X								į.
51	2005559	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (<i>PLOD1</i>) Sequencing and Deletion/Duplication												X
29	0090518	Ethanol, Urine, Qualitative - Medical				X								
29	<u>2012695</u>	Ethyl Glucuronide Screen Only, Urine				X								1
29	2007912	Ethyl Glucuronide Screen with Reflex to Confirmation, Urine				X								
29	0097720	Factor V Leiden (F5) R506Q Mutation				X								
29	2003220	Factor XIII (F13A1) V34L Variant				X								
30	3002110	Familial Hypercholesterolemia Panel, Sequencing											X	
30	0051468	Fanconi Anemia, Group C (FANCC), 2 Variants				X								
30	<u>2012284</u>	Fentanyl, Urine Screen with Reflex to Quantitation				X	X							



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3003279	Gastrointestinal Pathogens Panel by PCR											X	
0051438	Gaucher Disease (GBA), 8 Variants				X								
<u>3000258</u>	Reflex to Methylation				X								
<u>0051684</u>	Mutations		х		X								
<u>2013740</u>	Glycogen Storage Disease, Type 1A (<i>G6PC</i>), 9 Variants				X								
0055656	Hemochromatosis (HFE) 3 Mutations				X								
0030144	Heparin Anti-Xa, Low Molecular Weight Heparin			X	X								
0030143	Heparin Anti-Xa, Unfractionated			X	X								
2005457	High-Specificity Antiphospholipid Antibodies, IgG and IgM												X
2002429	HLA-B*57:01 for Abacavir Sensitivity				X								
<u>2003020</u>	Human Epididymis Protein 4 (HE4)				X								
3003760	Qualitative NAAT											x	
2014234	Human Immunodeficiency Virus 1 (HIV-1) by Qualitative Transcription-Mediated Amplification (TMA)												X
3001474	Human Immunodeficiency Virus 1 (HIV-1) Qualitative by NAAT, Whole Blood												X
<u>3001579</u>	Hypertrophic Cardiomyopathy Panel, Sequencing											X	
<u>3003751</u>	JAK2 (V617F) Mutation by ddPCR, Quantitative											X	
0040168	JAK2 Gene, V617F Mutation, Quantitative												X
<u>2013909</u>	Joubert Syndrome Type 2 (TMEM216), 1 Variant				X								
<u>2009302</u>	Li-Fraumeni (TP53) Sequencing												X
<u>2013735</u>	Variants				X								
<u>3001603</u>	Long QT Panel, Sequencing and Deletion/Duplication											X	
0092079	Magnesium, RBC					Х	X				X		
2010162	Mammaglobin by Immunohistochemistry				Х								
2013730	Maple Syrup Urine Disease, Type 1B (<i>BCKDHB</i>), 3 Variants				х								
0051205	Medium Chain Acyl-CoA Dehydrogenase (<i>ACADM</i>) 2 Mutations				X								
2012288	Meperidine, Urine Screen with Reflex to Quantitation				х								
	3003279 0051438 3000258 0051684 2013740 0055656 0030144 0030143 2005457 2002429 2003020 3003760 2014234 3001474 3001579 3003751 0040168 2013909 2013735 3001603 0092079 2010162 2013730 0051205	3003279 Gastrointestinal Pathogens Panel by PCR 0051438 Gaucher Disease (GBA), 8 Variants 3000258 Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation 0051684 Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations 2013740 Glycogen Storage Disease, Type 1A (G6PC), 9 Variants 0055656 Hemochromatosis (HFE) 3 Mutations 0030144 Heparin Anti-Xa, Low Molecular Weight Heparin 0030143 Heparin Anti-Xa, Unfractionated 2005457 High-Specificity Antiphospholipid Antibodies, IgG and IgM 2002429 HLA-B*57:01 for Abacavir Sensitivity 2003020 Human Epididymis Protein 4 (HE4) 3003760 Qualitative NAAT Human Immunodeficiency Virus 1 (HIV-1) by Qualitative Transcription-Mediated Amplification (TMA) 3001474 Human Immunodeficiency Virus 1 (HIV-1) Qualitative by NAAT, Whole Blood 3001579 Hypertrophic Cardiomyopathy Panel, Sequencing 3003751 JAK2 (V617F) Mutation by ddPCR, Quantitative 0040168 JAK2 Gene, V617F Mutation, Quantitative 2013909 Joubert Syndrome Type 2 (TMEM216), 1 Variant 2009302 Li-Fraumeni (TP53) Sequencing 2013735 Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants 3001603 Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants 3001603 Magnesium, RBC 2010162 Mammaglobin by Immunohistochemistry 2013730 Magle Syrup Urine Disease, Type 1B (BCKDHB), 3 Variants 0051205 Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations	3003279 Gastrointestinal Pathogens Panel by PCR 0051438 Gaucher Disease (GBA), 8 Variants 3000258 Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation 0051684 Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations 2013740 Glycogen Storage Disease, Type 1A (G6PC), 9 Variants 0055656 Hemochromatosis (HFE) 3 Mutations 0030144 Heparin Anti-Xa, Low Molecular Weight Heparin 0030143 Heparin Anti-Xa, Unfractionated 2005457 High-Specificity Antiphospholipid Antibodies, IgG and IgM 2002429 HLA-B*57:01 for Abacavir Sensitivity 2003020 Human Epididymis Protein 4 (HE4) Human Immunodeficiency Virus 1 (HIV-1) by Qualitative NAAT Human Immunodeficiency Virus 1 (HIV-1) by Qualitative Transcription-Mediated Amplification (TMA) 3001474 Qualitative Transcription-Mediated Amplification (TMA) 3001579 Hypertrophic Cardiomyopathy Panel, Sequencing 3003751 JAK2 (V617F) Mutation by ddPCR, Quantitative 0040168 JAK2 Gene, V617F Mutation, Quantitative 2013909 Joubert Syndrome Type 2 (TMEM216), 1 Variant 2009302 Li-Fraumeni (TP53) Sequencing 2013735 Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants 3001603 Long QT Panel, Sequencing and Deletion/Duplication 0092079 Magnesium, RBC 2010162 Mammaglobin by Immunohistochemistry Maple Syrup Urine Disease, Type 1B (BCKDHB), 3 Variants Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations Meperidine, Urine Screen with Reflex to	3003279 Gastrointestinal Pathogens Panel by PCR 0051438 Gaucher Disease (GBA), 8 Variants Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation 0051684 Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations 2013740 Glycogen Storage Disease, Type 1A (G6PC), 9 Variants 0055656 Hemochromatosis (HFE) 3 Mutations 0030144 Heparin Anti-Xa, Low Molecular Weight Heparin 0030143 Heparin Anti-Xa, Unfractionated High-Specificity Antiphospholipid Antibodies, IgG and IgM 2002429 HLA-B*57:01 for Abacavir Sensitivity 2003020 Human Epididymis Protein 4 (HE4) 3003760 Human Immunodeficiency Virus 1 (HIV-1) by Qualitative NAAT Human Immunodeficiency Virus 1 (HIV-1) by Qualitative Transcription-Mediated Amplification (TMA) 3001474 Human Immunodeficiency Virus 1 (HIV-1) Qualitative by NAAT, Whole Blood 3001579 Hypertrophic Cardiomyopathy Panel, Sequencing 3003751 JAK2 (V617F) Mutation by ddPCR, Quantitative 0040168 JAK2 Gene, V617F Mutation, Quantitative 2013909 Joubert Syndrome Type 2 (TMEM216), 1 Variant 2009302 Li-Fraumeni (TP53) Sequencing 2013735 Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants 3001603 Long QT Panel, Sequencing and Deletion/Duplication 0092079 Magnesium, RBC 2010162 Mammaglobin by Immunohistochemistry Maple Syrup Urine Disease, Type 1B (BCKDHB), 3 Variants 0051205 Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations Meperidine, Urine Screen with Reflex to	3003279 Gastrointestinal Pathogens Panel by PCR	3003279 Gastrointestinal Pathogens Panel by PCR 0051438 Gaucher Disease (GBA), 8 Variants x 3000258 Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation x x 0051684 Mutations x x x x x x x x x	3003279 Gastrointestinal Pathogens Panel by PCR						



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37	<u>2012245</u>	Methadone, Urine Screen with Reflex to Quantitation				х								
37	0055655	Methylenetetrahydrofolate Reductase (MTHFR) 2 Variants				Х								
38	<u>3001593</u>	MODY and Neonatal Diabetes Panel, Sequencing											X	
38	0051448	Mucolipidosis Type IV (MCOLN1), 2 Variants				X								
38	<u>2005023</u>	Narcolepsy (HLA-DQB1*06:02) Genotyping				X								
39	<u>2013745</u>	NEB-Related Nemaline Myopathy, 1 Variant				X								
39	0051458	Niemann-Pick Type A (SMPD1), 4 Variants				X								
51	0080281	NMP22, Urine												X
39	<u>2014599</u>	Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping				X								
51	<u>2004195</u>	Noonan Syndrome (SOS1) Sequencing												X
39	2005096	Opiates, Screen Only, Urine				X								
39	2005093	Opiates, Urine Screen with Reflex to Quantitation				X								
39	2008767	Opioid Receptor, mu <i>OPRM1</i> Genotype, 1 Variant				X								
40	<u>3001607</u>	Osteogenesis Imperfecta and Low Bone Density Panel, Sequencing											X	
40	2005103	Oxycodone/Oxymorphone Screen Only, Urine				X								
40	2005100	Oxycodone/Oxymorphone, Urine Screen with Reflex to Quantitation				X								
40	<u>2012265</u>	Phencyclidine, Urine Screen with Reflex to Quantitation				X								
41	2004980	Plasminogen Activator Inhibitor-1, PAI-1 (SERPINE1) Genotyping				X								
41	0030160	Platelet Aggregation Studies				X								
41	3001170	Platelet Antigen 1 Genotyping (HPA-1)				X								
41	3000193	Platelet Antigen Genotyping Panel				X								
42	3003723	Propoxyphene and Metabolite, Serum or Plasma											X	
51	2010464	Propoxyphene and Metabolite, Serum or Plasma, Quantitative												X
42	3003726	Propoxyphene and Metabolite, Urine											X	
51	2010468	Propoxyphene and Metabolite, Urine, Quantitative												Х
43	2012269	Propoxyphene, Urine Screen with Reflex to Quantitation				X			X			X		
43	0056060	Prothrombin (F2) c.*97G>A (G20210A) Pathogenic Variant				X								
51	2002722	PTEN-Related Disorders (PTEN) Sequencing												X



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43	3001053	Red Blood Cell Antigen Genotyping				X								
51	0051378	Rett Syndrome (MECP2), Full Gene Sequencing												X
43	0051368	RhD Gene (RHD) Copy Number				X								
44	<u>2012618</u>	Risk of Ovarian Malignancy Algorithm				X								
44	2003382	Ristocetin-Induced Platelet Aggregation				X								
44	<u>2008426</u>	SLCO1B1, 1 Variant				X								
51	<u>2011704</u>	Smith-Lemli-Opitz Syndrome (DHCR7) Sequencing, Fetal												X
45	<u>3001613</u>	Stickler Syndrome Panel, Sequencing											X	
45	2012294	Tapentadol Urine Screen with Reflex to Quantitation				X								
45	0051428	Tay-Sachs Disease (HEXA), 7 Variants				X								
46	3003595	TCL1 by Immunohistochemistry											X	
46	3003407	TDP43 by Immunohistochemistry											X	
47	<u>2012270</u>	THC (Cannabinoids), Urine Screen with Reflex to Quantitation				X								
47	0056200	Thrombotic Risk, DNA Panel				X								
47	<u>3001535</u>	TPMT and NUDT15				X								
47	<u>2012297</u>	Tramadol, Urine Screen with Reflex to Quantitation				X	X							
47	<u>2013750</u>	Usher Syndrome, Types 1F and 3 (<i>PCDH15</i> and <i>CLRN1</i>), 2 Variants				X								
48	<u>3002581</u>	Vaginitis Panel by TMA											X	i .
48	<u>3003676</u>	Vedolizumab Quantitation with Antibodies, Serum											X	
51	<u>2002001</u>	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing												X
49	3001541	Warfarin Sensitivity (CYP2C8, CYP2C9, CYP4F2, VKORC1) Genotyping				X								
49	<u>2006352</u>	X-Chromosome Inactivation Analysis				X		X						
50	<u>3003758</u>	Zinc, Red Blood Cells											X	
50	<u>2012300</u>	Zolpidem Urine with Reflex to Quantitation				X								



2013725 ABCC8-Related Hyperinsulinism, 3 Variants

ABCC8

Specimen Required: Collect: Lavender (EDTA), Pink (K₂EDTA), yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051266 Achondroplasia (*FGFR3*) 2 Mutations

AD PCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Do not freeze. Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Serum. Frozen specimens in glass collection tubes. Hemolyzed specimens. <u>Stability (collection to initiation of testing):</u> Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051265 Achondroplasia (FGFR3) 2 Mutations, Fetal

AD PCR FE

Specimen Required: Collect: Fetal Specimen: Two T-25 flasks at 80% confluency of cultured amniocytes. If the client is unable to culture amniocytes,

this can be arranged by contacting ARUP Client Services at (800) 522-2787. Or amniotic fluid.

