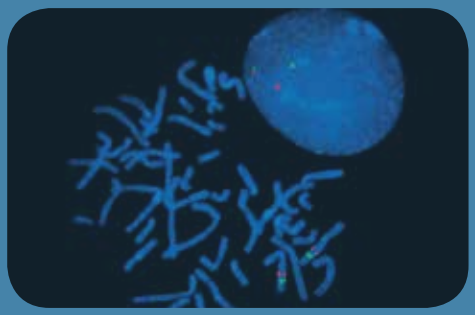
ARUP LABORATORIES | Genetics

genetic testing

PATIENTS. ANSWERS. RESULTS.


www.aruplab.com/genetics

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Information in this brochure is current as of November 2017. All content is subject to change. Please contact ARUP Client Services at (800) 522-2787 with any questions or concerns.

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.®

ISO 15189
CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

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NEW TESTS (November hot line)

TEST #	TEST NAME/DESCRIPTION
2014694	Maternal T Cell Engraftment in SCID, Pre-Engraftment Specimen
2014699	Maternal T Cell Engraftment in SCID
2014704	Maternal T Cell Engraftment in SCID, Maternal Specimen
2014599	Non-Alcoholic Fatty Liver Disease Susceptibility (*PNPLA3*) Genotyping

MATERNAL SERUM 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	
0081293	Maternal Screening, Sequential, Specimen#1
0081294	Maternal Screening, Sequential, Specimen#2
0081062	Maternal Serum Screening, Integrated, Specimen#1
0081064	Maternal Serum Screening, Integrated, Specimen#2
First-Trimester Screening	
0081150	Maternal Serum Screen, First Trimester

TEST #	TEST NAME/DESCRIPTION
Non-Invasive 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
Second-Trimester Screening	
0080434	Maternal Serum Screen, Alpha Fetoprotein (only)
0080269	Quad Screen: Alpha Fetoprotein, hCG, Estriol, and Inhibin A

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	Acylglycine, Quantitative, Urine
2011415	Alpha-Iduronidase Enzyme Activity in Leukocytes
0080137	Amino Acids Quantitative, 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
0093362	Biotinidase, Serum (with paired normal control)
0080068	Carnitine, Free and Total, Plasma
0081308	Carnitine, Free and Total, Urine
0080065	Carnitine, Free, Plasma
0081110	Carnitine Panel (Free and Total Carnitine, Acylcarnitine), Plasma or Serum
0080067	Carnitine, Total, Plasma
2002328	Creatine Disorders Panel, Plasma or Serum
2002333	Creatine Disorders Panel, Urine
0081106	Cystine Quantitative, Urine
0081105	Cystinuria Panel (Arginine, Cystine, Lysine, and Ornithine), Urine
0080351	Ehlers-Danlos Syndrome Type VI Screen, Urine
2013518	Fatty Acids Profile, Essential Serum and Plasma
0081296	Galactose-1-Phosphate in Red Blood Cells
0080125	Galactose-1-Phosphate Uridyltransferase, Whole Blood
0051175	Galactosemia Panel (Enzyme and DNA Testing for 7 Mutations and 2 Variants), Whole Blood
2014459	Gaucher Disease (GBA), Enzyme Activity in Leukocytes
2001510	Glutarylcarnitine Quantitative, Urine
2008129	Hexosaminidase A and Total Hexosaminidase in Plasma with Reflex to Hexosaminidase A and Total Hexosaminidase in Leukocytes
2008125	Hexosaminidase A and Total Hexosaminidase, Leukocytes
2008121	Hexosaminidase A and Total Hexosaminidase, Plasma or Serum
2012259	Keratan Sulfate, Quantitative by LC-MS/MS, Urine
2005255	Methylmalonic Acid, Serum or Plasma (Metabolic Disorders)
0081352	Mucopolysaccharides Electrophoresis and Quantitation, Urine
0081357	Mucopolysaccharides, Quantitative, Urine
2007599	Mucopolysaccharidosis Type I, Total HS and NRE (Sensi-Pro) Quantitative, Serum or Plasma
2007488	Mucopolysaccharidosis Type I, Total HS and NRE (Sensi-Pro) Quantitative, Urine
2008775	Mucopolysaccharidosis Type II, Total HS and NRE (Sensi-Pro) Quantitative, Serum or Plasma
2009282	Mucopolysaccharidosis Type II, Total HS and NRE (Sensi-Pro) Quantitative, Urine
0092458	Orotic Acid and Orotidine, Urine
0098389	Organic Acids, 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, Urine
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 post-treatment.

