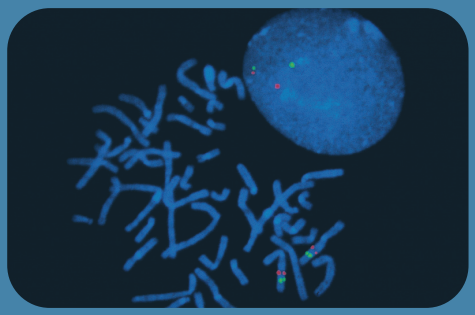
ARUP LABORATORIES | Genetics

genetic testing

PATIENTS.ANSWERS.RESULTS.®



aruplab.com/geneticstesting

SEPTEMBER 2021

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 believe that healthcare should be delivered as close to the patient as possible, which is why we support your efforts to be the principal healthcare provider in the communities you serve by offering highly complex assays and accompanying consultative support.

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.®

ISO 15189
CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

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PRENATAL ANEUPLOIDY SCREENING

Maternal serum screening (MSS) testing at ARUP is offered to help identify pregnancies at increased risk for Down syndrome, trisomy 18, or open neural tube defects such as spina bifida. ARUP offers all MSS testing recommended by the American College of Obstetrics and Gynecology. Second-trimester-only tests measure the levels of specific protein markers in maternal blood to predict risk. First-trimester-only and cross-trimester tests combine fetal ultrasound measurements* along with measurements of biochemical markers in maternal blood to predict risk.

* Sonographer providing NT measurement must be certified by NTQR (SMFM) or FMF. Please contact a genetic counselor at (800) 242-2787, ext. 2141, prior to sending first sample to ensure acceptability.

TEST #	TEST NAME/DESCRIPTION
Combined First- and Second-Trimester Screening	
3000146	Maternal Screening, Sequential, Specimen #1, hCG, PAPP-A, NT
3000148	Maternal Screening, Sequential, Specimen #2, Alpha Fetoprotein, hCG, Estriol, and Inhibin A
3000147	Maternal Serum Screening, Integrated, Specimen #1, PAPP-A, NT
3000149	Maternal Serum Screening, Integrated, Specimen #2, Alpha Fetoprotein, hCG, Estriol, and Inhibin A
First-Trimester Screening	
3000145	Maternal Serum Screen, First Trimester, hCG, PAPP-A, NT
Second-Trimester Screening	
3000144	Maternal Serum Screen, Alpha Fetoprotein
3000143	Maternal Serum Screen, Alpha Fetoprotein, hCG, Estriol, and Inhibin A (Quad)
3000142	Alpha Fetoprotein (Amniotic Fluid) with Reflex to Acetylcholinesterase and Fetal Hemoglobin

TEST #	TEST NAME/DESCRIPTION
Noninvasive Prenatal Screening	
2007537	Non-Invasive Prenatal Testing for Fetal Aneuploidy
2010232	Non-Invasive Prenatal Testing for Fetal Aneuploidy with Microdeletions
2013142	Non-Invasive Prenatal Testing for Fetal Aneuploidy with 22q11.2 Microdeletion

BIOCHEMICAL GENETICS

The Biochemical Genetics Laboratory performs testing on a variety of biological specimens to aid in the identification of aminoacidopathies, organic acidemias, fatty acid oxidation disorders, and other inherited metabolic diseases. Early identification of a metabolic disorder may prevent death, as well as other serious health problems.

Available tests include amino acid quantitation, assessment of organic acids, acylcarnitine profile, and various other assays to diagnose suspected inborn errors of metabolism or confirm abnormal newborn screen results.

Consultation with ARUP's genetic counselors or medical directors is available.