AND Maternal Specimen: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Cultured Amniocytes: Fill flasks with culture media. Transport two T-25 flasks at 80% confluency of

cultured amniocytes. Backup cultures must be retained at the client's institution until testing is complete.

OR Amniotic Fluid: Transport 10 mL unspun fluid. (Min: 5 mL) **AND Maternal Specimen:** Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours

of shipment due to liability of cells. **Amniotic Fluid:** Room temperature. **Maternal Specimen:** Room temperature.

Remarks: Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination, Maternal Specimen. This can be arranged by contacting ARUP genetic counselors at (800) 242-2787 ext. 2141. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.

Unacceptable Conditions: Frozen specimens in glass collection tubes

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Maternal Specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month



2011708

Alpha Globin (*HBA1* and *HBA2*) Sequencing and Deletion/Duplication (Extended TAT as of 1/11/21-no referral available)

HBA FGA

Interpretive Data:

Background Information for Alpha Globin (HBA1 and HBA2) Sequencing and Deletion/Duplication Characteristics:

Alpha thalassemia is caused by decreased or absent synthesis of the hemoglobin alpha chain resulting in variable clinical presentations. Alpha (+) thalassemia results from variants of a single *HBA2* globin gene (-a/aa) and is clinically asymptomatic (silent carrier). Alpha (0) thalassemia (trait) is caused by variants of both *HBA2* globin

genes (-a/-a) or variants in the *HBA1* and *HBA2* globin genes on the same chromosome (--/aa) and results in mild microcytic anemia. Hemoglobin H disease occurs due to variants of three alpha globin genes (--/-a) and results in hemolysis with Heinz bodies, moderate anemia, and splenomegaly. Hb Bart Hydrops Fetalis Syndrome results when variants occur in all four alpha globin genes (--/--) and is lethal in the fetal or early neonatal period. Alpha globin gene triplications result in three active alpha globin genes on a single chromosome. Nondeletional alpha globin variants may be pathogenic or benign; both may result in an abnormal protein detectable by hemoglobin evaluation. Pathogenic nondeletional variants often have a more severe effect than single gene deletions

Incidence: Carrier frequency in Mediterranean (1:30-50), Middle Eastern, Southeast Asian (1:20), African, African American (1:3).

Inheritance: Autosomal recessive.

Cause: Pathogenic variants in the alpha globin gene cluster.

Clinical Sensitivity: 99 percent.

Methodology: Bidirectional sequencing of the *HBA1* and *HBA2* coding regions, intron-exon boundaries and 3' polyadenylation signal. Multiplex ligation-dependent probe amplification (MLPA) of the alpha globin gene cluster (*HBZ*, *HBM*, *HBA1*, *HBA2*, *HBQ1*) and its HS-40 regulatory region.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. Sequence analysis will not detect all regulatory region variants or variants in alpha globin cluster genes other than *HBA1* and *HBA2*. Sequencing of both *HBA1* and *HBA2* may not be possible in individuals harboring large alpha globin deletions on both alleles. This assay is unable to sequence *HBA2-HBA1* fusion genes; thus, *HBA1* or *HBA2* sequence variants occurring in cis with a 3.7 kb deletion or other *HBA2-HBA1* hybrid gene will not be detected. It may not be possible to determine phase of identified sequence variants. Specific breakpoints of large deletions/duplications will not be determined; therefore, it may not be possible to distinguish variants of similar size. Individuals carrying both a deletion and duplication within the alpha globin gene cluster may appear to have a normal number of alpha globin gene copies. Rare syndromic or acquired forms of alpha thalassemia associated with *ATRX* variants will not be detected.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.



New Test

3003651

Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring

HBA DDCS

Available Now Click for Pricing



Additional Technical Information

Methodology: Multiplex Ligation-dependent Probe Amplification with Sanger sequencing confirmation of HbCS

Performed: Varies

Reported: 10-14 days; 21 days if reflexed

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 2 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Stability (collection to initiation of testing): Room Temperature: 7 days; Refrigerated: 1 month

Reference Interval: By report

Interpretive Data:

Background Information: Alpha Globin (HBA1 and HBA2) Deletion/Duplication

Characteristics: Decreased or absent synthesis of the hemoglobin (Hb) alpha-chain resulting in clinical presentations ranging from asymptomatic silent carriers to severe anemia and fetal lethality. Alpha thalassemia silent carrier commonly results from deletion of a single alpha globin gene (-a/aa) and is clinically asymptomatic. Alpha thalassemia trait may be caused by deletion of a single alpha globin gene from both chromosomes (-a/-a), or deletion of the HBA1 and HBA2 globin genes from the same chromosome (--/aa). Heterozygosity for Hb Constant Spring (HbCS) is usually asymptomatic but may be associated with mild microcytic anemia. Homozygous HbCS is characterized by over hemolytic anemia, jaundice and splenomegaly. Hemoglobin H disease occurs due to inactivation of three alpha globin genes and results in hemolysis with Heinz bodies, moderate anemia, and splenomegaly. Hb Bart hydrops fetalis syndrome results from deletion of all four alpha globin genes (--/--) and is lethal in the fetal or early neonatal period. Alpha globin gene duplication results in three or more active alpha globin genes on a single chromosome.

Epidemiology: Carrier frequency of alpha thalassemia in African, African-American (1:3), Mediterranean (1:30-50), Middle Eastern, Southeast Asian (1:20).

Inheritance: Autosomal recessive.

Cause: Pathogenic variants in the alpha globin gene cluster (HBZ, HBM, HBA2, HBA1, HBQ1) or regulatory region.

Clinical Sensitivity: Varies by ethnicity, at least 90 percent.

Methodology: Multiplex ligation-dependent probe amplification (MLPA) for the *HBZ*, *HBM*, *HBA2*, *HBA1*, and *HBQ1* genes, the HS-40 regulatory region, and Hb Constant Spring (HbCS) *HBA2* c.427T>C; p.Ter143Gln. To determine copy number of HbCS in absence of a concurrent deletion of *HBA2*, PCR and bidirectional sequencing for HbCS is performed.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. Specific breakpoints of large deletions/duplications will not be determined; therefore, it may not be possible to distinguish variants of similar size. Non-deletional variants within the coding or regulatory regions of the alpha globin cluster genes, other than HbCS, will not be targeted. Individuals carrying both a deletion and duplication within the alpha globin gene cluster may appear to have a normal number of alpha globin gene copies. Rare syndromic or acquired forms of alpha thalassemia associated with *ATRX* gene variants will not be detected.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: If a concurrent deletion of *HBA2* is not identified, PCR and bidirectional sequencing for the HbCS copy number will be performed. Additional charges apply.

CPT Code(s): 81269; if reflexed, add 81257

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



New Test

3003656

Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring, Fetal

HBA DDCSFE

Available Now Click for Pricing



Additional Technical Information



Patient History Form for Fetal Molecular Testing

Methodology: Multiplex Ligation-dependent Probe Amplification with Sanger sequencing confirmation of HbCS

Performed: Varies **Reported:** 10 days

Specimen Required: Collect: Fetal Specimen: Two T-25 flasks at 80 percent confluency of cultured amniocytes or CVS. If the client is unable to culture

amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Or amniotic fluid.

AND Maternal Specimen: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Cultured Amniocytes or Cultured CVS: Fill flasks with culture media. Transport two T-25 flasks at 80 percent confluency of cultured amniocytes or cultured CVS. Backup cultures must be retained at the client's institution until testing is complete.

OR Amniotic Fluid: Transport 10 mL unspun fluid.

AND Maternal Specimen: Transport 2 mL whole blood. (Min: 1 mL) <u>Storage/Transport Temperature</u>: **Amniotic Fluid:** Room temperature.

Cultured Fetal Cells: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to viability of cells.

Maternal Specimen: Room temperature.

Remarks: Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission. Patient History Form is

available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.

Unacceptable Conditions:

Stability (collection to initiation of testing): Fetal Specimen: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen:

Unacceptable

Maternal Specimen: Room temperature: 7 days; Refrigerated: 1 month

Reference Interval: By report

Interpretive Data:

Background Information: Alpha Globin (HBA1 and HBA2) Deletion/Duplication

Characteristics: Decreased or absent synthesis of the hemoglobin (Hb) alpha-chain resulting in clinical presentations ranging from asymptomatic silent carriers to severe anemia and fetal lethality. Alpha thalassemia silent carrier commonly results from deletion of a single alpha globin gene (-a/aa) and is clinically asymptomatic. Alpha thalassemia trait may be caused by deletion of a single alpha globin gene from both chromosomes (-a/-a), or deletion of the HBA1 and HBA2 globin genes from the same chromosome (--/aa). Heterozygosity for Hb Constant Spring (HbCS) is usually asymptomatic but may be associated with mild microcytic anemia. Homozygous HbCS is characterized by overt hemolytic anemia, jaundice and splenomegaly. Hemoglobin H disease occurs due to inactivation of three alpha globin genes and results in hemolysis with Heinz bodies, moderate anemia, and splenomegaly. Hb Bart hydrops fetalis syndrome results from deletion of all four alpha globin genes (--/--) and is lethal in the fetal or early neonatal period. Alpha globin gene duplication results in three or more active alpha globin genes on a single chromosome.

Epidemiology: Carrier frequency of alpha thalassemia in African, African-American (1:3), Mediterranean (1:30-50), Middle Eastern, Southeast Asian (1:20).

Inheritance: Autosomal recessive.

Cause: Pathogenic variants in the alpha globin gene cluster (HBZ, HBM, HBA2, HBA1, HBQ1) or regulatory region.

Clinical Sensitivity: Varies by ethnicity, at least 90 percent.

Methodology: Multiplex ligation-dependent probe amplification (MLPA) for the *HBZ*, *HBM*, *HBA2*, *HBA1*, and *HBQ1* genes, the HS-40 regulatory region, and Hb Constant Spring (HbCS) *HBA2* c.427T>C; p.Ter143Gln. To determine copy number of HbCS in absence of a concurrent deletion of *HBA2*, PCR and bidirectional sequencing for HbCS is performed.