TEST #	TEST NAME/DESCRIPTION
Constitutional/Postnatal Testing	
0040208	Aneuploidy Panel by FISH
2002289	Chromosome Analysis, Peripheral Blood
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
2005749	Chromosome Analysis, Breakage, Ataxia Telangiectasia, Whole Blood
0097688	Chromosome Analysis, Breakage, Fanconi Anemia
2010795	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue—Products of Conception
2003414	Cytogenomic SNP Microarray
2006267	Cytogenomic SNP Microarray Buccal Swab
2009353	Cytogenomic SNP Microarray, with Five-Cell ChromosomeStudy, Peripheral Blood
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
2002293	Chromosome Analysis, Amniotic Fluid
2008367	Chromosome Analysis, Amniotic Fluid, with Reflex to Genomic Microarray
2002291	Chromosome Analysis, Chorionic Villus
2005763	Chromosome Analysis, Peripheral Blood, with Reflex to Genomic Microarray
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
2002297	Chromosome FISH, Prenatal
2002366	Cytogenomic SNP Microarray—Fetal

For more on Genetics, including white papers and additional technical information, please visit

Constitutional FISH Probes—order test 2002299		
MICRODELETION SYNDROMES		
Suspected Diagnosis	**Probe Target**	**Gene(s)/Unique Sequence**
Aneuploidy, Common (ARUP test code 0040208)	13/18/21/X/Y	
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)		
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	
2002298	Chromosome FISH, Interphase
2002294	Multiple Myeloma Panel by FISH
2002295	Chromosome FISH, CLL Panel
2002360	Myeloproliferative Disorders Panel by FISH
2002363	*PML-RARA* Translocation by FISH
2002378	Eosinophilia Panel by FISH
2011132	Acute Myelogenous Leukemia Panel by FISH
2002647	Acute Lymphocytic Leukemia Panel by FISH, Adult
2002719	Acute Lymphocytic Leukemia Panel by FISH, Pediatric
2002650	Lymphoma (Aggressive) Panel by FISH
2002653	AML with MDS or Therapy Related AML by FISH
2002709	Myelodysplastic Syndrome Panel by FISH
2002298	**EWSR1* Break Apart Rearrangement by FISH
2002298	**SYT* Rearrangement by FISH
Testing provided by the ARUP Cytopathology Laboratory	
2002528	Pancreatobiliary FISH
2001181	UroVysion FISH

* FISH probes can be tested on touch prep samples.

TEST #	TEST NAME/DESCRIPTION
Testing provided by the ARUP Immunohistochemistry Laboratory on Paraffin-Embedded Tissue	
2008604	1p/19q Deletion by FISH
2006102	*ALK* Gene Rearrangements by FISH, Lung
2010107	*BCL6* (3q27) Gene Rearrangement by FISH
2007223	*DDIT3* (*CHOP*) (12q13) Gene Rearrangement by FISH
2008605	*EGFR* Gene Amplification by FISH
2008603	*ERBB2* (*HER-2/neu*) Gene Amplification by FISH
2007225	*EWSR1* (22q12) Gene Rearrangement by FISH
2001497	*FOXO1* (*FKHR*) (13q14) Gene Rearrangement by FISH
2001536	*IGH-BCL2* Fusion, t(14;18) by FISH for Detection in Follicular Lymphoma
2007226	*IGH-CCND1* Fusion, t(11;14) by FISH for Detection in Mantle Cell Lymphoma
2001538	*IGH-MYC* Fusion, t(8;14) by FISH for Detection in Burkitt Lymphoma
2003016	*MDM2* Gene Amplification by FISH
2002345	*MYC* (8q24) Gene Rearrangement by FISH
2007227	*MYCN* (*N-MYC*) Gene Amplification by FISH
2008418	*ROS1* by FISH
2007222	*SS18* (*SYT*) (18q11) Rearrangement by FISH

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

To order individual probes, use ARUP test code 2002298, Chromosome, FISH Interphase, and specify the desired probe(s).

Testing provided by the ARUP Cytopathology Laboratory			
FISH PROBES AVAILABLE FOR ONCOLOGY SAMPLES			
Indication		**Probe Target**	**Gene(s)/Unique Sequence**
Acute Lymphocytic 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 Myelogenous 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 with Myelodysplastic Syndrome or Therapy-Related AML Panel: 2002653		del(5)(q31)	*EGR1*
		del(7)(q31)/-7	D7S486
		11q23	*KMT2A* (*MLL*)
Chronic Lymphocytic Leukemia Panel (CLL): 2002295		del(11)(q22.3)	*ATM*
		+12	D12Z3
		del(13)(q14.3)	D13S319
		del(17)(p13.1)	*TP53* (*p53*)
Chronic Myelogenous Leukemia (CML): 2002298		t(9;22)(q34;q11.2)	*BCR-ABL1*, *ASS1*
Eosinophilia Panel: 2002378		4q12	*PDGFRA-CHIC2-FIP1L1*
		5q33.1	*PDGFRB*
		8p12	*FGFR1*
		inv(16)	*CBFB*

