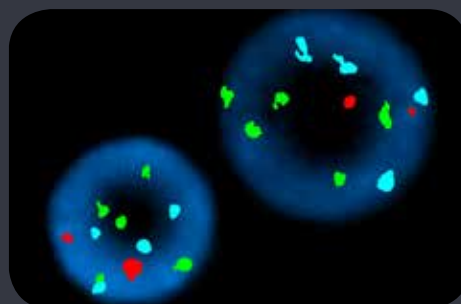




# molecular oncology services

PATIENTS.ANSWERS.RESULTS.



## ARUP LABORATORIES

As a nonprofit, academic institution of the University of Utah and its Department of Pathology, ARUP believes in collaborating, sharing knowledge, and contributing to laboratory science in ways that benefit our clients and their patients.

Our test menu is one of the broadest in the industry, encompassing more than 3,000 tests, including highly specialized and esoteric assays. We offer comprehensive testing in the areas of genetics, molecular oncology, pediatrics, and pain management, among others.

ARUP's clients include many of the nation's university teaching hospitals and children's hospitals, as well as multihospital groups, major commercial laboratories, and group purchasing organizations. We do not compete with our clients for physician office business, choosing instead to support clients' existing test menus by offering highly complex assays and accompanying consultative support so clients can provide exceptional patient care in their local communities.

Offering analytics, consulting, and decision support services, ARUP provides clients with the utilization management tools necessary to prosper in this time of value-based care. Our UM+ program helps clients control utilization, reduce costs, and improve patient care. In addition, ARUP is a worldwide leader in innovative laboratory research and development, led by the efforts of the ARUP Institute for Clinical and Experimental Pathology®.

ARUP's reputation for quality is supported by our ability to meet or exceed the requirements of multiple regulatory and accrediting agencies and organizations. ARUP participates in the CAP laboratory accreditation program and has CLIA certification through the Centers of Medicare and Medicaid Services. In December 2016, ARUP earned accreditation to the ISO 15189:2012 standard under CAP.

We believe in  
collaborating,  
sharing  
knowledge, and  
contributing  
to laboratory  
science in ways  
that provide  
the best value  
for the patient.  
Together,  
ARUP and  
its clients will  
improve patient  
care today and  
in the future.

**patients. answers. results.**

A laboratory test is more than a number; it is a person,  
an answer, a diagnosis.



## MOLECULAR ONCOLOGY SERVICES

Molecular diagnostics is an important component of clinical oncology, supplying pertinent information for diagnosis, prognosis, and prediction of response to tailored chemotherapeutic agents. ARUP Laboratories offers a wide range of molecular diagnostic tests designed to answer important clinical questions regarding diagnosis, prognosis, and pharmacogenetics. Using state-of-the-art methodologies, including fluorescence in situ hybridization (FISH), polymerase chain reaction (PCR), and next-generation sequencing analysis, ARUP Laboratories supplies pertinent clinical information for a variety of cancers.

**This brochure has been organized into two sections:**

- 1 Test Categories, which includes diagnostic markers, pharmacogenetic markers, and prognostic markers
- 2 Diagnostic Categories/Tumor Type

ARUP Laboratories is committed to supplying high-quality molecular diagnostic testing in a timely fashion and will continuously expand its test menu as new procedures and markers of clinical utility are identified.