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. Specific breakpoints of large deletions/duplications will not be determined; therefore, it may not be possible to distinguish variants of similar size. Non-deletional variants within the coding or regulatory regions of the alpha globin cluster genes, other than HbCS, will not be detected. Fetuses carrying both a deletion and duplication within the alpha globin gene cluster may appear to have a normal number of alpha globin gene copies. Rare syndromic or acquired forms of alpha thalassemia associated with *ATRX* gene variants will not be detected.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: If a concurrent deletion of *HBA2* is not identified, PCR and bidirectional sequencing for the HbCS copy number will be performed. Additional charges apply.



CPT Code(s): 81269; 81265; if reflexed, add 81257

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0050002 Alpha-1-Acid Glycoprotein

A1A GLYCO

Specimen Required: Collect: Serum separator tube or green (sodium or lithium heparin).

Specimen Preparation: Separate serum or plasma from cells ASAP or within 2 hours of collection. Transfer 1 mL serum or plasma to

an ARUP Standard Transport Tube. (Min: 0.5 mL) Storage/Transport Temperature: Refrigerated. Unacceptable Conditions: Hemolyzed specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: 8 hours; Refrigerated: 8 days; Frozen: 3 months

ODS1256 Alpha-1-Antitrypsin (SERPINA1) Enzyme Concentration and 2 Mutations with Reflex to Alpha-1-Antitrypsin Phenotype

A1A GENO

Specimen Required: Collect: Serum separator tube AND lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Allow serum to clot completely at room temperature. Separate serum from cells ASAP or within 2 hours of

collection. Transport 0.5 mL serum AND 3 mL whole blood. (Min: 0.5 mL serum AND 0.5 mL whole blood)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Serum: Ambient: 1 week; Refrigerated: 3 months; Frozen: 3 months (avoid repeat

freeze/thaw cycles)

Whole Blood: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month.

2012209 Amphetamines Urine Screen with Reflex to Quantitation

AMP RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4.0 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2.0 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Breast milk.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2005419 Androstanediol Glucuronide Quantitative, Serum or Plasma

ANDRO GLUC

Performed: Varies
Reported: 8-18 days

Specimen Required: Collect: Plain Red, Lavender (EDTA), or Serum Separator Tube (SST).

Specimen Preparation: Separate from cells within 45 minutes. Transfer 1 mL serum or plasma to an ARUP Standard Transport Tube.

(Min: 0.5 mL)

Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.

Storage/Transport Temperature: Frozen. Also acceptable: Room temperature or refrigerated. Stability (collection to initiation of testing): Ambient: 6 days; Refrigerated: 6 days; Frozen: 3 years

2005077 Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR

AS PWS

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Frozen specimens in glass collection tubes.



2012232 Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR,

AS PWS FE

Fetal

Specimen Required: Collect: Fetal Specimen: Four (4) T-25 flasks at 80 percent confluency of cultured amniocytes. If the client is unable to culture

amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Or amniotic fluid.

AND Maternal Specimen: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Cultured Amniocytes: Fill flasks with culture media. Transport four (4) T-25 flasks at 80 percent confluency

of cultured amniocytes. Backup cultures must be retained at the client's institution until testing is complete.

OR Amniotic Fluid: Transport 20 mL unspun fluid. (Min: 10 mL) **AND Maternal Specimen**: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours

of shipment due to liability of cells. **Amniotic Fluid**: Room temperature. **Maternal Specimen**: Room temperature.

Remarks: Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination, Maternal Specimen. This can be arranged by contacting ARUP genetic counselors at (800) 242-2787 ext. 2141. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.

Unacceptable Conditions: Frozen specimens in glass collection tubes

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Maternal Specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0050392 Ankylosing Spondylitis (HLA-B27) Genotyping

HLAB27 PCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Do not freeze. Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum; collection of specimen in sodium heparin tubes. Frozen specimens in glass collection

tubes

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3000601 Antinuclear Antibody (ANA) with HEp-2 Substrate, IgG by IFA with Reflex by ANA AB PAT

Pattern

Specimen Required: Collect: Serum Separator Tube (SST).

Specimen Preparation: Separate from cells ASAP or within 2 hours of collection. Transfer 2 mL serum to an ARUP Standard

Transport Tube. (Min: 1.0 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Non-serum specimens. Contaminated, grossly hemolyzed, heat-inactivated, severely lipemic specimens. <u>Stability (collection to initiation of testing):</u> After separation from cells: Ambient: 48 hours; Refrigerated: 2 weeks; Frozen: 1 year

(avoid repeated freeze/thaw cycles)

0030192 APC Resistance Profile with Reflex to Factor V Leiden

APC R

Specimen Required: Collect: Light Blue (Sodium Citrate) AND Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B). Refer to Specimen

Handling at aruplab.com for hemostasis/thrombosis specimen handling guidelines.

Specimen Preparation: Transport 1.5 mL platelet-poor plasma AND 3 mL whole blood. (Min: 1 mL/each)

Storage/Transport Temperature: Plasma: CRITICAL FROZEN. Separate specimens must be submitted when multiple tests are

ordered.

Whole Blood: Refrigerated.

Unacceptable Conditions: Serum, clotted or hemolyzed specimens. Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Plasma: Ambient: 4 hours; Refrigerated: Unacceptable; Frozen at -20°C: 3 months;

Frozen at -70°C: 6 months

Whole Blood: Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

0055654 Apolipoprotein B (APOB) Mutation Detection

APO B

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.



2013341 Apolipoprotein E (*APOE*) Genotyping, Alzheimer Disease Risk

APOE AZ

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Remarks:</u> Testing of fetal specimens or specimens from patients under the age of 18 years is not offered. <u>Unacceptable Conditions:</u> Plasma or serum. Heparinized specimens. <u>Frozen specimens in glass collection tubes.</u> <u>Stability (collection to initiation of testing):</u> Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month.

2013337 Apolipoprotein E (*APOE*) Genotyping, Cardiovascular Risk

APOE CR

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Remarks: This test is not recommended for nonsymptomatic patients under 18 years of age.

 $\underline{Unacceptable\ Conditions:}\ Plasma\ or\ serum.\ He parinized\ specimens.\ Frozen\ specimens\ in\ glass\ collection\ tubes.$

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051415 Ashkenazi Jewish Diseases, 16 Genes

AJP

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

<u>3000724</u>

B-Lymphoblastic Leukemia (B-ALL) Minimum Residual Disease Detection by Flow Cytometry

B-ALL MRD

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Note: This assay is a minimal residual disease assessment of B-ALL by flow cytometry.

Available Markers*: CD3, CD9, CD10, CD13, CD19, CD20, CD33, CD34, CD38, CD45, CD58, CD71, Syto 16, CD66b, CD24, CD22

*Not all markers will be reported in all cases.

If COG panel is not specified, a 10 marker panel will be run: CD10, CD19, CD20, CD22, CD24, CD34, CD38, CD45, CD58, CD66b

If COG panel is specified, indicate time point and specimen type:

DAY 8 Peripheral Blood sample will have CD10, CD19, CD20, CD34, CD45, and Syto 16 run and reported. (6 markers total).

Day 29 Bone Marrow sample will have CD3, CD9, CD10, CD13, CD19, CD20, CD33, CD34, CD38, CD45, CD58, CD71, Syto 16 run and reported. (13 markers total).

The report will include a pathologist interpretation and a marker interpretation range corresponding to CPT codes of 2-8 markers, 9-15 markers interpreted. Charges apply per marker.

HOTLINE NOTE: There is a clinically significant charting name change associated with this test.

Change the charting name for component 3000738, B-ALL MRD (COG Protocol) Interpretation from B-ALL MRD (COG Protocol) Interpretation to B-ALL MRD Interpretation.



New Test 3002528 Bacterial Strain Typing by Next Generation Sequencing STRAIN NGS

Available Now Click for Pricing

Methodology: Massively Parallel Sequencing
Performed: Varies; Batch tested every two weeks

Reported: 1-3 weeks

Specimen Required: Collect: Multiple bacterial isolates that are epidemiologically related

Specimen Preparation: Transport individual isolates on agar slants or on swabs in bacterial transport media. Package isolates together

in a sealed container.

Storage/Transport Temperature: Room temperature.

Remarks: Contact the Microbiology Laboratory at (800) 242-2787, extension 2576, prior to submission of the isolates.

One report will be generated for each batch tested; billing is per isolate. Order one test, using "Infection control, ####" (your ARUP client number) as the patient name. Include a list of isolate identifiers (do not use patient names) on the requisition or as an order note

for electronic orders. Identifiers on the requisition must match identifiers on the isolate samples.

<u>Unacceptable Conditions:</u> Mixed cultures or non-viable organisms.

Stability (collection to initiation of testing): Isolate: Ambient: 1 week; Refrigerated: Unacceptable; Frozen: Unacceptable.

Reference Interval: By report

Interpretive Data:

Method

Whole Genome Sequencing (WGS) is performed using Ion Torrent sequencing chemistry. Reference-free pairwise comparisons are performed using short, overlapping sequence matching (kmer) analysis. Relationships are determined by the percent of kmers that match between isolate pairs.

Interpretation

Predicted relatedness is based on the total number of differences between the isolates, applying the thresholds shown in the table. The dendrogram and relationship matrix (see enhanced report) illustrate isolate relatedness. Interpretation of strain relatedness should be performed by an investigator knowledgeable about whole genome strain typing procedures and based on all available epidemiological evidence. Inferred relationships based on any strain typing method should not be used for individual patient management.

WGS Strain Typing provides substantial improvements in resolution and reproducibility when compared to pulsed-field gel electrophoresis (PFGE) and can be performed on a broad range of microorganisms. Test was validated for *Staphylococcus*, *Acinetobacter*, *Enterococcus*, *Escherichia*, *Pseudomonas*, *Stenotrophomonas*, *Serratia*, and *Klebsiella* species.

Category	Kmer Identity	Epidemiological Interpretation
Indistinguishable	≥99.9	Part of the outbreak
Closely related	99.8-99.2	Probably part of the outbreak
Possibly related	99.1-95.0	Possibly part of the outbreak
Unrelated	<95.0	Not part of the outbreak

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Note: Each Isolate billed separately

CPT Code(s): 87153

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



New Test 3002582 Bacterial Vaginosis by TMA BV TMA

Click for Pricing

Methodology: Qualitative Transcription-Mediated Amplification

Performed: Tue, Thu, Sat **Reported:** 1-4 days

Specimen Required: Patient Prep: Patient must be 14 years of age or older.

Collect: Vaginal specimen collected with pink swab from Aptima MultiTest Swab Collection kit (ARUP supply #55224 PK/50 or

#55229 PK/10) available online through eSupply using ARUP Connect or contact Client Services at (800) 522-2787. Specimen Preparation: Place swab in MultiTest Swab Specimen Transport Tube, break shaft at scoreline then recap tube.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens in any transport media other than indicated above. Specimen in MultiTest swab transport media

without a swab.

Stability (collection to initiation of testing): Ambient: 30 days; Refrigerated: 30 days; Frozen: 90 days

Reference Interval:

Test Number	Components	Reference Interval
	Bacterial Vaginosis by TMA	
		Negative

Interpretive Data:

A negative result does not preclude a possible infection.

A single qualitative result is determined based on relative amounts of the following target organisms: Lactobacillus (L. gasseri, L. crispatus, and L. jensenii), Gardnerella vaginalis, and Atopobium vaginae. This assay does not report individual organisms.

Results should be interpreted in conjunction with other clinical data. This test has not been validated for use with specimens collected by patients at home.

This test is intended for medical purposes only and is not valid for the evaluation of suspected sexual abuse or for other forensic purposes.