TEST #	TEST NAME/DESCRIPTION
0040033	Acylcarnitine Quantitative Profile, Plasma
0081170	Acylglycines, Quantitative, Urine
2011415	Alpha-Iduronidase Enzyme Activity in Leukocytes
0080137	Amino Acids Quantitative by LC-MS/MS, CSF
2009389	Amino Acids Quantitative by LC-MS/MS, Plasma
2009419	Amino Acids Quantitative by LC-MS/MS, Urine
2014314	Autism and Intellectual Disability Comprehensive Panel
2014312	Autism and Intellectual Disability Metabolic Panel
0092610	Bile Acids, Fractionated and Total by LC-MS/MS
0093362	Biotinidase, Serum (with Paired Normal Control)
0080068	Carnitine, Free & Total (Includes Carnitine, Esterified)
0081308	Carnitine, Free and Total, Urine
0081110	Carnitine Panel (Free and Total Carnitine, Acylcarnitine), Plasma or Serum
2002328	Creatine Disorders Panel, Plasma or Serum
2002333	Creatine Disorders Panel, Urine
0081106	Cystine Quantitative, Urine
0081105	Cystinuria Panel
0080351	Ehlers-Danlos Syndrome Type VI Screen, Urine
3003086	Fatty Acids Profile, Essential in Red Blood Cells
2013518	Fatty Acids Profile, Essential Serum or Plasma
0081296	Galactose-1-Phosphate in Red Blood Cells
3001790	Galactose-1-Phosphate Uridyltransferase (GALT Enzyme), RBC
0051175	Galactosemia (GALT) Enzyme Activity and 9 Mutations
2014459	Gaucher Disease (GBA), Enzyme Activity in Leukocytes
2001510	Glutarylcarnitine Quantitative, Urine
2008129	Hexosaminidase A Percent and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A Percent and Total Hexosaminidase in Leukocytes
2008125	Hexosaminidase A and Total Hexosaminidase, Leukocytes
2008121	Hexosaminidase A Percent and Total Hexosaminidase, Plasma or Serum
2012259	Keratan Sulfate, Quantitative by LC-MS/MS, Urine
2012266	Lysosomal Acid Lipase Activity, Dried Blood Spot
2005255	Methylmalonic Acid, Serum or Plasma (Metabolic Disorders)
0081352	Mucopolysaccharides Screen - Electrophoresis and Quantitation, Urine
0081357	Mucopolysaccharides, Quantitative, Urine
3003566	Mucopolysaccharidoses Type 1/2, Total Heparan Sulfate and NRE (Sensi-Pro®) Quantitative, Serum or Plasma
3003552	Mucopolysaccharidoses Type 1/2, Total Heparan Sulfate and NRE (Sensi-Pro®) Quantitative, Urine
3003487	Mucopolysaccharidoses Type 4A/6 Total Chondroitin Sulfate and Dermatan Sulfate with NRE (Sensi-Pro®) Quantitative, Serum
3003539	Mucopolysaccharidoses Type 4A/6 Total CS-DS and NRE (Sensi-Pro®) Quantitative, Urine
3000704	Orotic Acid, Urine
0080336	Phenylalanine and Tyrosine, Plasma *monitoring only*
0080315	Phenylalanine Monitoring, Plasma *monitoring only*
2007406	Pipecolic Acid, Serum or Plasma
2008131	Pipecolic Acid, Urine
2014463	Pompe Disease (GAA), Enzyme Activity in Leukocytes
0080342	Pyridinoline and Deoxypyridinoline by HPLC
2013352	Pyridoxine-Dependent Epilepsy Panel, Serum or Plasma
2013355	Pyridoxine-Dependent Epilepsy Panel, Urine
2007401	Succinylacetone, Quantitative, Urine
0080355	Tyrosine, Plasma *monitoring only*
2004250	Very Long-Chain and Branched-Chain Fatty Acids Profile

CYTOGENETICS AND GENOMIC MICROARRAY

ARUP performs microarray, chromosome, and FISH analysis for both constitutional and cancer diagnoses. Patients with indications such as developmental delay, autism, recurrent fetal loss, and multiple congenital anomalies have the option of tests ranging from classic karyotype analysis, FISH studies for specific microdeletion/microduplication syndromes, or microarray.

Patients with confirmed or suspected cancer diagnoses have microarray, chromosome, and FISH analyses available to them. These studies may help determine the specific type of cancer present, predict disease course, determine a course of treatment, and enable physicians to monitor treatment effectiveness and look for residual disease following treatment.

TEST #	TEST NAME/DESCRIPTION
Constitutional/Postnatal Testing	
0040208	Aneuploidy Panel by FISH
2002289	Chromosome Analysis, Consitutional Peripheral Blood
2005763	Chromosome Analysis, Constitutional Blood, with Reflex to Genomic Microarray
2002288	Chromosome Analysis, Products of Conception
2005762	Chromosome Analysis, Products of Conception, with Reflex to Genomic Microarray
2002287	Chromosome Analysis, Rule Out Mosaicism
2002286	Chromosome Analysis, Skin Biopsy
2002299	Chromosome FISH, Metaphase
0097688	Chromosome Analysis - Breakage, Fanconi Anemia, Whole Blood
2003414	Cytogenomic SNP Microarray
2006267	Cytogenomic SNP Microarray Buccal Swab
2009353	Cytogenomic SNP Microarray with Five-Cell Chromosome Study, Constitutional Blood
2014314	Autism and Intellectual Disability Comprehensive Panel
2010795	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue - Products of Conception
2005633	Genomic SNP Microarray, Products of Conception

TEST #	TEST NAME/DESCRIPTION
Oncology	
2002292	Chromosome Analysis, Bone Marrow
2007130	Chromosome Analysis, Bone Marrow with Reflex to Genomic Microarray
2002290	Chromosome Analysis, Leukemic Blood
2007131	Chromosome Analysis, Leukemic Blood with Reflex to Genomic Microarray
2002300	Chromosome Analysis, Lymph Node
2002296	Chromosome Analysis, Solid Tumor
2010229	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue - Oncology
2006325	Cytogenomic SNP Microarray - Oncology
Prenatal Testing	
0040203	Chorionic Villus, FISH
2002297	Chromosome FISH, Prenatal
2002293	Chromosome Analysis, Amniotic Fluid
2008367	Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray
2002291	Chromosome Analysis, Chorionic Villus
2002288	Chromosome Analysis, Products of Conception
2005762	Chromosome Analysis, Products of Conception, with Reflex to Genomic Microarray
2011130	Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray
2011131	Chromosome FISH, Chorionic Villus with Reflex to Chromosome Analysis or Genomic Microarray
2010795	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue—Products of Conception
2002366	Cytogenomic SNP Microarray - Fetal