# TEST CATEGORIES

## Diagnostic Markers

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
2008604	1p/19q Deletion by FISH	P	2002378	Eosinophilia Panel by FISH	WB, BM
2002647	Acute Lymphocytic Leukemia Panel by FISH, Adult	WB, BM	2002298	<i>ETV6-RUNX1 (TEL-AML1)</i> Fusion, t(12;21)(p13;q22) by FISH	WB, BM
2002719	Acute Lymphocytic Leukemia Panel by FISH, Pediatric	WB, BM	2002298	<i>EWSR1 (22q12)</i> Rearrangement by FISH	+
2011132	Acute Myeloid Leukemia Panel by FISH	WB, BM	2007225	<i>EWSR1 (22q12)</i> Gene Rearrangement by FISH	P
2002653	Acute Myelogenous Leukemia (AML) with Myelodysplastic Syndrome (MDS) or Therapy-Related AML by FISH	WB, BM	2004863	Familial Adenomatous Polyposis ( <i>APC</i> ) Sequencing	WB
2012710	Aggressive B-Cell Lymphoma FISH Reflex, Tissue	P	2004915	Familial Adenomatous Polyposis Panel: <i>APC</i> Sequencing, ( <i>APC</i> ) Sequencing and Deletion/Duplication, ( <i>MUTYH</i> ) 2 Mutations	WB
2006102	<i>ALK</i> Gene Rearrangements by FISH, Lung	P	2001961	Familial Mutation, Target Sequencing (HNPCC/Lynch Syndrome)	WB
2006193	B-Cell Clonality Screening (IgH and IgK) by PCR	WB, BM, FF	2002298	<i>FOXO1 (FKHR)</i> (13q13) Gene Rearrangement by FISH	+
2010107	<i>BCL6</i> (3q27) Gene Rearrangement by FISH	P	2001497	<i>FOXO1 (FKHR)</i> (13q14) Gene Rearrangement by FISH	P
2002298	<i>BCL6</i> Rearrangement (3q27)	WB, BM	2013449	Gastrointestinal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 16 Genes	WB
2002298	<i>BCR-ABL1</i> Fusion t(9;22)(q34;q11.2) by FISH	WB, BM	2002674	Gastrointestinal Stromal Tumor Mutation	P
2005017	<i>BCR-ABL1</i> Major (p210), Quantitative	WB, BM	2010757	Hereditary Cancer Panel, Deletion/Duplication, 36 Genes	WB
2005016	<i>BCR-ABL1</i> Minor (p190), Quantitative	WB, BM	2007167	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> , <i>SDHC</i> , and <i>SDHD</i> ) Sequencing and Deletion/Duplication Panel	WB
2005010	<i>BCR-ABL1</i> , Qualitative with Reflex to <i>BCR-ABL1</i> , Quantitative	WB, BM	2007108	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> ) Sequencing and Deletion/Duplication	WB
2002298	<i>MALT1</i> (18q21) gene rearrangement by FISH	WB, BM	2007117	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHC</i> ) Sequencing and Deletion/Duplication	WB
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	P	2007122	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHD</i> ) Sequencing and Deletion/Duplication	WB
0051750	<i>BRAF</i> Codon 600 Mutation Detection with Reflex to <i>MLH1</i> Promoter Methylation	P	0051650	HNPCC/Lynch Syndrome ( <i>MLH1</i> ) Sequencing and Deletion/Duplication	WB
2007132	<i>BRAF</i> V600E Mutation Detection in Hairy Cell Leukemia by Real-Time PCR, Quantitative	WB, BM	0051654	HNPCC/Lynch Syndrome ( <i>MSH2</i> ) Sequencing and Deletion/Duplication	WB
2010673	<i>CALR</i> (Calreticulin) Exon 9 Mutation Analysis by PCR	WB, BM	0051656	HNPCC/Lynch Syndrome ( <i>MSH6</i> ) Sequencing and Deletion/Duplication	WB
2002298	<i>CBFB</i> Rearrangement inv(16)(p13.3q22) by FISH	WB, BM	0051737	HNPCC/Lynch Syndrome ( <i>PMS2</i> ) Sequencing and Deletion/Duplication	WB
2011114	<i>CBFB-MYH11</i> inv(16) Detection, Quantitative	WB, BM	2001728	HNPCC/Lynch Syndrome Deletion/Duplication— <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , or <i>PMS2</i>	WB
2010188	Central Nervous System Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 15 Genes	WB	2002298	Hyperdiploidy with Trisomy 4 and 10 for Pediatric ALL	WB, BM
2002292	Chromosome Analysis, Bone Marrow	BM	2002298	<i>IGH</i> Rearrangement 14q32	WB, BM
2002300	Chromosome Analysis, Lymph Node	+	2001536	<i>IGH-BCL2</i> Fusion, t(14;18) by FISH	P
2002290	Chromosome Analysis, Leukemic Blood	WB	2002298	<i>IGH-BCL2</i> Fusion, t(14;18)(q32;q21) by FISH	WB, BM
2002296	Chromosome Analysis, Solid Tumor	+	2007226	<i>IGH-CCND1</i> Fusion, t(11;14) by FISH	P
2010229	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue—Oncology	P	2002298	<i>IGH-CCND1</i> Fusion, t(11;14)(q13;q32) by FISH	WB, BM
2006325	Cytogenomic SNP Microarray, Oncology	WB, BM			
2002298	<i>DDIT3 (CHOP)</i> (12q13) Gene Rearrangement by FISH	+, TP			
2007223	<i>DDIT3 (CHOP)</i> (12q13) Gene Rearrangement by FISH	P			
2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	P, FNA			
2010193	Endocrine Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 13 Genes	WB			

+ Fresh tissue, unfixed specimen. Use test code 2002298 to order and specify probe.