ONCOLOGY FISH continued

Testing provided by the ARUP Cytopathology Laboratory

FISH PROBES AVAILABLE FOR ONCOLOGY SAMPLES

Indication		Probe Target	Gene(s)/Unique Sequence
Lymphoma (Aggressive) Panel: 2002650		3q27	*BCL6*
		8q24	*MYC*
		t(14;18)(q32;q21)	*IGH-BCL2*
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: 2002294		1q21	*CKS1B*
		+9	*ASS1*
		t(11;14)(q13;q32)	*IGH-CCND1*
		14q32	*IGH* rearrangement
		+15	*PML*
		del(17)(p13.1)	*TP53 (p53)*
		NOTE: If IGH is positive, additional testing will include:	
		t(4;14)(p16;q32)	*IGH-FGFR3*
		t(14;16)(q32;q23.1)	*IGH-MAF*
Myelodysplastic Syndrome Panel (MDS): 2002709		del(5)(q31)	*EGR1*
		del(7)(q31)/-7	D7S486
		+8	CEP8
		del(20)(q12)	D20S108
Myeloproliferative Disorder Panel (MPD): 2002360		4q12	*PDGFRA-CHIC2-FIP1L1*
		5q33.1	*PDGFRB*
		8p12	*FGFR1*
		t(9;22)(q34;q11.2)	*BCR-ABL1*
Sarcoma: 2002298	Synovial	18q11.2	*SS18* (*SYT*)
	Ewing	22q12.2	*EWSR1*
Myxoid Liposarcoma: 2002298		12q13	*DDIT3* (*CHOP*)
Alveolar Rhabdomyosarcoma: 2002298		13q13	*FKHR* (*FOX01*)

MOLECULAR GENETICS WITH SEQUENCING, FRAGMENT ANALYSIS, AND GENOMICS

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 multi-gene 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.

MOLECULAR GENETICS MULTI-GENE PANELS (GENOMICS)

TEST #	TEST NAME/DESCRIPTION
2006540	Aortopathy Panel, Sequencing and Deletion/Duplication, 21 Genes
2012026	Breast and Ovarian Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 20 Genes
2012032	Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes
2010183	Cardiomyopathy and Arrhythmia Panel, Sequencing (83 Genes) and Deletion/Duplication (81 Genes)
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 (25 Genes) and Deletion/Duplication (24 Genes)
2006340	Exome Sequencing, Familial Control, Tracking
2006336	Exome Sequencing Symptom-Guided Analysis, Patient Only
2006332	Exome Sequencing with Symptom-Guided Analysis
2008803	Expanded Hearing Loss Panel, Sequencing (56 Genes) and Deletion/Duplication (53 Genes)
2013449	Gastrointestinal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 16 Genes
2012052	Hereditary Hemolytic Anemia Sequencing, 28 Genes
2009337	Hereditary Hemorrhagic Telangiectasia (HHT) Panel, Sequencing and Deletion/Duplication, 5 Genes

TEST #	TEST NAME/DESCRIPTION
2008848	*Holoprosencephaly Panel, Nonsyndromic, Sequencing and Deletion/Duplication, 11 Genes
2007535	Infantile Epilepsy Panel, Sequence Analysis and Exon-Level Deletion/Duplication
2006065	Mitochondrial Disorders (mtDNA) Sequencing
2006054	Mitochondrial Disorders Panel (mtDNA Sequencing and Deletion/Duplication , 121 Nuclear Genes Sequencing, 119 Nuclear Genes Deletion/Duplication)
2010772	*Noonan Spectrum Disorders Panel, Sequencing, 15 Genes
2007370	Periodic Fever Syndromes Panel, Sequencing, 7 Genes, and Deletion/Duplication, 6 Genes
2011156	Primary Antibody Deficiency Panel, Sequencing (35 Genes) and Deletion/Duplication (26 Genes)
2007533	Progressive Myoclonic Epilepsy (PME) Panel, Sequence Analysis and Exon-Level Deletion/Duplication
2009345	Pulmonary Arterial Hypertension (PAH) Panel, Sequencing and Deletion/Duplication, Multigene
2010214	Renal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 15 Genes
2007085	Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Sequencing and Deletion/Duplication, 53 Genes
2012015	*Skeletal Dysplasia Panel, Sequencing (39 Genes) and Deletion/Duplication (35 Genes)
2007384	Vascular Malformations Panel, Sequencing and Deletion/Duplication, 14 Genes