CONSTITUTIONAL FISH PROBES		
Suspected Diagnosis	**Probe Target**	**Gene(s)/Unique Sequence**
Aneuploidy, Common (ARUP test code 0040208)	13/18/21/X/Y	
Microdeletion Syndromes—order test 2002299 and specify probes		
4p-	4p16.3	*WHSC1*
5p-	5p15.2	D5S23-D5S721
15q11.2-13 duplication	15q11.2-13	D15S11, D15S10
22qter deletion	22q13.3	22qtel (*SHANK3*)
Angelman	15q11.2-13	D15S10
Cri-du-chat	5p15.2	D5S23-D5S721
DiGeorge	22q11.2	*TUPLE-1* (*HIRA*)
Kallmann	Xp22.3	*KAL1*
Male detection (SRY)	Yp11.3	*SRY*
Miller-Dieker (lissencephaly	17p13.3	*LIS1*
Phelan-McDermid	22q13.3	22qtel (*SHANK3*)
Prader-Willi	15q11.2-13	D15S10
SHOX	Xp22.3	*SHOX*
Smith-Magenis	17p11.2	*SHMT1, TOP3, FL11, LLGL1*
SRY	Yp11.3	*SRY*
Steroid sulfatase deficiency (STS, X-linked ichthyosis)	Xp22.3	*STS*
Velocardiofacial (VCF)	22q11.2	*TUPLE-1* (*HIRA*)
Williams (elastin)	7q11.23	*ELN, LIMK1*, D7S613
Wolf-Hirschhorn	4p16.3	*WHSC1*
Miscellaneous (Please contact the lab prior to ordering)—order test 2002299 or 2002298 and specify probes		
Suspected Diagnosis	**Probe Target**	**Gene(s)/Unique Sequence**
Acrocentric p-arm		NOR regions of all acrocentric chromosomes
X centromere	Xcen	DXZ1
X inactivation locus	Xq13	*XIST*
Y centromere	Ycen	DYZ3
Yp11.3	Yp11.3	*SRY*
Yq12	Yq12	DYZ1-YsatIII

ONCOLOGY FISH TESTING SERVICES

TEST #	TEST NAME/DESCRIPTION
Testing provided by the ARUP Cytogenetics Laboratory	
2002647	Acute Lymphoblastic Leukemia (ALL) Panel by FISH, Adult
2002719	Acute Lymphoblastic Leukemia (ALL) Panel by FISH, Pediatric
2011132	Acute Myeloid Leukemia Panel by FISH
2002653	Acute Myelogenous Leukemia (AML) with Myelodysplastic Syndrome (MDS) or Therapy-Related AML, by FISH
2002295	Chromosome FISH, CLL Panel
2002298	Chromosome FISH, Interphase *To order tests with overlapping probes, order 2002298 and specify probes.*
2002378	Eosinophilia Panel by FISH
3002737	FISH, Interphase, CD138+ Cells
2002650	Lymphoma (Aggressive) Panel by FISH
3002063	Multiple Myeloma Panel by FISH
2002709	Myelodysplastic Syndrome (MDS) Panel by FISH
2002360	Myeloproliferative Disorders Panel by FISH
3000455	Ph-Like Acute Lymphoblastic Leukemia (ALL) Panel by FISH
2002363	*PML-RARA* Translocation by FISH
Testing provided by the ARUP Cytopathology Laboratory	
2002528	Pancreatobiliary FISH
2001181	UroVysion FISH

TEST #	TEST NAME/DESCRIPTION
Testing provided by the ARUP Immunohistochemistry Laboratory on Paraffin-Embedded Tissue	
3001309	1p/19q Deletion by FISH
3001302	*ALK* Gene Rearrangements by FISH, Lung
3001311	*BCL6* (3q27) Gene Rearrangement by FISH
3001304	*DDIT3* (*CHOP*) (12q13) Gene Rearrangement by FISH
3001310	*EGFR* Gene Amplification by FISH
2008603	*ERBB2* (*HER-2/neu*) Gene Amplification by FISH with Reflex, Tissue
3001305	*EWSR1* (22q12) Gene Rearrangement by FISH
3004075	*FGFR1* Gene Amplification by FISH
3001297	*FOXO1* (*FKHR*) (13q14) Gene Rearrangement by FISH
3000548	*FUS* (16p11) Gene Rearrangement by FISH
3001298	*IGH-BCL2* Fusion, t(14;18) by FISH
3001306	*IGH-CCND1* Fusion, t(11;14) by FISH
3001299	*IGH-MYC* Fusion, t(8;14) by FISH
3001568	*IRF4/DUSP22* (6p25) Gene Rearrangement by FISH
3001301	*MDM2* Gene Amplification by FISH
3001313	*MET* Gene Amplification by FISH
3001300	*MYC* (8q24) Gene Rearrangement by FISH
3001307	*MYCN* (*N-MYC*) Gene Amplification by FISH
3001312	*RET* Gene Rearrangements by FISH
3001308	*ROS1* by FISH
3001303	*SS18* (*SYT*) (18q11) Rearrangement by FISH
3002633	*TFE3* Gene Rearrangement by FISH

* FISH probes can be tested on touch prep samples.

Refer to the aliases under test code 2002298 on the Laboratory Test Directory on the ARUP website for a complete list of available probes. http://ltd.aruplab.com/Tests/Pub/2002298