BM—bone marrow

FF—fresh frozen tissue

For more on ARUP Molecular Oncology, please visit

# TEST CATEGORIES

## Diagnostic Markers, continued

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
2001538	<i>IGH-MYC</i> Fusion, t(8;14) by FISH	P	2009318	<i>MYD88</i> L265P Mutation Detection by PCR, Quantitative	WB, BM, P
2002357	<i>JAK2</i> Exon 12 Mutation Analysis by PCR	WB, BM	2002528	Pancreatobiliary FISH	VARIABLES
0051245	<i>JAK2</i> Gene, V617F Mutation, Qualitative	WB, BM	2010102	PCA3—Prostate Cancer Biomarker	UR
2012085	<i>JAK2</i> Gene, V617F Mutation, Qualitative with Reflex to <i>JAK2</i> Exon 12 Mutation Analysis by PCR	WB, BM	2002298	<i>PDGFRA</i> Rearrangement 4q12 by FISH	WB, BM
2012084	<i>JAK2</i> Gene, V617F Mutation, Qualitative with Reflex to <i>CALR</i> (Calreticulin) Exon 9 Mutation Analysis by PCR with Reflex to <i>MPL</i> codon 515 Mutation Detection by Pyrosequencing, Quantitative	WB, BM	2002298	<i>PDGFRB</i> Rearrangement 5q33.1 by FISH	WB, BM
0040168	<i>JAK2</i> Gene, V617F Mutation, Quantitative	WB	2002871	<i>PML-RARA</i> Translocation, t(15;17) by RT-PCR, Quantitative	WB, BM
0051510	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing	WB	2002363	<i>PML-RARA</i> Translocation by FISH	BM
2001971	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing and Deletion/Duplication	WB	2002722	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing	WB
2004988	Juvenile Polyposis Syndrome ( <i>BMPRIA</i> ) Sequencing	WB	2002470	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing and Deletion/Duplication	WB
2004992	Juvenile Polyposis Syndrome ( <i>BMPRIA</i> ) Sequencing and Deletion/Duplication	WB	2010214	Renal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 15 Genes	WB
0040137	<i>KIT</i> (D816V) Mutation by PCR	WB, BM, P, FF	2002298	<i>RUNX1-RUNX1T1</i> ( <i>AML1-ETO</i> ) Fusion, t(8;21) (q22;q22) by FISH	WB, BM
2002437	<i>KIT</i> Mutations in AML by Fragment Analysis and Sequencing	WB, BM	2010138	<i>RUNX1-RUNX1T1</i> ( <i>AML1-ETO</i> ) t(8;21) Detection, Quantitative	WB, BM
2002695	<i>KIT</i> Mutations, Melanoma	P	2007222	<i>SS18</i> ( <i>SYT</i> ) (18q11) Gene Rearrangement by FISH	P
2009302	Li-Fraumeni ( <i>TP53</i> ) Sequencing	WB	2002298	<i>SS18</i> ( <i>SYT</i> ) Rearrangement by FISH	+, TP
2009313	Li-Fraumeni ( <i>TP53</i> ) Sequencing and Deletion/Duplication	WB	0055567	T-Cell Clonality Screening by PCR	WB, BM, FF, P
2010209	Melanoma Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 6 Genes	WB	2002298	<i>TCF3</i> ( <i>E2A</i> ) Rearrangement 19p13 by FISH	WB, BM
2009310	<i>MGMT</i> Methylation Detection by PCR	P	2001181	UroVysion FISH	UR
0051740	Microsatellite Instability (MSI), HNPCC/Lynch Syndrome by PCR	P	2002970	von Hippel-Lindau ( <i>VHL</i> ) Sequencing	WB
2002499	<i>MLH1</i> Promoter Methylation, Paraffin	P	2002965	von Hippel-Lindau ( <i>VHL</i> ) Sequencing and Deletion/Duplication	WB
2005545	<i>MPL</i> Codon 515 Mutation Detection by Pyrosequencing, Quantitative	WB			
2005359	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing	WB			
2005360	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing and Deletion/Duplication	WB			
0051390	Multiple Endocrine Neoplasia Type 2 ( <i>MEN2</i> ), <i>RET</i> Gene Mutations by Sequencing	WB			
2004911	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations	WB			
2006307	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations with Reflex to Sequencing	WB			
2006191	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) Sequencing	WB			
2002345	<i>MYC</i> (8q24) Gene Rearrangement by FISH	P			
2002298	<i>MYC</i> Rearrangement 8q24 by FISH	WB, BM			
2007227	<i>MYCN</i> ( <i>N-MYC</i> ) Gene Rearrangement by FISH	P			

FNA—FNA smear

P—paraffinized tissue

PL—plasma

TP—touch prep

WB—whole blood

UR—urine

# TEST CATEGORIES

## Pharmacogenetic and Targeted Therapy Markers

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
2007228	5-Fluorouracil (5-FU) Toxicity and Chemotherapeutic Response, 7 Mutations	WB	0040137	<i>KIT</i> (D816V) Mutation by PCR	WB, BM, P, FF
2006102	<i>ALK</i> Gene Rearrangements by FISH, Lung	P	2002695	<i>KIT</i> Mutations, Melanoma	P
2002298	<i>BCR-ABL1</i> Fusion, t(9;22)(q34;q11.2) by FISH	+	0040248	<i>KRAS</i> Mutation Detection	P
2008420	<i>BCR-ABL1</i> Mutation Analysis by Next-Generation Sequencing	WB, BM	2001932	<i>KRAS</i> Mutation Detection with Reflex to <i>BRAF</i> Codon 600 Mutation Detection	P
2013921	<i>BRAF</i> V600E Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB	2008894	Lung Cancer Panel	P
2011616	Colon Cancer Gene Panel, Somatic	P	2008895	Lung Cancer Panel with <i>KRAS</i>	P
0051104	Cytochrome P450 2C19 ( <i>CYP2C19</i> ) 9 Mutations	WB	0055655	Methylenetetrahydrofolate Reductase ( <i>MTHFR</i> ) 2 Mutations	WB
0051103	Cytochrome P450 2C9 ( <i>CYP2C9</i> ) 2 Mutations	WB	2003123	<i>NRAS</i> Mutation Detection by Pyrosequencing	P
2014547	Cytochrome P450 2D6 ( <i>CYP2D6</i> ) 15 Variants and Gene Duplication	WB	2002298	<i>PDGFRA</i> by FISH	WB, BM
2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	P, FNA	2002298	<i>PDGFRB</i> by FISH	WB, BM
2012868	<i>EGFR</i> T790M Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB	2002363	<i>PML-RARA</i> Translocation by FISH	WB, BM
2008603	<i>ERBB2</i> (HER-2/ <i>neu</i> ) Gene Amplification by FISH	P	2002871	<i>PML-RARA</i> Translocation, t(15;17) by RT-PCR, Quantitative	WB, BM
2014683	LeukoStrat CDx <i>FLT3</i> Mutation Detection by PCR	WB, BM	2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P, FNA
2002674	Gastrointestinal Stromal Tumor Mutation	P	0051332	UDP-Glucuronosyltransferase 1A1 ( <i>UGT1A1</i> ) Genotyping	WB