CPT Code(s): 81513

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2012211 Barbiturates, Urine Screen with Reflex to Quantitation BARB RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4.0 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2.0 mL)

 $\underline{Storage/Transport\ Temperature:}\ Room\ temperature.$

<u>Unacceptable Conditions:</u> Breast milk. Pharmaceutical preparation.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2012225 Benzodiazepines, Urine Screen with Reflex to Quantitation BENZ RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room Temperature

Unacceptable Conditions: Breast Milk. Pharmaceutical preparation. Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0051433 Bloom Syndrome (BLM), 1 Variant

BLM

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2012273 Buprenorphine, Urine Screen with Reflex to Quantitation

BUPR RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Unknown fluids, pharmaceutical preparations, and breast milk. Specimens exposed to repeated freeze/thaw

cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0051453 Canavan Disease (ASPA), 4 Variants

ASPA

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.



New Test 3002583 Candida glabrata, Candida species, and Trichomonas vaginalis by CVTV TMA

TMA

Click for Pricing

Methodology: Qualitative Transcription-Mediated Amplification

Performed: Tue, Thu, Sat **Reported:** 1-4 days

Specimen Required: Patient Prep: Patient must be 14 years of age or older.

Collect: Vaginal specimen collected with pink swab from Aptima MultiTest Swab Collection kit (ARUP supply #55224 PK/50 or #55229 PK/10) available online through eSupply using ARUP Connect or contact Client Services at (800) 522-2787.

Specimen Preparation: Place swab in MultiTest Swab Specimen Transport Tube, break shaft at scoreline then recap tube.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens in any transport media other than indicated above. Specimen in MultiTest swab transport media

without a swab.

Stability (collection to initiation of testing): Ambient: 30 days; Refrigerated: 30 days; Frozen: 90 days

Reference Interval:

Test Number	Components	Reference Interval
	Trichomonas vaginalis by TMA	
		Negative
	Candida glabrata by TMA	
		Negative
	Candida species (other) by TMA	
		Negative

Interpretive Data:

A negative result does not preclude a possible infection.

This test detects *Trichomonas vaginalis*, *Candida glabrata*, and other *Candida* species (*C. albicans*, *C. parapsilosis*, *C. dubliniensis*, and *C. tropicalis*). The assay does not differentiate among organisms in the *Candida* species group.

Results should be interpreted in conjunction with other clinical data. This test has not been validated for use with specimens collected by patients at home.

This test is intended for medical purposes only and is not valid for the evaluation of suspected sexual abuse or for other forensic purposes.

CPT Code(s): 87481, 87661

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



New Test

3003634

Capillary Malformation-Arteriovenous Malformation (CM-

AVM) Panel, Sequencing and Deletion/Duplication

Available Now **Click for Pricing**



Additional Technical Information



Patient History for Capillary Malformation-Arteriovenous Malformation (CM-AVM) Panel, Sequencing and Deletion/Duplication **Testing**

Methodology: Massively Parallel Sequencing/ Multiplex Ligation-dependent Probe Amplification

Performed: Varies Reported: 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: EPHB4** (NM 004444), RASA1 (NM 002890)

** Deletion/duplication detection is not available for this gene.

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2012278 Carisoprodol Urine with Reflex to Quantitation

CARIS RFXU

CMAVM NGS

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Breast milk. Pharmaceutical preparation. Specimens exposed to multiple freeze thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month



New Test 3003704 CD71 by Immunohistochemistry CD71 IHC

Available Now Click for Pricing

Methodology: Immunohistochemistry

Performed: Mon-Fri **Reported:** 1-3 days

Specimen Required: Collect: Tissue.

Specimen Preparation: Formalin fix (10 percent neutral buffered formalin) and paraffin-embed specimen (cells must be prepared into a cellblock). Protect paraffin block and/or slides from excessive heat. Transport tissue block or 5 unstained (3- to 5-micron thick sections), positively charged slides in a tissue transport kit recommended but not required. ARUP supply #47808 available online through eSupply using ARUP Connect or contact ARUP Client Services at (800) 522-2787. (Min: 2 slides) If sending precut slides, do not over help.

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated. Ship in cooled container during summer months.

<u>Unacceptable Conditions:</u> Specimens submitted with nonrepresentative tissue type. Depleted specimens.

Stability (collection to initiation of testing): Ambient: Indefinitely; Refrigerated: Indefinitely; Frozen: Unacceptable

Interpretive Data:

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Note: This test is performed as a stain and return (technical) service only.

CPT Code(s): 88342

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2005018 Celiac Disease (*HLA-DQ2*, and *HLA-DQ8*) Genotyping HLA CELIAC

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2012155 Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, *PMP22* CMT REFLEX Deletion/Duplication with Reflex to Sequencing Panel

Specimen Required: Collect: Lavender (EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

2012231 Cocaine, Urine Screen with Reflex to Quantitation COCA RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month



3002463 Connective Tissue Disease First Line Panel with Reflex

CTD PAN

Reference Interval:

Test Number	Components	Reference Interval				
0050215	Double-Stranded DNA (dsDNA) Antibody, IgG	Effective May 17, 2021				
	by ELISA with Reflex to	Test Number	Components		Reference Interva	
	dsDNA Antibody, IgG by		Double-Stranded DNA (dsl	ONA)	24 IUs or less	Negative
	IFA		Antibody, IgG by ELISA		25 to 30 IUs	Borderline Positive
			,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,		30 to 60 IUs	Low Positive
					60 to 200 IUs	Positive
					201 IUs or greater	Strong Positive
		2002693	Double-Stranded DNA (dsl Antibody, IgG by IFA (usin luciliae)		Less than 1:10	1 - 0
2002693	Double-Stranded DNA (dsDNA) Antibody, IgG by IFA (using <i>Crithidia</i> <i>luciliae</i>)	Less than 1:10				
0050470	Smith/RNP (ENA) Antibody, IgG					
		29 AU/mL or less		Negative		
		30-40 AU/mL		Equivocal		
		41 AU/mL or greater		Positive		
0050085	Smith (ENA) Antibody, IgG					
		29 AU/mL or less		Negative		
		30-40 AU/mL		Equivocal		
		41 AU/mL or greater		Positive		
2012074	SSA 52 and 60 (Ro) (ENA) Antibodies, IgG					
		Test Number	Components		Reference Interva	
			SSA-52 (Ro52) (ENA) Ant	ibody, IgG	29 AU/mL or Less:	Negative
					30-40 AU/mL: Equ	
					41 AU/mL or great	
			SSA-60 (Ro60) (ENA) Ant	ibody, IgG	29 AU/mL or Less: 30-40 AU/mL: Equ 41 AU/mL or great	ivocal
		_	_	•		-
0050692	SSB (La) (ENA) Antibody, IgG					
		29 AU/mL or less		Negative		
				Equivocal		
		30-40 AU/mL				
		41 AU/mL or greater		Positive		
0099592	Jo-1 Antibody, IgG					
0099592	Jo-1 Antibody, IgG					
0099592	Jo-1 Antibody, IgG	41 AU/mL or greater 29 AU/mL or less 30-40 AU/mL		Positive		
0099592	Jo-1 Antibody, IgG	41 AU/mL or greater 29 AU/mL or less		Positive Negative		
0099592	Scleroderma (Scl-70)	41 AU/mL or greater 29 AU/mL or less 30-40 AU/mL		Positive Negative Equivocal		
		29 AU/mL or less 30-40 AU/mL 41 AU/mL or greater		Negative Equivocal Positive		
	Scleroderma (Scl-70)	41 AU/mL or greater 29 AU/mL or less 30-40 AU/mL		Positive Negative Equivocal		

HOTLINE NOTE: There is a numeric map change associated with this test.

Change the numeric map for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from no numeric map to XXXX.

There is a result type change associated with this test.

Change the result type for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from Alpha to Numeric.

There is a unit of measure change associated with this test.

Change the unit of measure for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from no unit of measure to IU.



New Test 3003756 Copper, Red Blood Cells CU RBC

Click for Pricing

Methodology: Quantitative Inductively Coupled Plasma-Mass Spectrometry

Performed: Sun-Sat **Reported:** 1-4 days

Specimen Required: Collect: Royal Blue (EDTA).

Specimen Preparation: Centrifuge whole blood and separate RBCs from plasma within 2 hours of collection. Transfer 2 mL RBCs to an ARUP Trace Element-Free Transport Tube (ARUP supply #43116) available online through eSupply using ARUP ConnectTM or

contact ARUP Client Services at (800) 522-2787. (Min: 0.6 mL)

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated.

Unacceptable Conditions: Specimens collected in tubes other than royal blue (EDTA). Specimens transported in containers other than

Royal Blue (EDTA) tube or Trace Element-Free Transport Tube. Clotted or grossly hemolyzed specimens.

Stability (collection to initiation of testing): After separation from plasma: Ambient: 1 week; Refrigerated: 1 week; Frozen:

Unacceptable

Reference Interval: 59.0-91.0 mcg/dL

Interpretive Data:

Copper concentrations in RBCs reflect the intracellular stores and general homeostasis of Copper. Results may be falsely elevated if RBCs in the submitted specimen are lysed or not promptly separated from plasma.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

CPT Code(s): 82525

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0050180 C-Reactive Protein CRP

Specimen Required: Collect: Serum separator tube. Also acceptable: Plasma separator tube, green (lithium heparin)

Specimen Preparation: Allow serum tube to clot completely at room temperature. Separate serum or plasma from cells within 2 hours of collection. Transfer 1 mL serum to an ARUP Standard Transport Tube. (Min: 0.3 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Hemolyzed specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: 2 Weeks; Refrigerated: 3 Weeks; Frozen: 12

Months (if frozen within 24 hours)

0092572 Cutaneous Direct Immunofluorescence, Biopsy

CUTDIF

Specimen Required: Collect: Tissue: skin, mucosa (oral, conjunctival, genital, esophageal), other epithelium (gastrointestinal, respiratory, urinary).

<u>Specimen Preparation:</u> Transport tissue (optimal 4-6 mm) in Michel's medium (ARUP supply #45462) available online through eSupply using ARUP Connector call ARUP Client Services at (800) 522-2787. Also acceptable: Zeus tissue fixative. <u>Label container with transport medium type</u>, if not an ARUP-supplied vial.

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated.

<u>Unacceptable Conditions:</u> Formalin-fixed tissue. Solid organs or solid organ tissue. <u>Tissue in container of unknown or unacceptable</u>

transport medium.

Stability (collection to initiation of testing): Ambient: 10 days; Refrigerated: 10 days; Frozen: Unacceptable



3001508 *CYP2C19* 2C19GENO

Specimen Required: Collect: Lavender (EDTA), Pink (K₂EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Specimens collected in sodium heparin or lithium heparin. <u>Stability (collection to initiation of testing):</u> <u>Ambient:</u> 72 hours; Refrigerated: 1 week; Frozen: 1 month

3001513 CYP2D6 2D6GENO

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated

Remarks:

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

CF VAR

collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2013661 Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2013663 Cystic Fibrosis (CFTR) 165 Pathogenic Variants with Reflex to Sequencing CF VAR SEQ

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution).

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.



2013664 Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants with Reflex to Sequencing and

CFVAR COMP

Reflex to Deletion/Duplication

Specimen Required: Collect: Lavender (K₂EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 5 mL whole blood. (Min: 3 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2013662 Cystic Fibrosis (CFTR) 165 Pathogenic Variants, Fetal

CF VAR FE

Specimen Required: Collect: Fetal Specimen: Two T-25 flasks of cultured amniocytes at 80 percent confluency. *If the client is unable to culture

amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.

Maternal Specimen: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution).