ONCOLOGY FISH PROBES

TEST NAME / DESCRIPTION AND TEST #		PROBE TARGET	GENE(S)/UNIQUE SEQUENCE
To order individual probes, use ARUP test code 2002298, Chromosome, FISH Interphase, and specify the desired probe(s). Testing provided by the ARUP Cytogenetics Laboratory			
Acute Lymphoblastic Leukemia Panel (ALL)	Adult: 2002647	8q24	*MYC*
		t(9;22)(q34;q11.2)	*BCR-ABL1*
		11q23	*KMT2A* (*MLL*)
		14q32	*IGH*
		19p13	*TCF3* (*E2A*)
	Pediatric: 2002719	+4,+10	CEP4, CEP10
		t(9;22)(q34;q11.2)	*BCR-ABL1*
		11q23	*KMT2A* (*MLL*)
		t(12;21)(p13;q22)	*ETV6-RUNX1* (*TEL-AML1*)
Acute Myeloid Leukemia Panel (AML): 2011132		t(15;17)(q24;q21)	*PML-RARA*
		t(8;21)(q22;q22)	*RUNX1T1-RUNX1* (*ETO-AML1*)
		inv(16)(p13.3q22)	*CBFB*
		11q23	*KMT2A* (*MLL*)
		inv(3) or t(3;3)	*RPN1/MECOM* (*EVI1*)
		del(5)(q31)	*EGR1*
		del(7)(q31)/-7	D7S486
Acute Myelogenous Leukemia (AML) with Myelodysplastic Syndrome (MDS) or Therapy-Related AML, by FISH: 2002653		del(5)(q31)	*EGR1*
		del(7)(q31)/-7	D7S486
		11q23	*KMT2A* (*MLL*)
Alveolar Rhabdomyosarcoma *Probes ordered individually as test code 2002298*		13q13	*FKHR* (*FOX01*)
Chromosome FISH, CLL Panel: 2002295		del(11)(q22.3)	*ATM*
		+12	D12Z3
		del(13)(q14.3)	D13S319
		del(17)(p13.1)	*TP53* (*p53*)
Chronic Myelogenous Leukemia (CML): 2002298 *Probes ordered individually as test code 2002298*		t(9;22)(q34;q11.2)	*BCR-ABL1*, *ASS1*
Eosinophilia Panel by Fish: 2002378		4q12	*PDGFRA-CHIC2-FIP1L1*
		5q32	*PDGFRB*
		8p12	*FGFR1*
		inv(16)	*CBFB*
Lymphoma (Aggressive) Panel by Fish: 2002650		3q27	*BCL6*
		8q24	*MYC*
		t(14;18)(q32;q21)	*IGH-BCL2*

ONCOLOGY FISH PROBES continued

TEST NAME / DESCRIPTION AND TEST #		PROBE TARGET	GENE(S)/UNIQUE SEQUENCE
To order individual probes, use ARUP test code 2002298, Chromosome, FISH Interphase, and specify the desired probe(s). Testing provided by the ARUP Cytogenetics Laboratory			
Lymphoma *Probes ordered individually as test code 2002298*	Burkitt	8q24	*MYC*
	Diffuse large cell	3q27	*BCL6*
	Follicular	t(14;18)(q32;q21)	*IGH-BCL2*
	IgH rearrangement	14q32	*IGH*
	Mantle cell	t(11;14)(q13;q32)	*IGH-CCND1*
	MALT	18q21	*MALT1*
Multiple Myeloma Panel by FISH: 3002063		1q21	*CKS1B*
		+9	*ASS1*
		t(11;14)(q13;q32)	*IGH-CCND1*
		t(14;20)(q32;q12)	*IGH-MAFB*
		del(17)(p13.1)	*TP53 (p53)*
		t(4;14)(p16;q32)	*IGH-FGFR3*
		t(14;16)(q32;q23.1)	*IGH-MAF*
Myelodysplastic Syndrome (MDS) Panel by FISH: 2002709		del(5)(q31)	*EGR1*
		del(7)(q31)/-7	D7S486
		+8	CEP8
		del(20)(q12)	D20S108
Myeloproliferative Disorders Panel by FISH: 2002360		4q12	*PDGFRA-CHIC2-FIP1L1*
		5q32	*PDGFRB*
		8p12	*FGFR1*
		t(9;22)(q34;q11.2)	*BCR-ABL1*
Myxoid Liposarcoma *Probes ordered individually as test code 2002298*		12q13	*DDIT3* (*CHOP*)
Ph-Like Acute Lymphoblastic Leukemia (ALL) Panel by FISH: 3000455		Xp22.33/Yp11.32	*CRLF2*
		9p24	*JAK2*
		19p13.2	*EPOR*
		5q32	*CSF1R*
		9q34.1	*ABL1*
		1q25.2	*ABL2*
		5q32	*PDGFRB*
Sarcoma *Probes ordered individually as test code 2002298*	Synovial	18q11.2	*SS18* (*SYT*)
	Ewing	22q12.2	*EWSR1*
TFE3 Gene Rearrangement by FISH: 3002633		Xp11.2	*TFE3*

MOLECULAR GENETICS AND GENOMICS FOR CONSTITUTIONAL DISORDERS

The Molecular Genetics Department at ARUP provides a comprehensive test menu to assist physicians in the diagnosis of patients with inherited genetic disorders. The laboratory offers diagnostic testing, carrier screening for common genetic conditions, fetal testing, presymptomatic testing, molecular confirmation of abnormal newborn screening results, assessment for genetic variants affecting drug metabolism (pharmacogenetics), and multigene panels.

The molecular genetics menu offers diagnostic testing for more than 60 different conditions. Disorders for which molecular genetic testing is available include: cystic fibrosis, fragile X, alpha and beta thalassemia, Huntington disease, hemophilia A and B, hearing loss, hereditary hemorrhagic telangiectasia, hereditary non-polyposis colon cancer, pancreatitis, Rett syndrome, hemochromatosis, factor V leiden, and common Ashkenazi Jewish disorders.

Once a causative mutation is identified, targeted testing for the familial mutation can be requested for at-risk family members. Pharmacogenetic testing may identify genetic variants influencing the metabolism or efficacy of commonly prescribed drugs. Available pharmacogenetic tests include: tamoxifen (*CYP2D6*), irinotecan (*UGT1A1*), warfarin (*CYP2C9* and *VKORC1*), and numerous others.