MOLECULAR GENETICS WITH GENETIC SEQUENCING AND FRAGMENT ANALYSIS

TEST #	TEST NAME/DESCRIPTION
2013725	*ABCC8*-Related Hyperinsulinism, 3 Variants
0051266	*Achondroplasia (*FGFR3*) 2 Mutations
2011902	Adrenoleukodystrophy, X- Linked (*ABCD1*) Sequencing
2011906	‡Adrenoleukodystrophy, X-Linked (*ABCD1*) Sequencing and Deletion/Duplication
2006230	Alagille Syndrome (*JAG1*) by Targeted 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
0051495	*Alpha Thalassemia *HBA1* and *HBA2* Gene Deletions
2001961	*Alpha Thalassemia *HBA1* and *HBA2* Sequencing, Familial Mutation
0051710	Alport Syndrome, X-linked (*COL4A5*) 3 Mutations
0051786	Alport Syndrome, X-linked (*COL4A5*) Sequencing
2002398	‡Alport Syndrome, X-linked (*COL4A5*) Sequencing and Deletion/Duplication
2001961	*Alport Syndrome, X-linked (*COL4A5*) Sequencing, Familial Mutation
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
2001961	*Angelman Syndrome (*UBE3A*) Sequencing, Familial Mutation
0050392	Ankylosing Spondylitis (*HLAB27*) Genotyping
0030192	APC Resistance Profile with Reflex to Factor V Leiden
0055654	Apolipoprotein B Mutation Detection
2013341	Apolipoprotein E (APOE) Genotyping, Alzheimer Disease Risk
2013337	Apolipoprotein E (APOE) Genotyping, Cardiovascular Risk
2011144	Arginine:Glycine Amidinotransferase (*GATM*) Deficiency sequencing
0051415	Ashkenazi Jewish Diseases, 16 Genes
2007872	*ATP7A*-Related Copper Transport Disorders (*ATP7A*) Sequencing
2008471	‡*ATP7A*-Related Copper Transport Disorders (*ATP7A*) Sequencing and Deletion/Duplication
2001961	**ATP7A*-Related Copper Transport Disorders (*ATP7A*) Sequencing, Familial Mutation

TEST #	TEST NAME/DESCRIPTION
2014314	Autism and Intellectual Disability Comprehensive Panel (includes Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis)
0051288	Beta-2-Adrenergic Receptor Haplotyping
0051421	*Beta Globin Gene Mutations for HbS, HbC, and HbE by PCR
0050578	*Beta Globin (*HBB*) Sequencing
2001961	*Beta Globin (*HBB*) Sequencing, Familial Mutation
2010117	‡Beta Globin (*HBB*) Sequencing and Deletion/Duplication
0051700	Biotinidase Deficiency (*BTD*) 5 Mutations
0051730	Biotinidase Deficiency (*BTD*) Sequencing
2001961	*Biotinidase Deficiency (*BTD*) Sequencing, Familial Mutation
2004739	Blood Group Genotyping by Microarray
0051433	*Bloom Syndrome (*BLM*), 1 Variant
0051750	*BRAF* V600E Mutation with Reflex to *MLH1* Promoter Methylation, Paraffin
2011954	Breast and Ovarian Hereditary Cancer Syndrome (*BRCA1* and *BRCA2*) Sequencing
2011949	Breast and Ovarian Hereditary Cancer Syndrome (*BRCA1* and *BRCA2*) Sequencing and Deletion/Duplication
0051453	*Canavan Disease (*ASPA*), 4 Variants
0051682	Carnitine Deficiency, Primary (*SLC22A5*) Sequencing
2004203	‡Carnitine Deficiency, Primary (*SLC22A5*) Sequencing and Deletion/Duplication
2001961	*Carnitine Deficiency, Primary (*SLC22A5*) Sequencing, Familial Mutation
2004931	*CDKL5*-Related Disorders (*CDKL5*) Sequencing
2004935	‡*CDKL5*-Related Disorders (*CDKL5*) Sequencing and Deletion/Duplication
2001961	**CDKL5*-Related Disorders (*CDKL5*) Sequencing, Familial Mutation
2005018	Celiac Disease (*HLA-DQA1**05, *HLA-DQB1**02, and *HLA-DQB1**03:02) Genotyping
2001961	*‡Cerebral Cavernous Malformation (*CCM1*, *CCM2*, or *CCM3*) Sequencing, Familial Mutation
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
2012609	CHARGE Syndrome (*CHD7*) Sequencing
2012717	CHARGE Syndrome (*CHD7*) Sequencing, Fetal

MOLECULAR GENETICS WITH GENETIC SEQUENCING AND FRAGMENT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2006356	Chronic Granulomatous Disease (*CYBB* Gene Scanning and *NCF1* Exon 2 GT Deletion) with Reflex to *CYBB* Sequencing
2001961	*Chronic Granulomatous Disease (*CYBB*) Sequencing, Familial Mutation
2006366	Chronic Granulomatous Disease (*NCF1*) Exon 2 GT Deletion
2006361	Chronic Granulomatous Disease, X-Linked (*CYBB*) Gene Scanning with Reflex to Sequencing
2006261	Citrin Deficiency (*SLC25A13*) Sequencing
2001961	*Citrin Deficiency (*SLC25A13*) Sequencing, Familial Mutation
2007069	Citrullinemia, Type I (*ASS1*) Sequencing
2001961	*Citrullinemia, Type I (*ASS1*) Sequencing, Familial Mutation
2006220	Congenital Amegakaryocytic Thrombocytopenia (CAMT) Sequencing
2008615	Creatine Transporter Deficiency (*SLC6A8*) Sequencing
2008610	‡Creatine Transporter Deficiency (*SLC6A8*) Sequencing and Deletion/Duplication
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
2001961	*Cystic Fibrosis (*CFTR*) Sequencing, Familial Mutation
0051640	Cystic Fibrosis (*CFTR*) Sequencing with Reflex to Deletion/Duplication
0056006	Cystic Fibrosis Cis-Trans (*CFTR*) R117H and 5T Mutations
2006236	Diamond-Blackfan Anemia (*RPL11*) Sequencing
2006234	Diamond-Blackfan Anemia (*RPL5*) Sequencing
2006238	Diamond-Blackfan Anemia (*RPS19*) Sequencing
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	*Fanconi Anemia, Group C (*FANCC*), 2 Variants
2006244	Dyskeratosis Congenita, Autosomal (*TERC*) Sequencing
2006228	Dyskeratosis Congenita, X-linked (*DKC1*) Sequencing

TEST #	TEST NAME/DESCRIPTION
2005559	‡Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Sequencing and Deletion/Duplication
2001961	*Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Sequencing, Familial Mutation
2014680	Expanded Carrier Screen by Next Generation Sequencing
2014677	Expanded Carrier Screen by Next Generation Sequencing with Fragile X
2014674	Expanded Carrier Screen Genotyping
2014671	Expanded Carrier Screen Genotyping with Fragile X
0097720	Factor V Leiden (*F5*) R506Q Mutation
2001549	Factor V, R2 Mutation
2003220	Factor XIII (*F13A1*) V34L Mutation
2004863	Familial Adenomatous Polyposis (*APC*) Sequencing
2001961	*Familial Adenomatous Polyposis (*APC*) Sequencing, Familial Mutation
2004915	Familial Adenomatous Polyposis Panel: *APC* Sequencing, *APC* Deletion/Duplication, and *MYH* 2 Mutations
2002658	Familial Mediterranean Fever (*MEFV*) Sequencing
2001961	*Familial Mediterranean Fever (*MEFV*) Sequencing, Familial Mutation
2001961	*Familial Mutation, Targeted Sequencing
2001980	Familial Mutation, Targeted Sequencing, Fetal
2014035	Familial Transthyretin Amyloidosis (TTR) Sequencing
0051468	*Fanconi Anemia Group C (*FANCC*) 2 Mutations
2007883	Filaggrin (*FLG*) 2 mutations
2009033	Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis
2009034	Fragile X (*FMR1*) PCR with Reflex to Methylation Analysis, Fetal
2002662	Freeman-Sheldon Syndrome (*MYH3*) Sequencing Exon 17
0051176	*Galactosemia (*GALT*) 9 Mutations
0051175	Galactosemia (*GALT*) Enzyme Activity and 9 Mutations
2006697	Galactosemia (*GALT*) Sequencing
2001961	*Galactosemia (*GALT*) Sequencing, Familial Mutation
0051438	*Gaucher Disease (*GBA*), 8 Variants
0051476	Glaucoma, Primary Congenital (*CYP1B1*) Sequencing
2001961	*Glaucoma, Primary Congenital (*CYP1B1*) Sequencing, Familial Mutation
2011470	*GLI3*-related disorders (*GLI3*) Sequencing
2011465	‡*GLI3*-Related Disorders (*GLI3*) Sequencing and Deletion/Duplication
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing

MOLECULAR GENETICS WITH GENETIC SEQUENCING AND FRAGMENT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2001961	*Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing, Familial Mutation
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
0040116	Haptoglobin (HP) Genotyping
0051374	Hearing Loss, Nonsyndromic, Connexin 26 (*GJB2*) Sequencing
2001961	*Hearing Loss, Nonsyndromic, Connexin 26 (*GJB2*) Sequencing, Familial Mutation
2001956	Hearing Loss, Nonsyndromic, Connexin 30 (*GJB6*) 2 Deletions
2002044	Hearing Loss, Nonsyndromic, Mitochondrial DNA 2 Mutations
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
2004686	Hemoglobin Lepore (*HBD-HBB* Fusion) 3 Mutations
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
2001961	*Hemophilia A (*F8*) Sequencing, Familial Mutation
2001578	Hemophilia B (*F9*) Sequencing
2010494	Hemophilia B (*F9*) Sequencing and Deletion/Duplication
2001961	*Hemophilia B (*F9*) Sequencing, Familial Mutation
0051381	Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Sequencing
0051382	‡Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Sequencing and Deletion/Duplication
2009008	Hereditary Hemorrhagic Telangiectasia (*ACVRL1* and *ENG*) Sequencing and Deletion/Duplication with Reflex to Juvenile Polyposis (*SMAD4*) Sequencing and Deletion/Duplication
2001961	*Hereditary Hemorrhagic Telangiectasia (*ACVRL1* or *ENG*) Sequencing, Familial Mutation
2007167	‡Hereditary Paraganglioma-Pheochromocytoma (*SDHB*, *SDHC*, and *SDHD*) Sequencing and Deletion/Duplication
2007108	Hereditary Paraganglioma-Pheochromocytoma (*SDHB*) Sequencing and Deletion/Duplication
2001961	*Hereditary Paraganglioma-Pheochromocytoma (*SDHB*) Sequencing, Familial Mutation
2007117	Hereditary Paraganglioma-Pheochromocytoma (*SDHC*) Sequencing and Deletion/Duplication
2001961	*Hereditary Paraganglioma-Pheochromocytoma (*SDHC*) Sequencing, Familial Mutation
2007122	Hereditary Paraganglioma-Pheochromocytoma (*SDHD*) Sequencing and Deletion/Duplication
2011461	Hereditary Paraganglioma-Pheochromocytoma (*SDHA*) Sequencing
2001961	*Hereditary Paraganglioma-Pheochromocytoma (*SDHD*) Sequencing, Familial Mutation
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
2001961	*HNPCC/Lynch Syndrome Sequencing, Familial Mutation
0049302	HNPCC/Microsatellite Instability by IHC
0040018	◊Huntington Disease (*HD*) Mutation by PCR
0051367	Hypochondroplasia (*FGFR3*) 2 Mutations
2006274	Inherited Insulin Resistance Syndromes (*INSR*) Sequencing
2001961	*Inherited Insulin Resistance Syndromes (*INSR*) Sequencing, Familial Mutation
2013909	Joubert Syndrome Type 2 (*TMEM216*), 1 Variant
2004988	Juvenile Polyposis (*BMPR1A*) Sequencing
2004992	‡Juvenile Polyposis (*BMPR1A*) Sequencing and Deletion/Duplication
2001961	*Juvenile Polyposis (*BMPR1A*) Sequencing, Familial Mutation
0051510	Juvenile Polyposis (*SMAD4*) Sequencing
2001971	‡Juvenile Polyposis (*SMAD4*) Sequencing and Deletion/Duplication
2001961	*Juvenile Polyposis (*SMAD4*) Sequencing, Familial Mutation
2009306	Kabuki Syndrome (*KMT2D*) Sequencing
0051644	Kell Antigen Genotyping (*KEL1-KEL2*)
2002945	Legius Syndrome (*SPRED1*) Sequencing
2008347	‡Legius Syndrome (*SPRED1*) Sequencing and Deletion/Duplication
2001961	*Legius Syndrome (*SPRED1*) Sequencing, Familial Mutation
2009302	Li-Fraumeni (*TP53*) Sequencing
2009313	‡Li-Fraumeni (*TP53*) Sequencing and Deletion/Duplication

MOLECULAR GENETICS WITH GENETIC SEQUENCING AND FRAGMENT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2013735	Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants
2004543	‡*LMNA*-Related Disorders (*LMNA*) Sequencing
2001961	**LMNA*-Related Disorders (*LMNA*) Sequencing, Familial Mutation
2002705	Loeys-Dietz Syndrome (*TGFBR1* and *TGFBR2*) Sequencing
2001961	*Loeys-Dietz Syndrome (*TGFBR1* or *TGFBR2*) Sequencing, Familial Mutation
0051674	Macular Degeneration, Age-Related, 2 DNA Markers
2013730	Maple Syrup Urine Disease, Type 1B (*BCKDHB*), 3 Variants
2005589	Marfan Syndrome, *FBN1* Sequencing
2005584	‡Marfan Syndrome, *FBN1* Sequencing and Deletion/Duplication
2001961	*Marfan Syndrome, *FBN1* Sequencing, Familial Mutation
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
2001961	*Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) Sequencing, Familial Mutation
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
2001961	*Multiple Endocrine Neoplasia Type 1 (*MEN1*) Sequencing, Familial Mutation
0051390	Multiple Endocrine Neoplasia, Type 2 (*RET*) Sequencing Exons 10, 11, 13–16
2001961	*Multiple Endocrine Neoplasia Type 2 (*RET*) Sequencing, Familial Mutation
2006307	*MUTYH*-Associated Polyposis (*MUTYH*) 2 Mutations with Reflex to Sequencing
2006191	*MUTYH*-Associated Polyposis (*MUTYH*) Sequencing
2001961	**MUTYH*-Associated Polyposis (*MUTYH*) Sequencing, Familial Mutation
2004911	*MYH*-Associated Polyposis (*MYH*) 2 Mutations
2005023	Narcolepsy (*HLA-DQB1*06:02) Genotyping