Specimen Preparation: Cultured Amniocytes: Fill flasks with culture media. Backup cultures must be retained at the client's

institution until testing is complete.

Maternal Specimen: Transport 3 mL whole blood. (Min. 1 mL)

Storage/Transport Temperature: CulturedAmniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of skinwart due to lability of colls

of shipment due to lability of cells. **MaternalSpecimen:** Refrigerated.

Remarks: Maternal sample is recommended for proper test interpretation; order Maternal Cell Contamination, Maternal

Specimen. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.

Unacceptable Conditions: Maternal Specimen: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes.

Frozen specimens in glass collection tubes

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Maternal Specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3001524 Cytochrome P450 Genotyping Panel

CYP PANEL

 $\textbf{Specimen Required:} \ \underline{Collect:} \ Lavender \ (K_2EDTA), \ Pink \ (K_2EDTA), \ or \ Yellow \ (ACD \ Solution \ A \ or \ B).$

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

<u>0030057</u> D-Dimer D-DI

Performed: Sun-Sat Reported: 1-2 days

Specimen Required: Collect: Light blue (sodium citrate). Refer to Specimen Handling at aruplab.com for hemostasis/thrombosis specimen handling

guidelines.

Specimen Preparation: Transfer 2 mL platelet poor plasma to an ARUP Standard Transport Tube. (Min: 0.5 mL)

Storage/Transport Temperature: CRITICAL FROZEN. Separate specimens must be submitted when multiple tests are ordered.

Remarks:

Unacceptable Conditions: Serum. EDTA plasma, clotted or hemolyzed specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: Unacceptable; Refrigerated: Unacceptable; Frozen:

l month

University of Utah Clients: after separation from cells: Ambient: 8 hours; Refrigerated: Unacceptable; Frozen: 1 month

Interpretive Data:

The presence of rheumatoid factor may lead to false-positive results with the D-Dimer test. This test should not be used to rule out venous thromboembolism.

Maximal values less than 10 µg/mL FEU are rarely indicative of DIC.

Results are reported in Fibrinogen Equivalent Units (FEU).



Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants 2012166

DPYD

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Heparinized specimens. Frozen specimens in glass collection tubes. Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month.

New Test

3001581

Dilated Cardiomyopathy Panel, Sequencing

DCM NGS

Click for Pricing

Methodology: Massively Parallel Sequencing

Performed: Varies Reported: 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: ABCC9, ACTC1, ACTN2, ALMS1, BAG3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FKTN, FLNC*, GLA, JUP, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, PKP2, PLN, PRDM16, PRKAG2*, RAF1, RBM20, RYR2, SCN5A, SGCD, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN*, TTR, VCL.

* One or more exons are not covered by sequencing for the indicated gene; see Additional Technical Information.

CPT Code(s): 81439

0050215

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

Double-Stranded DNA (dsDNA) Antibody, IgG by ELISA with Reflex to dsDNA

DNA

Antibody, IgG by IFA

Reference Interval: Effective May 17, 2021

Test Number	Components	Reference Interva	l
	Double-Stranded DNA (dsDNA) Antibody, IgG by ELISA	24 IUs or less	Negative
		25 to 30 IUs	Borderline Positive
		30 to 60 IUs	Low Positive
		60 to 200 IUs	Positive
		201 IUs or greater	Strong Positive
2002693	Double-Stranded DNA (dsDNA) Antibody, IgG by IFA (using Crithidia luciliae)	Less than 1:10	

HOTLINE NOTE: There is a numeric map change associated with this test.

Change the numeric map for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from no numeric map to XXXX.

There is a result type change associated with this test.

Change the result type for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from Alpha to Numeric.

There is a unit of measure change associated with this test

Change the unit of measure for component 0050215, dsDNA Ab, IgG w/ Reflex to IFA Titer from no unit of measure to IU.



0092184 Drug Panel 7, Urine - Screen with Reflex to Confirmation/Quantitation

CDASU 7

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 8 mL urine with no additives or preservative to ARUP Standard Transport Tubes. (Min: 4 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0092185 Drug Panel 7A, Urine - Screen with Reflex to Confirmation/Quantitation

CDASU 7A

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 8 mL urine with no additives or preservatives to ARUP Standard Transport Tubes. (Min: 4 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0092186 Drug Panel 9, Urine - Screen with Reflex to Confirmation/Quantitation

CDASU 9

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 8 mL urine with no additives or preservatives to ARUP Standard Transport Tubes. (Min: 4 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

HOTLINE NOTE: There is a reflexive pattern change associated with this test.

Add reflex to 3003726, Propoxyphene and Metabolite, Urine

Remove reflex from 2010468, Propoxyphene and Metabolite, Urine, Quantitative

0092187 Drug Panel 9A, Urine - Screen with Reflex to Confirmation/Quantitation

CDASU 9A

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 8 mL urine with no additives or preservatives in ARUP Standard Transport Tubes. (Min: 4 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

HOTLINE NOTE: There is a reflexive pattern change associated with this test.

Add reflex to 3003726, Propoxyphene and Metabolite, Urine

Remove reflex from 2010468, Propoxyphene and Metabolite, Urine, Quantitative



2012312 **Drug Profile, Screen with Reflex to Quantitation**

PAIN RFX U

Specimen Required: Collect: Random Urine.

Specimen Preparation: Transfer 4 mL each into two (2) ARUP Standard Transport Tubes urine with no additives or preservatives.

(Min: 2 mL each)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to multiple freeze/thaw cycles, Pharmaceutical preparation. Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; frozen: 1 month

Reference Interval:

Effective May 17, 2021

Drugs covered and range of cutoff concentrations

Drugs/Drug Classes	Screen
Amphetamines	300 ng/mL
Barbiturates	200 ng/mL
Benzodiazepines	200 ng/mL
Buprenorphine	5 ng/mL
Carisoprodol	100 ng/mL
Cocaine	150 ng/mL
Ethyl Glucuronide	500 ng/mL
Fentanyl	1 ng/mL
MDMA (Ecstasy)	500 ng/mL
Meperidine	200 ng/mL
Methadone	150 ng/mL
Opiates	300 ng/mL
Oxycodone	100 ng/mL
Phencyclidine	25 ng/mL
Propoxyphene	300 ng/mL
Tapentadol	200 ng/mL
Tramadol	100 ng/mL
THC (Cannabinoids)	20 ng/mL
Zolpidem	20 ng/mL

HOTLINE NOTE: There is a reflexive pattern change associated with this test.

Add reflex to 3003726, Propoxyphene and Metabolite, Urine

Remove reflex from 2010468, Propoxyphene and Metabolite, Urine, Quantitative

2007479 Drug Profile, Targeted by Tandem Mass Spectrometry and Enzyme

PAIN HYB U

Immunoassay, Urine

HOTLINE NOTE: There is a clinically significant charting name change associated with this test.

Change the charting name for component 2007660, Tramadol (cutoff 200 ng/mL) from Tramadol (cutoff 200 ng/mL) to Tramadol (cutoff 100 ng/mL).

2009288 Drug Profile, Targeted with Interpretation by Tandem Mass Spectrometry and

PAIN HYB 2

Enzyme Immunoassay, Urine

HOTLINE NOTE: There is a clinically significant charting name change associated with this test.

Change the charting name for component 2007660, Tramadol (cutoff 200 ng/mL) from Tramadol (cutoff 200 ng/mL) to Tramadol (cutoff 100 ng/mL).

0090448 Drugs of Abuse 7 Panel, Urine - Screen Only

CDTI7

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month



0090449 Drugs of Abuse 7A Panel, Urine - Screen Only CDTI7A

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0090453 Drugs of Abuse 9 Panel, Urine - Screen Only CDT19

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0090454 Drugs of Abuse 9A Panel, Urine - Screen Only CDT19A

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0092280 Drugs of Abuse Test, Alcohol, Urine - Screen with Reflex to CDASUALC

Confirmation/Quantitation

Specimen Required: Collect: Random urine.

Specimen Preparation: Transport 10 mL urine. (Min: 2 mL)

Newborn minimum: 1 mL for the initial test and 1 mL for the confirmation of screen positive.

Storage/Transport Temperature: Room temperature.

Remarks: If applicable, indicate NEWBORN prominently on the test request form. <u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication with Reflex to Sequencing

DMD REFLEX

2011241

Specimen Required: Collect: Lavender (EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

0051463 Dysautonomia, Familial (IKBKAP), 2 Variants IKBKAP

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

0090518 Ethanol, Urine, Qualitative - Medical

ETOH URN

Specimen Required: Patient Prep: For medical purposes only.

Collect: Fresh random urine.

Specimen Preparation: Mix specimen well. Transfer 10 mL aliquot urine to a tightly sealed container for storage and transport. (Min:

 $0.5 \, mL)$

Storage/Transport Temperature: Refrigerated.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2012695 Ethyl Glucuronide Screen Only, Urine

ETG SCR UR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min. 1 mL)

Storage/Transport Temperature: Refrigerated.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2007912 Ethyl Glucuronide Screen with Reflex to Confirmation, Urine

ETG SCR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions:

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0097720 Factor V Leiden (F5) R506Q Mutation

FACV

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum; collection of specimen in sodium heparin tubes. Frozen specimens in glass collection

tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2003220 Factor XIII (F13A1) V34L Variant

FAC 13 MUT

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Frozen specimens in glass collection tubes.



New Test
Available Now

Familial Hypercholesterolemia Panel, Sequencing

FH NGS

Click for Pricing



3002110

Patient History for Familial Hypercholesterolemia Panel

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: APOB, LDLR, LDLRAP1, PCSK9.

CPT Code(s): 81407; 81479; 81406

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0051468 Fanconi Anemia, Group C (FANCC), 2 Variants

FANCC

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2012284 Fentanyl, Urine Screen with Reflex to Quantitation

FENT RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives in ARUP Standard Transport Tubes. (Min: 1 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles. Samples collected in tubes with additives or

preservatives.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

Reference Interval:

Effective May 17, 2021

Screen cutoff Concentration: 1 ng/mL



New Test 3003279 Gastrointestinal Pathogens Panel by PCR GIPPCR

Available Now Click for Pricing

Methodology: Qualitative Polymerase Chain Reaction

Performed: Mon-Sun **Reported:** 1-3 days

Specimen Required: Collect: Stool.

Specimen Preparation: Transfer stool to enteric transport media (Cary-Blair) (ARUP supply #29799) available online through eSupply

using ARUP ConnectTM or contact ARUP Client Services at (800) 522-2787. (Min: 0.5 mL)

Storage/Transport Temperature: Refrigerated

Remarks: This test is New York DOH approved; however, per NYDOH regulations, testing cannot be performed for New York City

clients.

<u>Unacceptable Conditions:</u> <u>Unpreserved stool, stool in media other than Cary-Blair, rectal swabs, specimens outside stability.</u> <u>Stability (collection to initiation of testing):</u> In enteric transport media: Ambient: 4 days, Refrigerated: 4 days, Frozen: Unacceptable.

Note: This assay detects Campylobacter spp. (C. jejuni, C. coli, C. upsaliensis), Clostridium difficle toxin A/B, Plesiomonas shigelloides, Salmonella spp., Vibrio spp. (V. parahaemolyticus, V. vulnificus, V. cholerae) including specific identification of V. cholerae, Yersinia enterocolitica, Enteroaggregative Escherichia coli (EAEC), Enteropathogenic Escherichia coli (EPEC), Enterotoxigenic Escherichia coli (ETEC) lt/st, Shiga-like toxin-producing Escherichia coli (STEC) stx1/stx2 including specific identification of E. coli O157, Shigella/Enteroinvasive Escherichia coli (EIEC), Cryptosporidium spp., Cyclospora cayetanensis, Entamoeba histolytica, Giardia lamblia (also known as G. intestinalis and G. duodenalis), Adenovirus F40/41, Astrovirus, Norovirus GI/GII, Rotavirus A, and Sapovirus (Genogroups I, II, IV, and V).