Many of the hemoglobin-related tests supplement genetic testing for hemoglobinopathies/thalassemias, and include Hemoglobin Evaluation with Reflex to Electrophoresis and/or RBC Solubility, Oxygen Dissociation (P50) by Hemoximetry, and Hemoglobin Evaluation Reflexive Cascade.

ARUP's genetic counselors are available to answer questions regarding test selection or interpretation.

MULTIGENE PANELS

TEST #	TEST NAME/DESCRIPTION
3002685	Alport Syndrome Panel, Sequencing and Deletion/ Duplication
2006540	Aortopathy Panel, Sequencing and Deletion/Duplication
3001855	*BRCA1*and *BRCA2*-Associated HBOC Syndrome Panel, Sequencing and Deletion/Duplication
3003634	Capillary Malformation-Arteriovenous Malformation (CM-AVM) Panel, Sequencing and Deletion/Duplication
2010183	Cardiomyopathy and Arrhythmia Panel, Sequencing and Deletion/Duplication
3002286	Cerebral Cavernous Malformation Panel, Sequencing and Deletion/Duplication
2012151	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies Panel, Sequencing
2012155	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, *PMP22* Deletion/ Duplication with Reflex to Sequencing Panel
2007545	Childhood-Onset Epilepsy Panel, Sequencing and Deletion/Duplication
2011157	Cobalamin/Propionate/Homocysteine Metabolism Related Disorders Panel, Sequencing and Deletion/ Duplication
3001581	Dilated Cardiomyopathy Panel, Sequencing
3003917	Distal Arthrogryposis Panel, Sequencing
3001585	Early-Onset Alzheimer's Panel, Sequencing
3001839	Emery-Dreifuss Muscular Dystrophy Panel, Sequencing
2008803	Expanded Hearing Loss Panel, Sequencing and Deletion/ Duplication

TEST #	TEST NAME/DESCRIPTION
3002110	Familial Hypercholesterolemia Panel, Sequencing
2012026	Hereditary Breast and Ovarian Cancer Panel, Sequencing and Deletion/Duplication
2012032	Hereditary Cancer Panel, Sequencing and Deletion/ Duplication
2013449	Hereditary Gastrointestinal Cancer Panel, Sequencing and Deletion/Duplication
3000894	Hereditary Hemolytic Anemia Cascade
2012052	Hereditary Hemolytic Anemia Panel, Sequencing
2009337	Hereditary Hemorrhagic Telangiectasia (HHT) Panel, Sequencing and Deletion/Duplication
3001842	Hereditary Myeloid Neoplasms Panel, Sequencing
2010214	Hereditary Renal Cancer Panel, Sequencing and Deletion/ Duplication
3002682	Heterotaxyand Situs Inversus Panel, Sequencing
3002061	HLA Class I and II Panel (A,B,C,DRB1, DQA1, DQB1, DPB1) by Next Generation Sequencing
3002062	HLA Class I and II Panel (A,B,C,DRB1, DRB345, DQA1, DQB1, DPA1, DPB1) by Next Generation Sequencing
3002307	HLA Class I Panel (ABC) by Next Generation Sequencing
3002308	HLA Class II Panel (DRB1, DQA1and DQB1) by Next Generation Sequencing
2008848	*Holoprosencephaly Panel, Sequencing and Deletion/ Duplication
3001579	Hypertrophic Cardiomyopathy Panel, Sequencing
2007535	Infantile Epilepsy Panel, Sequence Analysis and Exon-Level Deletion/Duplication

MULTIGENE PANELS continued

TEST #	TEST NAME/DESCRIPTION
3003947	Loeys-Dietz Syndrome Core Panel, Sequencing
3001603	Long QT Panel, Sequencing and Deletion/Duplication
3002688	Malignant Hyperthermia Panel, Sequencing
3001965	Mitochondrial Disorders (mtDNA) Sequencing and Deletion Analysis by NGS
3001959	Mitochondrial Disorders Panel (mtDNA and Nuclear Genes)
3001593	MODY and Neonatal Diabetes Panel, Sequencing
3003927	Neurofibromatosis Type 1 Sequencing and Deletion/ Duplication and Legius Syndrome Sequencing Panel
2010772	*Noonan Spectrum Disorders Panel, Sequencing
3001607	Osteogenesis Imperfecta and Low Bone Density Panel, Sequencing
2007370	Periodic Fever Syndromes Panel, Sequencing and Deletion/Duplication

TEST #	TEST NAME/DESCRIPTION
2011156	Primary Antibody Deficiency Panel, Sequencing and Deletion/Duplication
3003927	Neurofibromatosis Type 1 Sequencing and Deletion/ Duplication and Legius Syndrome Sequencing Panel 3001621 Primary Ciliary Dyskinesia Panel, Sequencing
2009345	Pulmonary Arterial Hypertension (PAH) Panel, Sequencing and Deletion/Duplication
2012849	Rapid Mendelian Genes Sequencing Panel, Trio
2012015	*Skeletal Dysplasia Panel, Sequencing and Deletion/ Duplication
3001613	Sticker Syndrome Panel, Sequencing
3002100	Tuberous Sclerosis Complex Panel, Sequencing and Deletion/Duplication
3002096	Tuberous Sclerosis Complex Panel, Sequencing and Deletion/Duplication, Fetal
2007384	Vascular Malformations Panel, Sequencing and Deletion/ Duplication

EXOME TESTING

TEST #	TEST NAME/DESCRIPTION
3001457	Exome Reanalysis (Originally Tested at ARUP - No Specimen Required)

TEST #	TEST NAME/DESCRIPTION
2006336	Exome Sequencing, Proband
2006332	Exome Sequencing, Trio

FAMILIAL VARIANT TESTING

TEST #	TEST NAME/DESCRIPTION
2001961	Familial Mutation, Targeted Sequencing
2001980	Familial Mutation, Targeted Sequencing, Fetal

TEST #	TEST NAME/DESCRIPTION
3003144	Deletion/Duplication Analysis by MLPA

SANGER SEQUENCING, MLPA AND TARGETED VARIANT ANALYSIS

TEST #	TEST NAME/DESCRIPTION
2013725	*ABCC8*-Related Hyperinsulinism, 3 Variants
0051266	*Achondroplasia (*FGFR3*) 2 Mutations
2011906	‡Adrenoleukodystrophy, X-Linked (*ABCD1*) Sequencing and Deletion/Duplication
0051256	Alpha-1-Antitrypsin and A1A Genotype with Reflex to Phenotype
2011622	Alpha Globin (*HBA1* and *HBA2*) Deletion/Duplication
2011708	Alpha Globin (*HBA1* and *HBA2*) Sequencing and Deletion/Duplication
3003651	Alpha Thalassemia (*HBA1*and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring
3003656	Alpha Thalassemia (*HBA1*and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring, Fetal
2005077	Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR
2012232	Angelman Syndrome and Prader-Willi Syndrome by Methylation-Sensitive PCR, Fetal
2005564	Angelman Syndrome (*UBE3A*) Sequencing
0050392	Ankylosing Spondylitis (*HLAB27*) Genotyping
0030192	APC Resistance Profile with Reflex to Factor V Leiden
2013341	Apolipoprotein E (APOE) Genotyping, Alzheimer Disease Risk
2013337	Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk
0051415	Ashkenazi Jewish Diseases, 16 Genes
2014314	Autism and Intellectual Disability Comprehensive Panel (includes Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis)
3001635	Beckwith-Wiedemann Syndrome (BWS) and Russell-Silver Syndrome (RSS) by Methylation-Specific MLPA
0050578	*Beta Globin (*HBB*) Sequencing
2010117	‡Beta Globin (*HBB*) Sequencing and Deletion/Duplication
0051730	Biotinidase Deficiency (*BTD*) Sequencing
0051433	*Bloom Syndrome (*BLM*), 1 Variant
0051453	*Canavan Disease (*ASPA*), 4 Variants
2004203	‡Carnitine Deficiency, Primary (*SLC22A5*) Sequencing and Deletion/Duplication
2005018	Celiac Disease (*HLA-DQA1**05, *HLA-DQB1**02, and *HLA-DQB1**03:02) Genotyping

TEST #	TEST NAME/DESCRIPTION
3000531	Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy, CADASIL (NOTCH3), Sequencing
2012151	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies Panel Sequencing
2012155	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, PMP22 Deletion/Duplication with Reflex to Sequencing Panel
2012160	Charcot-Marie-Tooth Type 1A (CMT1A)/Hereditary Neuropathy with Liability to Pressure Palsies (HNPP), PMP22 Deletion/Duplication
2007069	Citrullinemia, Type I (*ASS1*) Sequencing
2013661	Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants
2013662	Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants, Fetal
2013663	Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants with Reflex to Sequencing
2013664	Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants with Reflex to Sequencing and Reflex to Deletion/Duplication
0051110	Cystic Fibrosis (*CFTR*) Sequencing
0051640	Cystic Fibrosis (*CFTR*) Sequencing with Reflex to Deletion/Duplication
2011235	Duchenne/Becker Muscular Dystrophy (*DMD*) Deletion/Duplication
2011231	Duchenne/Becker Muscular Dystrophy (*DMD*) Deletion/Duplication, Fetal
2011241	Duchenne/Becker Muscular Dystrophy (*DMD*) Deletion/Duplication with Reflex to Sequencing
2011153	Duchenne/Becker Muscular Dystrophy (*DMD*) Sequencing
0051463	Dysautonomia, Familial (*IKBKAP*), 2 Variants
0097720	Factor V Leiden (*F5*) R506Q Mutation
2014248	Factor V, R2 Mutation Detection by PCR
2003220	Factor XIII (*F13A1*) V34L Mutation
2004863	Familial Adenomatous Polyposis (*APC*) Sequencing
2004915	Familial Adenomatous Polyposis Panel: *APC* Sequencing, *APC* Deletion/Duplication, and *MYH* 2 Mutations
2002658	Familial Mediterranean Fever (*MEFV*) Sequencing
2014035	Familial Transthyretin Amyloidosis (TTR) Sequencing
0051468	*Fanconi Anemia Group C (*FANCC*) 2 Mutations