TEST #	TEST NAME/DESCRIPTION
2013745	*NEB*-Related Nemaline Myopathy, 1 Variant
2007159	Neurofibromatosis Type 1 (*NF1*) Sequencing
2007154	‡Neurofibromatosis Type 1 (*NF1*) Sequencing and Deletion/Duplication
2001961	*Neurofibromatosis Type 1 (*NF1*) Sequencing, Familial Mutation
0051458	*Niemann-Pick, Type A (*SMPD1*) 4 Variants
2009077	Non-Invasive Prenatal Testing for RHD Genotyping, Fetal
2014599	Non-Alcoholic Fatty Liver Disease Susceptibility (*PNPLA3*) Genotyping
2001961	*Noonan Syndrome (*PTPN11* or *SOS1*) Sequencing, Familial Mutation
0051805	Noonan Syndrome (*PTPN11*) Sequencing
2004189	Noonan Syndrome (*PTPN11*) Sequencing with Reflex to SOS1 Sequencing
2004195	Noonan Syndrome (*SOS1*) Sequencing
2004901	Ornithine Transcarbamylase Deficiency (*OTC*) Sequencing
2004896	‡Ornithine Transcarbamylase Deficiency (*OTC*) Sequencing and Deletion/Duplication
2001961	*Ornithine Transcarbamylase Deficiency (*OTC*) Sequencing, Familial Mutation
2010703	Pancreatitis (*CTRC*) Sequencing
2010876	Pancreatitis, Panel (*CTRC*, *CFTR*, *PRSS1*, *SPINK1*) Sequencing
2002016	Pancreatitis (*PRSS1*) Sequencing
2001961	*Pancreatitis (*PRSS1*) Sequencing, Familial Mutation
2002012	Pancreatitis (*SPINK1*) Sequencing
2001961	*Pancreatitis (*SPINK1*) Sequencing, Familial Mutation
2008394	Peutz-Jeghers Syndrome (*STK11*) Sequencing
2008398	‡Peutz-Jeghers Syndrome (*STK11*) Sequencing and Deletion/Duplication
2001961	*Peutz-Jeghers Syndrome (*STK11*) Sequencing, Familial Mutation
2004980	Plasminogen Activator Inhibitor-1, PAI-1 (*SERPINE1*) Genotyping
+	Platelet Antigen Genotyping: HPA 1, 2, 3, 4, 5, 6, and 15 (ordered individually or as panel)
2012250	Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing and Deletion/Duplication
2012255	Polycystic Kidney Disease, Autosomal Dominant (PKD1 and PKD2) Sequencing
2005077	Angelman Syndrome and Prader-Willi Syndrome by Methylation
0051476	Primary Congenital Glaucoma (*CYP1B1*) Sequencing
0056060	Prothrombin (*F2*) G20210A Mutation

MOLECULAR GENETICS WITH GENETIC SEQUENCING AND FRAGMENT ANALYSIS continued

TEST #	TEST NAME/DESCRIPTION
2005898	Protocadherin 19 (*PCDH19*) Sequencing
2002722	*PTEN*-Related Disorders (*PTEN*) Sequencing
2002470	‡*PTEN*-Related Disorders (*PTEN*) Sequencing and Deletion/Duplication
2001961	**PTEN*-Related Disorders (*PTEN*) Sequencing, Familial Mutation
2003410	Pulmonary Arterial Hypertension (*BMPR2*) Sequencing
2003405	‡Pulmonary Arterial Hypertension (*BMPR2*) Sequencing and Deletion/Duplication
2001961	*Pulmonary Arterial Hypertension (*BMPR2*) Sequencing, Familial Mutation
2002730	*RASA1*-Related Disorders (*RASA1*) Sequencing
2007852	‡*RASA1*-Related Disorders (*RASA1*) Sequencing and Deletion/Duplication
2001961	**RASA1*-Related Disorders (*RASA1*) Sequencing, Familial Mutation
0051378	Rett Syndrome (*MECP2*) Sequencing
0051614	‡Rett Syndrome (*MECP2*) Sequencing and Deletion/Duplication
2001961	*Rett Syndrome (*MECP2*) Sequencing, Familial Mutation
0050421	RhCc Antigen (*RHCE*) Genotyping
0051368	RhD Antigen (*RhD*) Genotyping
2009077	Non-Invasive Prenatal Testing for RhD Genotyping, Fetal
0050423	RhEe Antigen (*RHCE*) Genotyping
2005896	*SCN1A*-Related Seizure Disorders (*SCN1A*), Sequencing and Deletion/Duplication
2006948	SDHB with Interpretation by Immunohistochemistry
2006240	Shwachman-Diamond Syndrome (*SBDS*) Sequencing
2011457	Smith-Lemli-Opitz Syndrome (*DHCR7*) Sequencing
2011704	Smith-Lemli-Opitz Syndrome (*DHCR7*) Sequencing, Fetal
2013436	Spinal Muscular Atrophy (SMA) Copy Number Analysis
2013444	Spinal Muscular Atrophy (SMA) Copy Number Analysis, Fetal
2007569	TACI-Associated Common Variable Immunodeficiency (*TNFRSF13B*) Sequencing