CPT Code(s): 87507

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0051438 Gaucher Disease (GBA), 8 Variants

 $\textbf{Specimen Required:} \ \underline{\textbf{Collect:}} \ Lavender \ (EDTA), pink \ (K_2EDTA), or \ yellow \ (ACD \ Solution \ A \ or \ B).$

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

GBA

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3000258 Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation CF FX SMA

Specimen Required: Collect: Lavender (K₂EDTA). Also acceptable: Pink (K₂EDTA) or Yellow (ACD Solution A).

Specimen Preparation: Transport 5 mL whole blood. (Min: 3 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051684 Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations G6PD AFRIC

Methodology: Real-Time Polymerase Chain Reaction

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Frozen specimens in glass collection tubes.



2013740 Glycogen Storage Disease, Type 1A (G6PC), 9 Variants G6PC

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0055656 Hemochromatosis (HFE) 3 Mutations HFE PCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0030144 Heparin Anti-Xa, Low Molecular Weight Heparin **HA LMW**

Performed: Sun-Sat Reported: 1-2 days

Specimen Required: Collect: Lt. blue (sodium citrate). Refer to Specimen Handling at aruplab.com for hemostasis/thrombosis specimen handling

Specimen Preparation: Centrifuge specimen within one hour of collection. Transport 2 mL platelet-poor plasma. (Min: 1.5 mL)

Storage/Transport Temperature: Frozen.

Remarks: This test cannot be used to quantitate anticoagulants other than low molecular weight heparin. This includes, but is not

limited to, direct oral anticoagulants and Fondaparinux (Arixtra).

Unacceptable Conditions: Serum. EDTA, oxalate, heparin, or plasma separator tubes. Specimens refrigerated more than eight hours. Hemolyzed specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: Unacceptable; Refrigerated: Unacceptable; Frozen:

University of Utah Clients: After separation from cells: Ambient: 4 hours; Refrigerated: 8 hours; Frozen: 2 weeks

0030143 Heparin Anti-Xa, Unfractionated HA UN

Performed: Sun-Sat Reported: 1-2 days

Specimen Required: Collect: Lt. blue (sodium citrate). Refer to Specimen Handling at aruplab.com for hemostasis/thrombosis specimen handling

Specimen Preparation: Centrifuge specimen within one hour of collection. Transport 2 mL platelet-poor plasma. (Min: 1 mL)

Storage/Transport Temperature: Frozen.

Remarks: This test cannot be used to quantitate anticoagulants other than unfractionated heparin. This includes, but is not limited to,

direct oral anticoagulants and Fondaparinux (Arixtra)

<u>Unacceptable Conditions:</u> Serum. EDTA, oxalate, heparin, or plasma separator tubes. Hemolyzed or clotted specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: Unacceptable; Refrigerated: Unacceptable; Frozen:

University of Utah Clients: After separation from cells: Ambient: 4 hours; Refrigerated: 8 hours; Frozen: 2 weeks

2002429 HLA-B*57:01 for Abacavir Sensitivity HLA-B5701

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Heparinized specimens. Frozen specimens in glass collection tubes.



2003020 Human Epididymis Protein 4 (HE4)

HE4

Specimen Required: Collect: Serum Separator Tube (SST). Also acceptable: Green (Lithium Heparin), Lavender (K2 EDTA), or Lavender (K3 EDTA).

Specimen Preparation: Allow specimen to clot completely at room temperature. Transfer 1 mL serum or plasma to an ARUP Standard

Transport Tube. (Min: 0.5 mL)

Storage/Transport Temperature: Frozen.

Unacceptable Conditions: Grossly hemolyzed specimens.

Stability (collection to initiation of testing): Ambient: 5 hours; Refrigerated: 48 hours; Frozen: 3 months

New Test 3003760 Human Immunodeficiency Virus 1 (HIV-1) by Qualitative NAAT HIV1QUAL Click for Pricing

Methodology: Qualitative Transcription-Mediated Amplification

Performed: Sun-Sat **Reported:** 1-4 days

Specimen Required: Collect: Lavender (EDTA), Pink (K₂EDTA), Yellow (ACD), or Plasma Preparation Tube (PPT).

Specimen Preparation: Separate from cells within 24 hours of collection. Transfer 2 mL plasma to an ARUP Standard Transport Tube

and freeze. (Min: 0.8 mL)

 $\underline{Storage/Transport\ Temperature:}\ Frozen$

Unacceptable Conditions: Heparinized specimens.

Stability (collection to initiation of testing): After separation from cells: Ambient: Unacceptable; Refrigerated: 72 hours; Frozen: 90

davs

Reference Interval: Not detected

Interpretive Data:

This test detects human immunodeficiency virus type 1 (HIV-1) RNA from Group M, N and O subtypes; it does not detect HIV-1 proviral DNA. A result of "Not Detected" does not rule out HIV-1 RNA concentrations below the limit of detection of the assay or the presence of inhibitors in the patient specimen. The diagnosis of HIV-1 infection should not be made based solely on a single HIV-1 test result. Diagnosis requires repeat and confirmatory testing as recommended by U.S. Health and Human Services Guidelines. Improper specimen handling can cause false negatives or contamination.

This assay should not be used for blood donor screening, associated re-entry protocols, or for screening Human Cell, Tissues and Cellular Tissue-Based Products (HCT/P).

Note: Assay detects HIV-1 virus RNA. Proviral DNA will not be detected.

CPT Code(s): 87535

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



New Test 3001579 Hypertrophic Cardiomyopathy Panel, Sequencing HCM NGS

Click for Pricing

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: ACTC1, ACTN2, AGL, ALPK3, BRAF, CACNA1C, CSRP3, DES, FHL1, FLNC, GAA, GLA, HRAS, JPH2, KRAS, LAMP2, MAP2K1, MAP2K2, MYBPC3, MYH7, MYL2, MYL3, NRAS, PLN, PRKAG2, PTPN11, RAF1, RIT1, SOS1, TNNC1, TNN13, TNNT2, TPM1, TTR

CPT Code(s): 81439

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



New Test

3003751 JAK2

JAK2 (V617F) Mutation by ddPCR, Quantitative

JAK2V617FQ

Click for Pricing



Additional Technical Information

Methodology: Droplet Digital Polymerase Chain Reaction

Performed: DNA Isolation: Sun-Sat

Assay: Varies

Reported: 2-7 days

Specimen Required: Collect: Whole blood or bone marrow: Lavender (EDTA), preferred. Also acceptable: Green (sodium heparin)

Specimen Preparation: Whole Blood: Do not freeze. Transport 5 mL whole blood. (Min: 1 mL)

Bone Marrow: Do not freeze. Transport 3 mL bone marrow. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma, serum, FFPE tissue blocks/slides, or fresh or frozen tissue, DNA extracted by a non-CLIA certified

lab. Specimens collected in anticoagulants other than EDTA or sodium heparin. Clotted or grossly hemolyzed specimens.

Stability (collection to initiation of testing): Refrigerated: 7 days; Frozen: Unacceptable

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

CPT Code(s): 81270

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2013909 Joubert Syndrome Type 2 (*TMEM216*), 1 Variant

TMEM216

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2013735 Lipoamide Dehydrogenase Deficiency (*DLD*), 2 Variants

DLD

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.



New Test 3001603 Long QT Panel, Sequencing and Deletion/Duplication LQT NGS

Available Now Click for Pricing

Methodology: Massively Parallel Sequencing / Exonic Oligonucleotide-based CGH Microarray

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: CACNA1C, CALM1**, CALM2**, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A

 $\ensuremath{^{**}}$ - Deletion/duplication detection is not available for this gene.

CPT Code(s): 81403; 81404; 81406; 81407; 81414; 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0092079 Magnesium, RBC MG RBC

Reference Interval:

Effective May 17, 2021 3.6–7.5 mg/dL

Interpretive Data:

RBC magnesium results reflect the intracellular stores and general homeostasis of magnesium. Results may be falsely low if RBCs in the submitted specimen are lysed or not promptly separated from plasma.

RBC magnesium concentration is reported as milligrams per deciliter (mg/dL). To convert concentration to micromoles per liter (mmol/L), divide the result by 2.43.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes

HOTLINE NOTE: There is a unit of measure change associated with this test.

Change the unit of measure for component 0092079 Magnesium, RBC from mmol/L to mg/dL.



2010162 Mammaglobin by Immunohistochemistry

MAMMA IHC

Specimen Required: Collect: Tissue or cells.

Specimen Preparation: Formalin fix (10 percent neutral buffered formalin) and paraffin embed specimen (cells must be prepared into a cellblock). Protect paraffin block and/or slides from excessive heat. Transport tissue block or 3 unstained (4-micron thick sections), positively charged slides in a tissue transport kit (ARUP supply #47808) available online through eSupply using ARUP ConnectTM or contact ARUP Client Services at (800) 522-2787. (Min: 3 slides) If sending precut slides, do not oven bake.

Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated. Ship in cooled container during summer months.

<u>Unacceptable Conditions:</u> Specimens submitted with non-representative tissue type. Depleted specimens.

Stability (collection to initiation of testing): Ambient: Indefinitely; Refrigerated: Indefinitely; Frozen: Unacceptable

2013730 Maple Syrup Urine Disease, Type 1B (*BCKDHB*), 3 Variants

BCKDHB

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

<u>0051205</u> Medium Chain Acyl-CoA Dehydrogenase (ACADM) 2 Mutations

MCADPCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2012288 Meperidine, Urine Screen with Reflex to Quantitation

MEP RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens collected in tubes with additives or preservatives.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2012245 Methadone, Urine Screen with Reflex to Quantitation

METH RFX U

Specimen Required: Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

<u>Unacceptable Conditions:</u> Breast Milk. Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0055655 Methylenetetrahydrofolate Reductase (*MTHFR*) 2 Variants

MTHFR PCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Heparinized specimens. Frozen specimens in glass collection tubes.



New Test

3001593

MODY and Neonatal Diabetes Panel, Sequencing

MODY NGS

Click for Pricing



Additional Technical Information



Patient History for MODY and Neonatal Diabetes

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Serum or plasma; grossly hemolyzed or frozen specimens.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: ABCC8*, APPL1, BLK, CEL*, EIF2AK3, FOXP3, GATA4, GATA6, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11,

NEUROD1, NEUROG3, PAX4, PDX1, RFX6, SLC19A2, WFS1, ZFP57

*CEL (NM_001807) exons 1, 8, 9, 11 are not sequenced due to technical limitations of the assay.

*ABCC8 (NM_001351295) partial exon 14 (Chr11:17449973-17450018) is not sequenced due to technical limitations of the assay.

CPT Code(s): 81403; 81404; 81405; 81406; 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

0051448 Mucolipidosis Type IV (MCOLN1), 2 Variants

MCOLN1

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2005023 Narcolepsy (HLA-DQB1*06:02) Genotyping

NARCOLEPSY

Specimen Required: Collect: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.



2013745 *NEB*-Related Nemaline Myopathy, 1 Variant

NEB

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051458 Niemann-Pick Type A (SMPD1), 4 Variants

SMPD1

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2014599 Non-Alcoholic Fatty Liver Disease Susceptibility (PNPLA3) Genotyping

PNPLA3

Specimen Required: Collect: Lavender (EDTA).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2005096 Opiates, Screen Only, Urine

OPI SCR UR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2005093 Opiates, Urine Screen with Reflex to Quantitation

OPI RFX UR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2008767 Opioid Receptor, mu *OPRM1* Genotype, 1 Variant

OPRM1

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Heparinized specimens. Frozen specimens in glass collection tubes.