SANGER SEQUENCING, MLPA AND TARGETED VARIANT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2009033	Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis
2009034	Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis, Fetal
0051176	*Galactosemia (*GALT*) 9 Mutations
0051175	Galactosemia (*GALT*) Enzyme Activity and 9 Mutations
2006697	Galactosemia (*GALT*) Sequencing
3001957	Gamma Globin (*HBG1* and *HBG2*) Sequencing
3001648	Gaucher Disease (*GBA*) Sequencing
0051438	*Gaucher Disease (*GBA*), 8 Variants
3000258	Genetic Carrier Screen, (CF, FXS, and SMA) with Reflex to Methylation
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing
0051684	Glucose-6-Phosphate Dehydrogenase (*G6PD*) 2 Mutations, African Alleles
2013740	Glycogen Storage Disease, Type 1A (*G6PC*), 9 Variants
2011140	Guanidinoacetate Methyltransferase (*GAMT*) Deficiency Sequencing
0051374	Hearing Loss, Nonsyndromic, Connexin 26 (*GJB2*) Sequencing
2001956	Hearing Loss, Nonsyndromic, Connexin 30 (*GJB6*) 2 Deletions
2001992	Hearing Loss, Nonsyndromic Panel (*GJB2*) Sequencing, 2 Deletions, and Mitochondrial DNA, 2 Mutations
0055656	Hemochromatosis, Hereditary (*HFE*) 3 Mutations
2005792	Hemoglobin Evaluation Reflexive Cascade
2013399	Hemoglobin S, Sickle Solubility
2001759	*Hemophilia A (*F8*) 2 Inversions
2001614	‡Hemophilia A (*F8*) 2 Inversions with Reflex to Sequencing and Reflex to Deletion/Duplication
2001747	Hemophilia A (*F8*) Sequencing
2001578	Hemophilia B (*F9*) Sequencing
2010494	Hemophilia B (*F9*) Sequencing and Deletion/Duplication
3000894	Hereditary Hemolytic Anemia Cascade
2007167	‡Hereditary Paraganglioma-Pheochromocytoma (*SDHB*, *SDHC*, and *SDHD*) Sequencing and Deletion/Duplication
2007108	Hereditary Paraganglioma-Pheochromocytoma (*SDHB*) Sequencing and Deletion/Duplication
2007117	Hereditary Paraganglioma-Pheochromocytoma (*SDHC*) Sequencing and Deletion/Duplication
2007122	Hereditary Paraganglioma-Pheochromocytoma (*SDHD*) Sequencing and Deletion/Duplication
2011461	Hereditary Paraganglioma-Pheochromocytoma (*SDHA*) Sequencing

TEST #	TEST NAME/DESCRIPTION
0051650	‡HNPCC/Lynch Syndrome (*MLH1*) Sequencing and Deletion/Duplication
0051654	‡HNPCC/Lynch Syndrome (*MSH2*) Sequencing and Deletion/Duplication
0051656	‡HNPCC/Lynch Syndrome (*MSH6*) Sequencing and Deletion/Duplication
0051737	‡HNPCC/Lynch Syndrome (*PMS2*) Sequencing and Deletion/Duplication
0040018	◊Huntington Disease (*HD*) Mutation by PCR
2006274	Inherited Insulin Resistance Syndromes (*INSR*) Sequencing
2013909	Joubert Syndrome Type 2 (*TMEM216*), 1 Variant
2004992	‡Juvenile Polyposis (*BMPR1A*) Sequencing and Deletion/Duplication
3002001	Kell K/k (KEL) Antigen Genotyping
2009313	‡Li-Fraumeni (*TP53*) Sequencing and Deletion/Duplication
2013735	Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants
2004543	‡*LMNA*-Related Disorders (*LMNA*) Sequencing
2013730	Maple Syrup Urine Disease, Type 1B (*BCKDHB*), 3 Variants
2005589	Marfan Syndrome, *FBN1* Sequencing
2005584	‡Marfan Syndrome, *FBN1* Sequencing and Deletion/Duplication
2014699	Maternal T Cell Engraftment in SCID
2014704	Maternal T Cell Engraftment in SCID, Maternal Specimen
2014694	Maternal T Cell Engraftment in SCID, Pre-Engraftment Specimen
0051205	Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) 2 Mutations
0051758	Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) Sequencing
0055655	Methylenetetrahydrofolate Reductase (*MTHFR*) 2 Mutations
2005270	Mismatch Repair by IHC with Reflex to *MLH1* Promoter Methylation
0051755	Molar Pregnancy, 16 DNA Markers
0051448	*Mucolipidosis Type IV (*MCOLN1*) 2 Variants
2005359	Multiple Endocrine Neoplasia Type 1 (*MEN1*) Sequencing
2005360	‡Multiple Endocrine Neoplasia Type 1 (*MEN1*) Sequencing and Deletion/Duplication
0051390	Multiple Endocrine Neoplasia Type 2 (*MEN2*), RET Gene Mutations by Sequencing
2006191	*MUTYH*-Associated Polyposis (*MUTYH*) Sequencing
3001907	Myotonic Dystrophy Type 1 (*DMPK*) CTG Expansion
2005023	Narcolepsy (*HLA-DQB1**06:02) Genotyping

SANGER SEQUENCING, MLPA AND TARGETED VARIANT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2013745	*NEB*-Related Nemaline Myopathy, 1 Variant
0051458	*Niemann-Pick, Type A (*SMPD1*) 4 Variants
2014599	Non-Alcoholic Fatty Liver Disease Susceptibility (*PNPLA3*) Genotyping
0051805	Noonan Syndrome (*PTPN11*) Sequencing
2004896	‡Ornithine Transcarbamylase Deficiency (*OTC*) Sequencing and Deletion/Duplication
2010703	Pancreatitis (*CTRC*) Sequencing
2010876	Pancreatitis, Panel (*CTRC*, *CFTR*, *PRSS1*, *SPINK1*) Sequencing
3001768	Pancreatitis (*PRSS1*) Sequencing and Deletion/Duplication
2002012	Pancreatitis (*SPINK1*) Sequencing
2008398	‡Peutz-Jeghers Syndrome (*STK11*) Sequencing and Deletion/Duplication
2004980	Plasminogen Activator Inhibitor-1, PAI-1 (*SERPINE1*) Genotyping
3000193	Platelet antigen Genotyping Panel
3001170	Platelet Antigen Genotyping (HPA-1)
2012250	Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing and Deletion/Duplication
2012255	Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing
0056060	Prothrombin (*F2*) c.*97G>A (G20210A) Pathogenic Variant
2002470	‡*PTEN*-Related Disorders (*PTEN*) Sequencing and Deletion/Duplication
3001053	Red Blood Cell Antigen Genotyping
0051614	‡Rett Syndrome (*MECP2*) Sequencing and Deletion/Duplication
3002002	RhC/c (*RHCE*) Antigen Genotyping
0051368	RhD Gene (*RHD*) Copy Number
3002003	RhE/e (*RHCE*) Antigen Genotyping
3001401	SHOX-Related Disorders, Deletion/Duplication with Reflex to Sequencing

TEST #	TEST NAME/DESCRIPTION
2006240	Shwachman-Diamond Syndrome (*SBDS*) Sequencing
2011457	Smith-Lemli-Opitz Syndrome (*DHCR7*) Sequencing
2013436	Spinal Muscular Atrophy (SMA) Copy Number Analysis
2013444	Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal
2009298	*Tay-Sachs Disease (HEXA) Sequencing and 7.6kb Deletion
0051428	*Tay-Sachs (*HEXA*) 7 Mutations
2010015	Telangiectasia Syndrome (*BMP9/GDF2*) Sequencing
0051506	*Thanatophoric Dysplasia, Types I and II (*FGFR3*) 13 Mutations
0056200	Thrombotic Risk, DNA Panel
0030133	Thrombotic Risk, Inherited Etiologies (Most Common) with Reflex to Factor V Leiden
2006385	Thrombotic Risk Reflexive Panel
0050547	Twin Zygosity Testing
2013750	Usher Syndrome, Types 1F and 3 (*PCDH15* and *CLRN1*), 2 Variants
2004212	‡Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing and Deletion/Duplication
2002970	von Hippel-Lindau (*VHL*) Sequencing
2002965	‡von Hippel-Lindau (*VHL*) Sequencing and Deletion/Duplication
2005476	von Willebrand Disease, Platelet Type (*GP1BA*) 4 Mutations
2005480	von Willebrand Disease, Type 2A (*VWF*) Sequencing Exon 28 with Reflex to 9 Exons
2005486	von Willebrand Disease, Type 2B (*VWF*) Sequencing
2005490	von Willebrand Disease, Type 2M (*VWF*) Sequencing
2005494	von Willebrand Disease, Type 2N (*VWF*) Sequencing
2010716	Wilson Disease (*ATP7B*) Sequencing
2006352	X-Chromosome Inactivation Analysis
2001778	Y-Chromosome Microdeletion

PHARMACOGENETICS MARKERS

TEST #	TEST NAME/DESCRIPTION
3001501	*CYP2C8* and *CYP2C9*
3001508	*CYP2C19*
3001513	*CYP2D6*
3001518	*CYP3A4* and *CYP3A5*
3001524	Cytochrome P450 Genotype Panel
0055655	Methylenetetrahydrofolate Reductase (*MTHFR*) 2 Mutations

TEST #	TEST NAME/DESCRIPTION
2008767	Opioid Receptor, mu *OPRM1* Genotype, 1 Variant
2008426	*SLCO1B1*, 1 Variant
0092066	Thiopurine Methyltransferase, RBC
3001535	TMPT and NUDT15
0051332	UDP Glucuronosyltransferase 1A1 (*UGT1A1*) Genotyping—irinotecan
3001541	Warfarin Sensitivity (*CYP2C8*, *CYP2C9*, *CYP4F2*, *VKORC1*) Genotyping

HEMATOLOGIC DISORDERS

TEST #	TEST NAME/DESCRIPTION
3003651	Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring
3003656	Alpha Thalassemia (*HBA1* and *HBA2*) Deletion/Duplication with reflex to Hb Constant Spring, Fetal
2011622	Alpha Globin (*HBA1* and *HBA2*) Deletion/Duplication
0050578	*Beta Globin (*HBB*) Sequencing
2010117	‡Beta Globin (*HBB*) Sequencing and Deletion/Duplication
3001957	Gamma Globin (*HBG1* and *HBG2*) Sequencing
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing
0080135	Glucose-6-Phosphate Dehydrogenase (enzyme)
0051684	Glucose-6-Phosphate Dehydrogenase (*G6PD*) 2 Mutations

TEST #	TEST NAME/DESCRIPTION
0049090	Heinz Body Stain
2005792	Hemoglobin Evaluation Reflexive Cascade
0050610	Hemoglobin Evaluation with Reflex to Electrophoresis and/or RBC Solubility
3002645	Hemoglobin F with Reflex to Electrophoresis
3002644	Hemoglobin (*Hb*) A2 and F by Column with Reflex to Electrophoresis
0050520	Hemoglobin S, Evaluation with Reflex to RBC Solubility
2013399	Hemoglobin S, Sickle Solubility
0049020	Hemoglobin, Unstable
3000894	Hereditary Hemolytic Anemia Cascade
0080290	Pyruvate Kinase

+ Multiple tests available

* Fetal testing available

◊ Testing is not offered for patients under the age of 18

‡ For standalone deletion/duplication testing please contact the ARUP genetic counselors for more information

Please see the ARUP Molecular Oncology Services brochure for a complete listing of molecular tests related to oncology.

Refer to the Laboratory Test Directory on the ARUP website, **aruplab.com**, for more detailed test information.

ARUP LABORATORIES

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