TEST #	TEST NAME/DESCRIPTION
2001961	*TACI-Associated Common Variable Immunodeficiency (*TNFRSF13B*) Sequencing, Familial Mutation
2009298	*Tay-Sachs Disease (*HEXA*), 7 Variants
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
2002001	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing
2004212	‡Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing and Deletion/Duplication
2001961	*Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) Sequencing, Familial Mutation
2002970	von Hippel-Lindau (*VHL*) Sequencing
2002965	‡von Hippel-Lindau (*VHL*) Sequencing and Deletion/Duplication
2001961	*von Hippel-Lindau (*VHL*) Sequencing, Familial Mutation
2005476	von Willebrand Disease, Platelet Type (*GP1BA*) 4 Mutations
2001961	*von Willebrand Disease, Type 2A, 2B, 2M, or 2N (*VWF*) Sequencing, Familial Mutation
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
2007228	5-Fluorouracil (5-FU) Toxicity and Chemotherapeutic Response, 5 Mutations
2004247	CEBPA Mutation Detection
2002766	Cytochrome P450 2C9, *CYP2C9* - 2 Variants
2012769	Cytochrome P450 2C19, *CYP2C19* - 9 Variants
2012772	Warfarin Sensitivity, *CYP2C9* and *VKORC1*, 3 Variants
2012740	Cytochrome P450 3A5 Genotyping, *CYP3A5*, 2 Variants
2013098	Cytochrome P450 Genotype Panel
2014547	Cytochrome P450 2D6 (CYP2D6) 15 Variants and Gene Duplication
2012166	Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants
2002440	EGFR Mutation Detection by Pyrosequencing
2002674	Gastrointestinal Stromal Tumor Mutation (*C-KIT*)—imatinib
2008603	*ERBB2* (HER2/neu) Gene Amplification by FISH, Tissue

TEST #	TEST NAME/DESCRIPTION
2012049	HLA-B*15:02 Genotyping, Carbamazepine Hypersensitivity
2002429	HLA-B*5701 Genotyping for Abacavir Sensitivity
2004680	Interleukin 28B-Associated Variants, IL28B, 2 SNPs
0040248	*KRAS* Mutation Detection
2001932	*KRAS* Mutation Detection with *BRAF* Reflex
0055655	Methylenetetrahydrofolate Reductase (*MTHFR*) 2 Mutations
2008767	Opioid Receptor, Mu 1, *OPRM1* Genotype, 1 Variant
2002871	*PML-RARA* Translocation, t(15;17) by RT-PCR, Quantitative
2008426	Statin Sensitivity (*SLCO1B1*), 1 Variant
2012233	Thiopurine Methyltransferase (*TPMT*) Genotyping, 4 Variants
0051332	UDP Glucuronosyltransferase 1A1 (*UGT1A1*) Genotyping—irinotecan

HEMATOLOGIC DISORDERS

TEST #	TEST NAME/DESCRIPTION
0051495	Alpha Thalassemia (*HBA1* and *HBA2*) 7 Deletions
2001582	Alpha Thalassemia *HBA1* and *HBA2* Sequencing
2011622	Alpha Globin (*HBA1* and *HBA2*) Deletion/Duplication
0051421	Beta Globin Gene Mutations for *HbS*, *HbC*, and *HbE* by PCR
0050578	Beta Globin (*HBB*) Sequencing
2010117	‡Beta Globin (*HBB*) Sequencing and Deletion/Duplication
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing
0080135	Glucose-6-Phosphate Dehydrogenase (enzyme)
0051684	Glucose-6-Phosphate Dehydrogenase (*G6PD*) 2 Mutations
0049090	Heinz Body Stain

TEST #	TEST NAME/DESCRIPTION
2005792	Hemoglobin Evaluation Reflexive Cascade
0050610	Hemoglobin Evaluation with Reflex to Electrophoresis and/or RBC Solubility
0081348	Hemoglobin F
0050613	Hemoglobin (Hb) A2 and F by Column
2004686	Hemoglobin Lepore (*HBD-HBB* Fusion) 3 Mutations
0050520	Hemoglobin S, Evaluation with Reflex to RBC Solubility
2013399	Hemoglobin S, Sickle Solubility
0049020	Hemoglobin, Unstable
2002984	Oxygen Dissociation (P50) by Hemoximetry
0080290	Pyruvate Kinase
2002970	von Hippel-Lindau (*VHL*) Sequencing

+ 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, **www.aruplab.com**, for more detailed test information.

ARUP LABORATORIES

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