New Test 3001607 Osteogenesis Imperfecta and Low Bone Density Panel, Sequencing OI NGS

Click for Pricing

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: ALPL, ANO5, BMP1, CASR, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, CYP27B1, FKBP10, GORAB, IFITM5, LRP5*, P3H1, P4HB, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SLC34A3, SP7, SPARC, TMEM38B, WNT1

* One or more exons are not covered by sequencing for the indicated gene; see Additional Technical Information.

CPT Code(s): 81405; 81406; 81408; 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2005103 Oxycodone/Oxymorphone Screen Only, Urine

OXY SCR UR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2005100 Oxycodone/Oxymorphone, Urine Screen with Reflex to Quantitation

OXY RFX UR

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Specimens exposed to repeated freeze/thaw cycles.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

2012265 Phencyclidine, Urine Screen with Reflex to Quantitation

PCP RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Breast milk.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month



2004980 Plasminogen Activator Inhibitor-1, PAI-1 (SERPINE1) Genotyping

PAI-1 GENO

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

<u>0030160</u> Platelet Aggregation Studies

PLTAG

Specimen Required: Patient Prep: Instructions will be provided by the ARUP Hemostasis/Thrombosis lab upon receipt of Patient History for Platelet Aggregation Studies form.

Collect: Specimen collection to be scheduled by the ARUP Hemostasis/Thrombosis lab and performed at the ARUP Family Health Clinic.

Specimen Preparation: Performed by the ARUP Hemostasis/Thrombosis Lab.

Storage/Transport Temperature: CRITICAL ROOM TEMPERATURE.

<u>Remarks:</u> A completed Patient History Form must be submitted to Coagulation811@aruplab.com and approved prior to scheduling specimen collection. The Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.

<u>Unacceptable Conditions:</u> Specimens not scheduled by the ARUP Hemostasis/Thrombosis lab and not collected by ARUP.

Stability (collection to initiation of testing): Ambient: 1 hour; Refrigerated: Unacceptable; Frozen: Unacceptable

3001170 Platelet Antigen 1 Genotyping (HPA-1)

HPA-1 GENO

Specimen Required: Collect: Fetal Specimen: Amniotic fluid OR cultured amniocytes. If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.

WITH Maternal Cell Contamination Specimen (see Note): Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Parental Specimen: Lavender (EDTA).

Specimen Preparation: Amniotic Fluid: Transport 10 mL unspun fluid. (Min: 5 mL)

Cultured Amniocytes: Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.

Maternal Cell Contamination Specimen: Transport 3 mL whole blood. (Min: 1 mL)

Whole Blood (Parental Genotyping): Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Amniotic Fluid: Room temperature.

Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to liability of cells

Whole Blood or Maternal Cell Contamination Specimen: Refrigerated.

<u>Unacceptable Conditions:</u> Frozen specimens in glass collection tubes

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Whole Blood or Maternal Cell Contamination Specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3000193 Platelet Antigen Genotyping Panel

HPA GENO

Specimen Required: Collect: Fetal Genotyping: Amniotic fluid OR cultured amniocytes. If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787.

WITH Maternal Cell Contamination Specimen (see Note): Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Parental Genotyping: Lavender (EDTA).

Specimen Preparation: Amniotic Fluid: Transport 10 mL unspun fluid. (Min: 5 mL)

Cultured Amniocytes: Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.

Maternal Cell Contamination Specimen: Transport 3 mL whole blood. (Min: 1 mL)

Whole Blood (Parental Genotyping): Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Amniotic Fluid: Room temperature.

Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to liability of cells.

Whole Blood or Maternal Cell Contamination Specimen: Refrigerated.

<u>Unacceptable Conditions:</u> Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Whole Blood or Maternal Cell Contamination Specimen: Ambient: 72 hours; Refrigerated: 1 week, Frozen: 1 month



New Test 3003723 Propoxyphene and Metabolite, Serum or Plasma PROPOX SP

Click for Pricing

Methodology: Quantitative Gas Chromatography-Mass Spectrometry (GC-MS)

Performed: Varies **Reported:** 8 – 11 days

Specimen Required: Collect: Plain red, Lavender (K2EDTA or K3EDTA) or Pink (K2EDTA).

Specimen Preparation: Separate from cells within 2 hours. Transfer 2 mL serum or plasma to an ARUP Standard Transport Tube.

(Min: 0.7 mL)

Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.

Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature and frozen.

<u>Unacceptable Conditions:</u> Separator tubes.

Stability (collection to initiation of testing): Ambient: 2 weeks; Refrigerated: 2 weeks; Frozen: 3 months

Reference Interval: By report

Note: Amitriptyline is a known interference.

CPT Code(s): 80367 (Alt code: G0480)

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

New Test 3003726 Propoxyphene and Metabolite, Urine PROPOX U

Click for Pricing

Methodology: Quantitative Gas Chromatography-Mass Spectrometry (GC-MS)

Performed: Varies **Reported:** 8 – 11 days

Specimen Required: Collect: Urine

Specimen Preparation: Transfer 2 mL urine to an ARUP Standard Transport Tube. (Min: 0.7 mL)

Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.

Storage/Transport Temperature: Refrigerated. Also acceptable: Frozen

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 3 months

Reference Interval: By report

Note: Amitriptyline is a known interference.

CPT Code(s): 80367 (Alt code: G0480)

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



2012269 Propoxyphene, Urine Screen with Reflex to Quantitation

PPXY RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Breast milk.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

Note: If the specimen screens positive, then Confirmation/Quantitation by LC-MS/MS (ARUP test code 3003726) will be added to confirm result. Additional charges apply.

HOTLINE NOTE: There is a reflexive pattern change associated with this test.

Add reflex to 3003726, Propoxyphene and Metabolite, Urine

Remove reflex from 2010468, Propoxyphene and Metabolite, Urine, Quantitative

0056060 Prothrombin (F2) c.*97G>A (G20210A) Pathogenic Variant PT PCR

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum; collection of specimen in sodium heparin tubes. Frozen specimens in glass collection

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

3001053 **Red Blood Cell Antigen Genotyping**

RBC GENO

Specimen Required: Collect: Lavender (EDTA)

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum; collection of specimen in sodium heparin tubes. Frozen specimens in glass collection

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

0051368 RhD Gene (RHD) Copy Number

RHD

Specimen Required: Collect: Fetal Genotyping: Amniotic fluid OR two T-25 flasks at 80 percent confluency of cultured amniocytes. If the client is unable

to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787

WITH Maternal Cell Contamination Specimen (see Remarks): Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or

B).

Parental Genotyping: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Amniotic Fluid: Transport 10 mL unspun fluid. (Min: 5 mL)

Cultured Amniocytes: Fill flasks with culture media. Transport two T-25 flasks at 80 percent confluency of cultured amniocytes

filled with culture media. Backup cultures must be retained the client's institution until testing is complete.

Maternal Cell Contamination Specimen: Transport 3 mL whole blood (Min: 1 mL)

Whole Blood (Parental Genotyping): Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Amniotic fluid: Room temperature.

Cultured Amniocytes: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to liability of

cells.

Whole Blood or Maternal Cell Contamination Specimen: Refrigerated.

Remarks: Maternal specimen is recommended for proper test interpretation if contamination of the fetal specimen from the mother is suspected. Order Maternal Cell Contamination. Patient History Form is available on the ARUP website or by contacting ARUP Client Services.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): Fetal Specimen: Ambient: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Whole Blood or Maternal Cell Contamination Specimen: Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month



2012618 Risk of Ovarian Malignancy Algorithm

ROMA

Specimen Required: Collect: Serum Separator Tube (SST). Also acceptable: Green (Sodium or Lithium Heparin), Lavender (EDTA), or Pink (K2 EDTA).

Specimen Preparation: Allow specimen to clot completely at room temperature. Transfer 1.5 mL serum to an ARUP Standard

Transport Tube. (Min: 1 mL)

Storage/Transport Temperature: Frozen.

Unacceptable Conditions: Hemolyzed specimens.

Stability (collection to initiation of testing): Ambient: 5 hours; Refrigerated: 48 hours; Frozen: 3 months

2003382 Ristocetin-Induced Platelet Aggregation

RIPA

Specimen Required: Patient Prep: Instructions will be provided by the ARUP Hemostasis/Thrombosis lab upon receipt of Patient History for Platelet

Aggregation Studies form.

Collect: Specimen collection to be scheduled by the ARUP Hemostasis/Thrombosis lab and performed at the ARUP Family Health

Clinic.

Specimen Preparation: Performed by the ARUP Hemostasis/Thrombosis lab.

Storage/Transport Temperature: CRITICAL ROOM TEMPERATURE.

Remarks: A completed Patient History Form must be submitted to Coagulation811@aruplab.com and approved prior to scheduling specimen collection. The Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800)

522-2787

Unacceptable Conditions: Specimens not scheduled by the ARUP Hemostasis/Thrombosis lab and not collected by ARUP.

Stability (collection to initiation of testing): Ambient: 1 hour; Refrigerated: Unacceptable; Frozen: Unacceptable

2008426 SLCO1B1, 1 Variant

SLCO1B1

Specimen Required: Collect: Lavender (EDTA) or pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Heparinized specimens. Frozen specimens in glass collection tubes.



New Test 3001613 Stickler Syndrome Panel, Sequencing SS NGS

Available Now Click for Pricing

Methodology: Massively Parallel Sequencing

Performed: Varies **Reported:** 3-6 weeks

Specimen Required: Collect: Lavender (EDTA) or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1.5 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Serum or plasma; grossly hemolyzed or frozen specimens

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

Reference Interval: By report

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Note: Genes tested: COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, VCAN

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2012294 Tapentadol Urine Screen with Reflex to Quantitation TAPEN RFXU

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

 $\underline{Unacceptable\ Conditions:}\ Breast\ milk.\ Pharmaceutical\ preparation.\ Specimens\ exposed\ to\ repeated\ freeze/thaw\ cycles.$

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0051428 Tay-Sachs Disease (HEXA), 7 Variants HEXA

 $\textbf{Specimen Required:} \ \underline{Collect:} \ Lavender \ (EDTA), pink \ (K_2EDTA), or \ yellow \ (ACD \ Solution \ A \ or \ B).$

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in glass collection tubes.



TCL1 IHC 3003595 TCL1 by Immunohistochemistry **New Test**

Available Now **Click for Pricing**

Methodology: Immunohistochemistry

Performed: Mon-Fri Reported: 1-3 days

Specimen Required: Collect: Tissue.

Specimen Preparation: Formalin fix (10 percent neutral buffered formalin) and paraffin embed specimen (cells must be prepared into a cellblock). Protect paraffin block and/or slides from excessive heat. Transport tissue block or 5 unstained (3- to 5-micron thick sections), positively charged slides in a tissue transport kit, recommended but not required. ARUP supply #47808 available online through eSupply using ARUP Connect or contact ARUP Client Services at (800) 522-2787. (Min: 2 slides) If sending precut slides, do

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated. Ship in cooled container during summer months.

<u>Unacceptable Conditions:</u> Specimens submitted with non-representative tissue type. Depleted specimens.

Stability (collection to initiation of testing): Ambient: Indefinitely; Refrigerated: Indefinitely; Frozen: Unacceptable

Interpretive Data:

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Note: This test is performed as a stain and return (technical) service only.

CPT Code(s): 88342

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

TDP43 IHC **New Test** 3003407 TDP43 by Immunohistochemistry

Available Now **Click for Pricing**

Methodology: Immunohistochemistry

Performed: Mon-Fri Reported: 1-3 days

Specimen Required: Collect: Tissue.