## Prognostic Markers

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
2012710	Aggressive B-Cell Lymphoma FISH Reflex, Tissue	P	2012032	Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes	WB
2002298	<i>BCR-ABL1</i> Fusion, t(9;22)(q34;q11.2) by FISH	WB, BM	2002298	<i>CBFB</i> Rearrangement inv(16)(p13.3q22) by FISH	WB, BM
2008420	<i>BCR-ABL1</i> Mutation Analysis by Next-Generation Sequencing	WB, BM	2011114	<i>CBFB-MYH11</i> inv(16) Detection, Quantitative	WB, BM
2005017	<i>BCR-ABL1</i> Major (p210), Quantitative	WB, BM	2004247	<i>CEBPA</i> Mutation Detection	WB, BM
2005016	<i>BCR-ABL1</i> Minor (p190), Quantitative	WB, BM	2002295	Chronic Lymphocytic Leukemia (CLL) Panel by FISH	WB, BM
2005010	<i>BCR-ABL1</i> Qualitative with Reflex to <i>BCR-ABL1</i> Quantitative	WB, BM	2008605	<i>EGFR</i> Gene Amplification by FISH	P
2011954	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing	WB	2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	P, FNA
2011949	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing and Deletion/Duplication	WB	2002378	Eosinophilia Panel by FISH	WB, BM
2012026	Breast and Ovarian Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 20 Genes	WB	2002298	<i>ETV6-RUNX1</i> ( <i>TEL-AML1</i> ) Fusion, t(12;21)(p13;q22) by FISH	WB, BM
2010673	<i>CALR</i> (Calreticulin) Exon 9 Mutation Analysis by PCR	WB, BM	2004863	Familial Adenomatous Polyposis ( <i>APC</i> ) Sequencing	WB
			2004915	Familial Adenomatous Polyposis Panel: ( <i>APC</i> ) Sequencing and Deletion/Duplication, ( <i>MUTYH</i> ) 2 Mutations	WB

+ Fresh tissue, unfixed specimen. Use test code 2002298 to order and specify probe.

BM—bone marrow

FF—fresh frozen tissue

For more on ARUP Molecular Oncology, please visit

# TEST CATEGORIES

## Prognostic Markers, continued

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
2001961	Familial Mutation, Targeted Sequencing (HNPCC/Lynch Syndrome)	WB	2006307	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations with Reflex to Sequencing	WB
2007167	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> , <i>SDHC</i> , and <i>SDHD</i> ) Sequencing and Deletion/Duplication Panel	WB	2006191	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) Sequencing	WB
2007108	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> ) Sequencing and Deletion/Duplication	WB	2004911	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations	WB
2007117	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHC</i> ) Sequencing and Deletion/Duplication	WB	2002709	Myelodysplastic Syndrome (MDS) Panel by FISH	BM, WB
2007122	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHD</i> ) Sequencing and Deletion/Duplication	WB	0040174	<i>NPM1</i> Mutation by PCR and Fragment Analysis	WB, BM, P
0051650	HNPCC/Lynch Syndrome ( <i>MLH1</i> ) Sequencing and Deletion/Duplication	WB	2010102	PCA3—Prostate Cancer Biomarker by Transcription-Mediated Amplification	UR
0051654	HNPCC/Lynch Syndrome ( <i>MSH2</i> ) Sequencing and Deletion/Duplication	WB	2002363	<i>PML-RAR<math>\alpha</math></i> FISH	WB, BM
0051656	HNPCC/Lynch Syndrome ( <i>MSH6</i> ) Sequencing and Deletion/Duplication	WB	2002871	<i>PML-RARA</i> Translocation, t(15;17) by RT-PCR, Quantitative	WB, BM
0051737	HNPCC/Lynch Syndrome ( <i>PMS2</i> ) Sequencing and Deletion/Duplication	WB	2010248	Prosigna Breast Cancer Prognostic Gene Signature	P
2006444	<i>IDH1</i> and <i>IDH2</i> Mutation Analysis, Exon 4	WB, BM	2002298	<i>RUNX1-RUNX1T1</i> ( <i>AML1-ETO</i> ) Fusion, t(8;21) (q22;q22) by FISH	WB, BM
2014188	<i>IDH1</i> and <i>IDH2</i> Mutation Analysis, Exon 4, Formalin-Fixed, Paraffin-Embedded (FFPE) Tissue	P	2010138	<i>RUNX1-RUNX1T1</i> ( <i>AML1-ETO</i> ) t(8;21) Detection, Quantitative	WB, BM
0040227	<i>IGHV</i> Mutation Analysis by Sequencing	WB, BM	2002298	<i>TCF3</i> ( <i>E2A</i> ) Rearrangement	WB, BM
2002437	<i>KIT</i> Mutations in AML by Fragment Analysis and Sequencing	WB, BM	2005766	<i>WT1</i> Mutations by Sequencing	WB, BM
2014683	LeukoStrat CDx <i>FLT3</i> Mutation Detection by PCR	WB, BM			
2009302	Li-Fraumeni ( <i>TP53</i> ) Sequencing	WB			
2009313	Li-Fraumeni ( <i>TP53</i> ) Sequencing and Deletion/Duplication	WB			
2013082	<i>MET</i> Gene Amplification by FISH	P			
0051740	Microsatellite Instability (MSI), HNPCC/Lynch Syndrome by PCR	P			
2002298	<i>MLL</i> Rearrangement 11q23 by FISH	WB, BM			
2005359	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing	WB			
2005360	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing and Deletion/Duplication	WB			
0051390	Multiple Endocrine Neoplasia Type 2 ( <i>MEN2</i> ), <i>RET</i> Gene Mutations by Sequencing	WB			
2002294	Multiple Myeloma Panel by FISH	WB, BM			

FNA—FNA smear

P—paraffinized tissue

PL—plasma

TP—touch prep

WB—whole blood

UR—urine

## DIAGNOSTIC CATEGORIES/TUMOR TYPE

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
<b>Alveolar Rhabdomyosarcoma</b>					
2002298	<i>FKHR (FOXO1)</i> 13q13 by FISH	+	0051650	HNPCC/Lynch Syndrome ( <i>MLH1</i> ) Sequencing and Deletion/Duplication	WB
2001497	<i>FOXO1 (FKHR)</i> (13q14) Gene Rearrangement by FISH	P	0051654	HNPCC/Lynch Syndrome ( <i>MSH2</i> ) Sequencing and Deletion/Duplication	WB
<b>Bladder Cancer (Urothelial Carcinoma)</b>					
2001181	UroVysion FISH	UR	0051656	HNPCC/Lynch Syndrome ( <i>MSH6</i> ) Sequencing and Deletion/Duplication	WB
<b>Breast Cancer (Breast Carcinoma)</b>					
2011954	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing	WB	0051737	HNPCC/Lynch Syndrome ( <i>PMS2</i> ) Sequencing and Deletion/Duplication	WB
2011949	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing and Deletion/Duplication	WB	2001728	HNPCC/Lynch Syndrome Deletion/Duplication— <i>MLH1, MSH2, MSH6, or PMS2</i>	WB
2012026	Breast and Ovarian Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 20 Genes	WB	0051510	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing	WB
2008603	<i>ERBB2 (HER-2/neu)</i> Gene Amplification by FISH	P	2001971	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing and Deletion/Duplication	WB
2010248	Prosigna Breast Cancer Prognostic Gene Signature	P	2004988	Juvenile Polyposis Syndrome ( <i>BMPR1A</i> ) Sequencing	WB
2002722	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing	WB	2004992	Juvenile Polyposis Syndrome ( <i>BMPR1A</i> ) Sequencing and Deletion/Duplication	WB
2002470	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing and Deletion/Duplication	WB	0040248	<i>KRAS</i> Mutation Detection	P
2009302	Li-Fraumeni ( <i>TP53</i> ) Sequencing	WB	2001932	<i>KRAS</i> Mutation Detection with Reflex to <i>BRAF</i> Codon 600 Mutation Detection	P
2009313	Li-Fraumeni ( <i>TP53</i> ) Sequencing and Deletion/Duplication	WB	2009302	Li-Fraumeni ( <i>TP53</i> ) Sequencing	WB
2008394	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing	WB	2009313	Li-Fraumeni ( <i>TP53</i> ) Sequencing and Deletion/Duplication	WB
2008398	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing and Deletion/Duplication	WB	0051740	Microsatellite Instability (MSI), HNPCC/Lynch Syndrome by PCR	P
<b>Colon Cancer (Colonic Adenocarcinoma)</b>					
2007228	5-Fluorouracil (5-FU) Toxicity and Chemotherapeutic Response, 7 Mutations	WB	2002327	Mismatch Repair by IHC with Reflex to <i>BRAF</i> Codon 600 Mutation and <i>MLH1</i> Promoter Methylation	P
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	P	2002499	<i>MLH1</i> Promoter Methylation, Paraffin	P
2013921	<i>BRAF</i> V600E Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB	2006307	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations with Reflex to Sequencing	WB
0051750	<i>BRAF</i> Codon 600 Mutation Detection with Reflex to <i>MLH1</i> Promoter Methylation	P	2006191	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) Sequencing	WB
2010757	Cancer Panel, Hereditary, Deletion/Duplication, 46 Genes	WB	2004911	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations	WB
2012032	Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes	WB	2003123	<i>NRAS</i> Mutation Detection by Pyrosequencing	P
2011616	Colon Cancer Gene Panel, Somatic	P	2008394	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing	WB
2013906	Epi proColon	WB	2008398	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing and Deletion/Duplication	WB
2004863	Familial Adenomatous Polyposis ( <i>APC</i> ) Sequencing	WB	2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P, FNA
2004915	Familial Adenomatous Polyposis Panel: ( <i>APC</i> ) Sequencing and Deletion/Duplication, ( <i>MUTYH</i> ) 2 Mutations	WB	0051332	UDP-Glucuronosyltransferase 1A1 ( <i>UGT1A1</i> ) Genotyping	WB
2001961	Familial Mutation, Targeted Sequencing (HNPCC/Lynch Syndrome)	WB	<b>CNS/ Renal Cell Carcinoma/ Pheochromocytoma</b>		
2013449	Gastrointestinal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 16 genes	WB	2010188	Central Nervous System Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 15 Genes	WB
			2002970	von Hippel-Lindau ( <i>VHL</i> ) Sequencing	WB
			2002965	von Hippel-Lindau ( <i>VHL</i> ) Sequencing and Deletion/Duplication	WB

+ Fresh tissue, unfixed specimen. Use test code 2002298 to order and specify probe.

BM—bone marrow

FF—fresh frozen tissue



# DIAGNOSTIC CATEGORIES/TUMOR TYPE

Test #	Test Name	Specimen Type
<b>Endometrial Carcinoma</b>		
2005270	Mismatch Repair by IHC with Reflex to <i>MLH1</i> Promoter Methylation	P
<b>Ewing Sarcoma</b>		
2007225	<i>EWSR1</i> (22q12) Gene Rearrangement by FISH	P
2002298	<i>EWSR1</i> Rearrangement by FISH	+
<b>Gastrointestinal Stromal Tumor (GIST)</b>		
2002674	Gastrointestinal Stromal Tumor Mutation	P
2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P
<b>Glioblastoma</b>		
2008605	<i>EGFR</i> Gene Amplification by FISH	P
2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	P, FNA
2014188	<i>IDH1</i> and <i>IDH2</i> Mutation Analysis, Exon 4	P
2009310	<i>MGMT</i> Methylation Detection by PCR	P
<b>Lung Carcinoma</b>		
2006102	<i>ALK</i> Gene Rearrangements by FISH, Lung	P
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	P
2013921	<i>BRAF</i> V600E Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB
2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	P, FNA
2012868	<i>EGFR</i> T790M Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB
0040248	<i>KRAS</i> Mutation Detection	P
2008894	Lung Cancer Panel	P
2008895	Lung Cancer Panel with <i>KRAS</i>	P
2013082	<i>MET</i> Gene Amplification by FISH	P
2012654	<i>RET</i> Gene Rearrangements by FISH	P
2008418	<i>ROS1</i> by FISH	P
2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P, FNA
<b>Melanoma</b>		
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	P
2013921	<i>BRAF</i> V600E Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB
2010757	Cancer Panel, Hereditary, Deletion/Duplication, 46 Genes	WB
2002695	<i>KIT</i> Mutations, Melanoma	P
2010209	Melanoma Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 6 Genes	WB
2003123	<i>NRAS</i> Mutation Detection by Pyrosequencing	P
2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P, FNA

Test #	Test Name	Specimen Type
<b>Neuroblastoma</b>		
2007227	<i>MYCN</i> ( <i>N-Myc</i> ) Gene Amplification by FISH	P
<b>Oligodendroglioma</b>		
2008604	1p/19q Deletion by FISH	P
<b>Paraganglioma/Pheochromocytoma</b>		
2007167	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> , <i>SDHC</i> , and <i>SDHD</i> ) Sequencing and Deletion/Duplication Panel	WB
2007108	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> ) Sequencing and Deletion/Duplication	WB
2007117	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHC</i> ) Sequencing and Deletion/Duplication	WB
2007122	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHD</i> ) Sequencing and Deletion/Duplication	WB
<b>Parathyroid/Pituitary/Pancreatic</b>		
2005359	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing	WB
2005360	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing and Deletion/Duplication	WB
<b>Round Cell/Myxoid Liposarcoma</b>		
2002298	<i>DDIT3</i> ( <i>CHOP</i> ) (12q13) Gene Rearrangement by FISH	TOUCH PREP, +
2007223	<i>DDIT3</i> ( <i>CHOP</i> ) (12q13) Gene Rearrangement by FISH	P
2003016	<i>MDM2</i> Gene Amplification by FISH	P
<b>Synovial Sarcoma</b>		
2007222	<i>SS18</i> ( <i>SYT</i> ) (18q11) Gene Rearrangement by FISH	P
<b>Systemic Mastocytosis</b>		
0040137	<i>KIT</i> (D816V) Mutation by PCR	WB, BM, FF, P
<b>Thyroid Carcinoma</b>		
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	P
2013921	<i>BRAF</i> V600E Mutation Detection in Circulating Tumor DNA by Digital Droplet PCR	WB
0051390	Multiple Endocrine Neoplasia Type 2 ( <i>MEN2</i> ), <i>RET</i> Gene Mutations by Sequencing	WB
2012654	<i>RET</i> Gene Rearrangements by FISH	P
2007991	Solid Tumor Mutation Panel by Next-Generation Sequencing	P, FNA

FNA—FNA smear

P—paraffinized tissue

PL—plasma

TP—touch prep

WB—whole blood

UR—urine

# LEUKEMIA/LYMPHOMA

Test *	Test Name	Specimen Type	Test *	Test Name	Specimen Type
<b>Acute Lymphoblastic Leukemia (ALL)</b>					
2002647	Acute Lymphocytic Leukemia (ALL) Panel by FISH, Adult	BM, WB	0040174	<i>NPM1</i> Mutation by PCR and Fragment Analysis	WB, BM, FF, P
2002719	Acute Lymphocytic Leukemia (ALL) Panel by FISH, Pediatric	BM, WB	2010138	<i>RUNX1-RUNX1T1 (AML1-ETO)</i> t(8;21) Detection, Quantitative	WB, BM
2002298	<i>BCR-ABL1</i> Fusion, t(9;22)(q34;q11.2) by FISH	WB, BM	2002298	<i>RUNX1-RUNX1T1 (AML1-ETO)</i> Fusion, t(8;21)(q22;q22) by FISH	WB, BM
2005010	<i>BCR-ABL1</i> Qualitative with Reflex to <i>BCR-ABL1</i> Quantitative	WB, BM	2005766	<i>WT1</i> Mutations by Sequencing	WB, BM
2005016	<i>BCR-ABL1</i> Minor (p190), Quantitative	WB, BM	<b>Acute Promyelocytic Leukemia (APL)</b>		
2008420	<i>BCR-ABL1</i> Mutation Analysis by Next-Generation Sequencing	WB, BM	2002363	<i>PML-RARA</i> Translocation by FISH	WB, BM
2002298	<i>CDKN2</i> p16 Deletion 9p21 by FISH	WB, BM	2002871	<i>PML-RARA</i> Translocation, t(15;17) by RT-PCR, Quantitative	WB, BM
2007130	Chromosome Analysis, Bone Marrow with Reflex to Genomic Microarray	BM	<b>B- and T-Cell Markers Lymphoma</b>		
2007131	Chromosome Analysis, Leukemic Blood with Reflex to Genomic Microarray	WB	2006193	B-Cell Clonality Screening (IgH and IgK) by PCR	WB, BM, FF
2006325	Cytogenomic SNP Microarray—Oncology	BM, WB	2009318	<i>MYD88</i> L265P Mutation Detection by PCR, Quantitative	WB, BM, P
2002298	<i>ETV6-RUNX1 (TEL-AML1)</i> Fusion, t(12;21)(p13;q22) by FISH	WB, BM	0055567	T-Cell Clonality Screening by PCR	WB, BM, FF, P
2002298	Hyperdiploidy with Trisomy 4 and 10	WB, BM	<b>Burkitt Lymphoma</b>		
2002298	<i>IGH</i> Rearrangement 14q32 by FISH	WB, BM	2012710	Aggressive B-Cell Lymphoma FISH Reflex, Tissue	P
0055655	Methylenetetrahydrofolate Reductase ( <i>MTHFR</i> ) 2 Mutations	WB	2010107	<i>BCL6</i> (3q27) Gene Rearrangement by FISH	P
2002298	<i>MLL</i> Rearrangement 11q23 by FISH	WB, BM	2001538	<i>IGH-MYC</i> Fusion t(8;14) by FISH	P
2002298	<i>TCF3 (E2A)</i> Rearrangement 19p13 by FISH	WB, BM	2002345	<i>MYC</i> (8q24) Gene Rearrangement by FISH	P
<b>Acute Myelogenous Leukemia (AML)</b>			2002298	<i>MYC</i> Rearrangement 8q24 by FISH	WB, BM
2011132	Acute Myeloid Leukemia Panel by FISH	WB, BM	<b>Chronic Lymphocytic Leukemia (CLL)</b>		
2002653	Acute Myelogenous Leukemia (AML) with Myelodysplastic Syndrome (MDS) or Therapy-Related AML by FISH	WB, BM	2002295	Chronic Lymphocytic Leukemia (CLL) Panel by FISH	WB, BM
2012222	Bone Marrow Failure Sequencing, 35 Genes	WB	0040227	<i>IGHV</i> Mutation Analysis by Sequencing	WB, BM
2002298	<i>CBFB</i> Rearrangement inv(16)(p13.3q22) by FISH	WB, BM	<b>Chronic Myelogenous Leukemia (CML)</b>		
2011114	<i>CBFB-MYH11</i> inv(16) Detection, Quantitative	WB, BM	2002298	<i>BCR-ABL1</i> Fusion, t(9;22)(q34;q11.2) by FISH	WB, BM
2004247	<i>CEBPA</i> Mutation Detection	WB, BM	2008420	<i>BCR-ABL1</i> Mutation Analysis by Next-Generation Sequencing	WB, BM
2007130	Chromosome Analysis, Bone Marrow with Reflex to Genomic Microarray	BM	2005017	<i>BCR-ABL1</i> Major (p210), Quantitative	WB, BM
2006325	Cytogenomic SNP Microarray—Oncology	BM, WB	2005016	<i>BCR-ABL1</i> Minor (p190), Quantitative	WB, BM
2006444	<i>IDH1</i> and <i>IDH2</i> Mutation Analysis, Exon 4	WB, BM	2005010	<i>BCR-ABL1</i> Qualitative with Reflex to <i>BCR-ABL1</i> Quantitative	WB, BM
2002437	<i>KIT</i> Mutations in AML by Fragment Analysis and Sequencing	WB, BM	0055655	Methylenetetrahydrofolate Reductase ( <i>MTHFR</i> ) 2 Mutations	WB
2014683	LeukoStrat CDx <i>FLT3</i> Mutation Detection by PCR	WB, BM	<b>Follicular Lymphoma/Diffuse Large-Cell Lymphoma</b>		
2002298	<i>MLL</i> Rearrangement 11q23 by FISH	WB, BM	2012710	Aggressive B-Cell Lymphoma FISH Reflex, Tissue	P
2011117	Myeloid Malignancies Panel by Next-Generation Sequencing	WB, BM	2002298	<i>BCL6</i> Rearrangement (3q27) by FISH	WB, BM
2012182	Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel	WB, BM	2010107	<i>BCL6</i> (3q27) Gene Rearrangement by FISH	P
			2002298	<i>IGH-BCL2</i> Fusion, t(14;18)(q32;q21) by FISH	WB, BM
			2001536	<i>IGH-BCL2</i> Fusion, t(14;18) by FISH	P

+ Fresh tissue, unfixed specimen. Use test code 2002298 to order and specify probe.

BM—bone marrow

FF—fresh frozen tissue

For more on ARUP Molecular Oncology, please visit

# LEUKEMIA/LYMPHOMA

Test #	Test Name	Specimen Type	Test #	Test Name	Specimen Type
<b>Hairy Cell Leukemia</b>			<b>Myeloproliferative Neoplasms (MPN)</b>		
2007132	<i>BRAF</i> V600E Mutation Detection in Hairy Cell Leukemia by Real-Time PCR, Quantitative	WB, BM	2010673	<i>CALR</i> (Calreticulin) Exon 9 Mutation Analysis by PCR	WB, BM
<b>Lymphoproliferative Disorders (LPD)</b>			2002378	Eosinophilia Panel by FISH	WB, BM
2002650	Lymphoma (Aggressive) Panel by FISH	BM, WB	2002357	<i>JAK2</i> Exon 12 Mutation Analysis by PCR	BM, WB
2002298	Trisomy 12 by FISH	WB, BM	0051245	<i>JAK2</i> Gene, V617F Mutation, Qualitative	BM, WB
<b>Mantle Cell Non-Hodgkins Lymphoma</b>			0040168	<i>JAK2</i> Gene, V617F Mutation, Quantitative	WB
2002298	<i>IGH</i> Rearrangement by FISH	WB, BM	2012085	<i>JAK2</i> Gene, V617F Mutation, Qualitative with Reflex to <i>JAK2</i> Exon 12 Mutation Analysis by PCR	WB, BM
2007226	<i>IGH-CCND1</i> Fusion, t(11;14) by FISH	P	2012084	<i>JAK2</i> Gene, V617F Mutation, Qualitative with Reflex to <i>CALR</i> (Calreticulin) Exon 9 Mutation Analysis by PCR with Reflex to <i>MPL</i> codon 515 Mutation Detection by Pyrosequencing, Quantitative	WB, BM
2002298	<i>IGH-CCND1</i> Fusion, t(11;14)(q13;q32) by FISH	WB, BM			
<b>Marginal Zone B-Cell Lymphoma</b>			2005545	<i>MPL</i> Codon 515 Mutation Detection by Pyrosequencing, Quantitative	WB
2002298	<i>MALT1</i> (18q21) gene rearrangement by FISH	BM, WB	2011117	Myeloid Malignancies Mutation Panel by Next Generation Sequencing	WB, BM
<b>Multiple Myeloma</b>			2012182	Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel	WB, BM
2002294	Multiple Myeloma Panel by FISH	BM, WB	2002360	Myeloproliferative Disorders Panel by FISH	BM, WB
<b>Myelodysplastic Syndrome (MDS)</b>			2002298	<i>PDGFRA-FIP1L1</i> Fusion by FISH ( <i>CHIC2</i> Deletion)	WB, BM
2002298	20q Deletion ( <i>D20S108</i> ) del(20)(q12) by FISH	WB, BM	2002298	<i>PDGFRB</i> Rearrangement 5q33.1 by FISH	WB, BM
2002298	5q Deletion ( <i>EGR1</i> )/Monosomy 5 del(5)(q31)/-5 by FISH	WB, BM	2002298	Trisomy 8 by FISH	WB, BM
2002298	7q Deletion ( <i>D7S486</i> )/Monosomy 7 del(7)(q31)/-7 by FISH	WB, BM	2002298	Trisomy 9 by FISH	WB, BM
2012222	Bone Marrow Failure Sequencing, 35 Genes	WB	<b>Primary Effusion Lymphoma</b>		
2010229	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue—Oncology	P	2002902	Epstein-Barr Virus (EBV) by in situ Hybridization, Paraffin	P
2002709	Myelodysplastic Syndrome (MDS) Panel by FISH	BM, WB			
2011117	Myeloid Malignancies Panel by Next-Generation Sequencing	WB, BM			
2012182	Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel	WB, BM			
2002298	Trisomy 8 by FISH	WB, BM			

FNA—FNA smear

P—paraffinized tissue

PL—plasma

TP—touch prep

WB—whole blood

UR—urine



[www.aruplab.com](http://www.aruplab.com) | [www.arupconsult.com](http://www.arupconsult.com)

**ARUP LABORATORIES**

500 Chipeta Way  
Salt Lake City, UT 84108-1221  
Phone: (800) 522-2787  
Fax: (801) 583-2712  
[www.aruplab.com](http://www.aruplab.com)

*ARUP is a nonprofit enterprise of the University of Utah  
and its Department of Pathology.*

© 2017 ARUP Laboratories  
BD-TS-026, Rev 6, October 2017