Specimen Preparation: Formalin fix (10 percent neutral buffered formalin) and paraffin embed specimen (cells must be prepared into a cellblock). Protect paraffin block and/or slides from excessive heat. Transport tissue block or 5 unstained (3- to 5-micron thick sections), positively charged slides in a tissue transport kit (recommended but not required). ARUP supply #47808 available online through eSupply using ARUP Connect or contact ARUP Client Services at (800) 522-2787. (Min: 2 slides) If sending precut slides, do

not oven bake.

Storage/Transport Temperature: Room temperature. Also acceptable: Refrigerated. Ship in cooled container during summer months.

<u>Unacceptable Conditions:</u> Specimens submitted with non-representative tissue type. Depleted specimens.

Stability (collection to initiation of testing): Ambient: Indefinitely; Refrigerated: Indefinitely; Frozen: Unacceptable

Interpretive Data:

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Note: This test is performed as a stain and return (technical) service only.

CPT Code(s): 88342

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



2012270 THC (Cannabinoids), Urine Screen with Reflex to Quantitation

THC RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

Unacceptable Conditions: Breast milk.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

0056200 Thrombotic Risk, DNA Panel

THROMDNA

Specimen Required: Collect: Lavender (EDTA), pink (K2 EDTA) or yellow (ACD Solution A or B).

Specimen Preparation: Transport 4 mL whole blood. (Min: 3 mL)

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum; collection of specimen in sodium heparin tubes. Frozen specimens in glass collection

tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month.

 TPMT2

Specimen Required: Collect: Lavender (EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2012297 Tramadol, Urine Screen with Reflex to Quantitation

TRAM RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

 $\underline{Storage/Transport\ Temperature:}\ Refrigerated.$

<u>Unacceptable Conditions:</u> Pharmaceutical preparation. Specimens exposed to repeated freeze/thaw cycles. <u>Stability (collection to initiation of testing):</u> Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month

Reference Interval:

Effective May 17, 2021

Screen Cutoff concentration 100 ng/mL

2013750 Usher Syndrome, Types 1F and 3 (PCDH15 and CLRN1), 2 Variants

USHER

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Plasma or serum. Specimens collected in sodium heparin or lithium heparin tubes. Frozen specimens in

glass collection tubes.



New Test 3002581 Vaginitis Panel by TMA VPAN TMA

Click for Pricing

Methodology: Qualitative Transcription-Mediated Amplification

Performed: Tue, Thu, Sat **Reported:** 1-4 days

Specimen Required: Patient Prep: Patient must be 14 years of age or older.

Collect: Vaginal specimen collected with pink swab from Aptima MultiTest Swab Collection kit (ARUP supply #55224 PK/50 or

#55229 PK/10) available online through eSupply using ARUP Connect or contact Client Services at (800) 522-2787. Specimen Preparation: Place swab in MultiTest Swab Specimen Transport Tube, break shaft at scoreline, then recap tube.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Specimens in any transport media other than indicated above. Specimen in MultiTest swab transport media

without a swab.

Stability (collection to initiation of testing): Ambient: 30 days; Refrigerated: 30 days; Frozen: 90 days

Reference Interval:

Test Number	Components	Reference Interval
3002582	Bacterial Vaginosis by Transcription-mediated Amplification (TMA)	
		Negative
3002583	Candida glabrata, Candida species, and Trichomonas vaginalis by Transcription-mediated Amplification (TMA)	
		Negative

Interpretive Data:

See report

CPT Code(s): 81513, 87481, 87661

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

New Test 3003676 Vedolizumab Quantitation with Antibodies, Serum VEDOL AB

Click for Pricing

Methodology: Quantitative Liquid Chromatography/Mass Spectrometry (LC-MS/MS) / Electrochemiluminescent Immunoassay

Performed: Varies Reported: 7-17 days

Specimen Required: Patient Prep: 12 hours prior to specimen collection discontinue multivitamins or dietary supplements containing biotin (vitamin B7),

commonly found in hair, skin, and nail supplements. Nivolumab (Opdivo) must be discontinued at least 4 weeks prior to testing.

<u>Collect:</u> Plain red. Also acceptable: Serum separator tube (SST). Collect immediately before next scheduled dose (trough specimen).

<u>Specimen Preparation:</u> Separate from cells ASAP or within 2 hours of collection. Transfer 1.5 mL serum to an ARUP Standard

Transport Tube. (Min: 0.75 mL)

Test is not performed at ARUP; separate specimens must be submitted when multiple tests are ordered.

Storage/Transport Temperature: Refrigerated. Also acceptable: Frozen.

Stability (collection to initiation of testing): Ambient: Unacceptable; Refrigerated: 28 days; Frozen: 28 days

Reference Interval: By Report

CPT Code(s): 80280; 82397

New York DOH Approved.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.



3001541 Warfarin Sensitivity (CYP2C8, CYP2C9, CYP4F2, VKORC1) Genotyping

WARF PAN

Specimen Required: Collect: Lavender (EDTA), Pink (K₂EDTA), or Yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL) OR Transport the Saliva Collection Device.

Storage/Transport Temperature: Refrigerated.

Unacceptable Conditions: Plasma or serum. Specimens collected in sodium heparin or lithium heparin. Frozen specimens in glass

collection tubes.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month

2006352 X-Chromosome Inactivation Analysis

XCI

Specimen Required: Collect: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: Refrigerated.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 week; Frozen: Unacceptable

Interpretive Data:

Characteristics: Females usually have two copies of the X-chromosome, one of which becomes randomly inactivated early in embryonic development in a process known as lyonization. If either the paternally or maternally derived X-chromosome is preferentially inactivated, this results in a nonrandom or "skewed" pattern of X-chromosome inactivation (XCI). The pattern of XCI may vary among tissue types. XCI ratios of 50:50 to 74:26 suggest random XCI, ratios greater than 85:15 suggest nonrandom XCI, and ratios from 75:25 to 85:15 should be interpreted with caution.

Cause: Nonrandom XCI may result by chance or from secondary cell selection in females who are heterozygous for X-chromosome rearrangements, carriers of pathogenic variants in X-linked genes, or affected with neoplastic disease.

Gene Tested: The androgen receptor (AR) gene on the X chromosome.

Clinical Sensitivity: Approximately 90 percent. An estimated 10-15 percent of females have skewed X-inactivation by chance. However, skewed XCI may be seen more frequently with increasing age.

Methodology: Methylation-sensitive restriction digest followed by PCR and fragment analysis.

Limitations: Testing is limited to XX females only. This assay will be uninformative in up to 20 percent of females due to homozygosity for the polymorphic AR gene locus analyzed. XCI patterns may differ among tissues; therefore, the XCI ratio reported is for the tissue type tested with a standard deviation 0.08 for XCI ratios of 50:50-79:50; 0.05 for XCI ratios 80:20 or greater. Although this test will detect the methylation status of the X-chromosomes, it will not determine if the X-inactivation pattern is associated with rearrangements of the X chromosome, pathogenic variants in X-linked genes or neoplastic disease. If a nonrandom XCI pattern is present, the parent of origin of the active X cannot be determined without testing parental samples. XCI ratios should not be used to predict prognosis for female carriers of X-linked disorders as variable expressivity may result due to other genetic or environmental modifiers. Because the level of XCI may differ in prenatal specimens and whole blood, this test is not recommended for prenatal diagnosis. Diagnostic errors can occur due to rare sequence variations.



New Test 3003758 Zinc, Red Blood Cells ZN RBC

Click for Pricing

Methodology: Quantitative Inductively Coupled Plasma-Mass Spectrometry

Performed: Sun-Sat **Reported:** 1-4 days

Specimen Required: Collect: Royal Blue (EDTA).

Specimen Preparation: Centrifuge whole blood and separate RBCs from plasma within 2 hours of collection. Transfer 2 mL RBCs to an ARUP Trace Element-Free Transport Tube (ARUP supply #43116) available online through eSupply using ARUP ConnectTM or

contact ARUP Client Services at (800) 522-2787. (Min: 0.6 mL)

<u>Storage/Transport Temperature:</u> Room temperature. Also acceptable: Refrigerated. <u>Unacceptable Conditions:</u> Specimens collected in tubes other than royal blue (EDTA). Specimens transported in containers other than

Royal Blue (EDTA) tube or Trace Element-Free Transport Tube. Clotted or grossly hemolyzed specimens.

Stability (collection to initiation of testing): After separation from plasma: Ambient: 1 week; Refrigerated: 1 week; Frozen:

Unacceptable

Reference Interval: 794.0-1470.0 mcg/dL

Interpretive Data:

Zinc concentrations in RBCs reflect the intracellular stores and general homeostasis of Zinc. Results may be falsely low if RBCs in the submitted specimen are lysed or not promptly separated from plasma.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

CPT Code(s): 84630

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.

2012300 Zolpidem Urine with Reflex to Quantitation ZOLP RFX U

Specimen Required: Collect: Random urine.

Specimen Preparation: Transfer 4 mL urine with no additives or preservatives to an ARUP Standard Transport Tube. (Min: 2 mL)

Storage/Transport Temperature: Room temperature.

<u>Unacceptable Conditions:</u> Samples collected in tube with additives or preservatives.

Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 1 month



The following will be discontinued from ARUP's test menu on May 17, 2021. Replacement test options are supplied if applicable.

Test Number	Test Name	Refer To Replacement
2011902	Adrenoleukodystrophy, X- Linked (ABCD1) Sequencing	
<u>0051495</u>	Alpha Thalassemia (HBA1 and HBA2) 7 Deletions	Alpha Globin (HBA1 and HBA2) Deletion/Duplication (2011622)
0050043	Alpha-1-Microglobulin, Urine	Retinol Binding Protein (0050467)
<u>2011144</u>	Arginine:Glycine Amidinotransferase (GATM) Deficiency Sequencing	
2008471	ATP7A-Related Copper Transport Disorders (ATP7A) Sequencing and Deletion/Duplication	
2008652	c-MET by Immunohistochemistry	
2005559	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (<i>PLOD1</i>) Sequencing and Deletion/Duplication	
2005457	High-Specificity Antiphospholipid Antibodies, IgG and IgM	Cardiolipin Antibodies, IgG and IgM or Phosphatidylserine Antibodies, IgG and IgM (0099344 or 2006495)
<u>2014234</u>	Human Immunodeficiency Virus 1 (HIV-1) by Qualitative Transcription-Mediated Amplification (TMA)	Human Immunodeficiency Virus 1 (HIV-1) by Qualitative NAAT (3003760)
3001474	Human Immunodeficiency Virus 1 (HIV-1) Qualitative by NAAT, Whole Blood	Human Immunodeficiency Virus 1 (HIV-1) by Qualitative NAAT (3003760)
0040168	JAK2 Gene, V617F Mutation, Quantitative	JAK2 (V617F) Mutation by ddPCR, Quantitative (3003751)
2009302	Li-Fraumeni (TP53) Sequencing	
0080281	NMP22, Urine	
2004195	Noonan Syndrome (SOS1) Sequencing	
<u>2010464</u>	Propoxyphene and Metabolite, Serum or Plasma, Quantitative	Propoxyphene and Metabolite, Serum or Plasma (3003723)
<u>2010468</u>	Propoxyphene and Metabolite, Urine, Quantitative	Propoxyphene and Metabolite, Urine (3003726)
<u>2002722</u>	PTEN-Related Disorders (PTEN) Sequencing	
<u>0051378</u>	Rett Syndrome (MECP2), Full Gene Sequencing	
<u>2011704</u>	Smith-Lemli-Opitz Syndrome (DHCR7) Sequencing, Fetal	
<u>2002001</u>	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing	