



# molecular testing

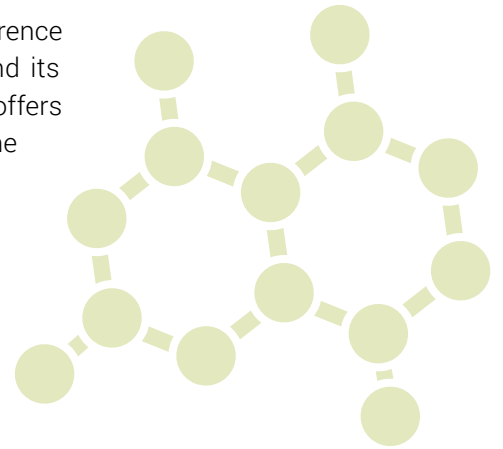
PATIENTS.ANSWERS.RESULTS.





## ARUP LABORATORIES

ARUP Laboratories is a national clinical and anatomic pathology reference laboratory and a nonprofit enterprise of the University of Utah and its Department of Pathology. Located in Salt Lake City, Utah, ARUP offers in excess of 3,000 tests and test combinations, ranging from routine screening tests to esoteric molecular and genetic assays. Rather than competing with its clients for physician office business, ARUP chooses instead to support clients' existing test menus by offering complex and unique tests, with accompanying consultative support, to enhance their abilities to provide local and regional laboratory services. ARUP's clients include many of the nation's university teaching hospitals and children's hospitals, as well as multihospital groups, major commercial laboratories, group purchasing organizations, military and other government facilities, and major clinics. 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®.



Since its formation in 1984 by the Department of Pathology at the University of Utah, ARUP has founded its reputation on reliable and consistent laboratory testing and service. This simple strategy contributes significantly to client satisfaction. When ARUP conducts surveys, clients regularly rate ARUP highly and respond that they would recommend ARUP to others.

As the most responsive source of quality information and knowledge, ARUP strives to be the reference

laboratory of choice for community healthcare systems. ARUP helps its clients meet the customized needs of their unique communities. **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.**



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## patients. answers. results.

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

The ARUP Integrated Oncology and Genetics Division provides a comprehensive test menu to assist physicians in the diagnosis of patients with inherited genetic disorders and offers diagnostic testing, carrier screening for common genetic conditions, fetal testing, presymptomatic testing, molecular confirmation of abnormal newborn screening results, and assessment for genetic variants affecting drug metabolism. In addition, molecular diagnostics is an important component of clinical oncology, supplying pertinent information for diagnosis, prognosis, and prediction of response to tailored chemotherapeutic agents.

The ARUP Infectious Disease Division offers an extensive menu, with full-service analytical capabilities in virology,

microbiology, parasitology, bacteriology, mycology, mycobacteriology, epidemiologic typing, and susceptibility testing. ARUP has the capability and expertise to perform an impressive range of testing, from the most routine bacterial cultures and serologic antibody assays to the latest in molecular-based techniques, including viral-load testing, microorganism identification by 16s rDNA sequencing, and viral genotyping.

ARUP leads the clinical diagnostics field by offering both traditional and cutting-edge technologies in genetics, infectious disease, and oncology testing, and continually expands these test menus as new procedures and clinically relevant targets are discovered.

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## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2008604	1p/19q Deletion by FISH	2001961	Angelman Syndrome ( <i>UBE3A</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2002298	20q Deletion ( <i>D20S108</i> ) del(20)(q12), Chromosome FISH, Interphase	0050392	Ankylosing Spondylitis ( <i>HLAB27</i> ) Genotyping
2007228	5-Fluorouracil (5-FU) Toxicity and Chemotherapeutic Response, 7 Mutations	2006540	Aortopathy Panel, Sequencing and Deletion/Duplication
2002298	5q Deletion ( <i>EGRT</i> )/Monosomy 5 del(5)(q31)/-5, Chromosome FISH, Interphase	2006617	Aortopathy Sequencing, 17 Genes
2002298	7q Deletion ( <i>D7S486</i> )/Monosomy 7 del(7)(q31)/-7, Chromosome FISH, Interphase	2006546	Aortopathy Deletion/Duplication, 17 Genes
0051266	Achondroplasia ( <i>FGFR3</i> ) 2 Mutations <sup>‡</sup>	2001961	Aortopathy ( <i>ACTA2</i> , <i>COL3A1</i> , <i>FBN1</i> , <i>FBN2</i> , <i>MYH11</i> , <i>MYLK</i> , <i>SMAD3</i> , <i>TGFBR1</i> , and <i>TGFBR2</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2002647	Acute Lymphocytic Leukemia (ALL) Panel by FISH, Adult	0030192	APC Resistance Profile with Reflex to Factor V Leiden
2002719	Acute Lymphocytic Leukemia (ALL) Panel by FISH, Pediatric	0055654	Apolipoprotein B Mutation Detection
2011132	Acute Myeloid Leukemia Panel by FISH	0055566	Apolipoprotein E Mutation Detection for Cardiovascular Risk
2002653	Acute Myelogenous Leukemia (AML) with Myelodysplastic Syndrome (MDS) or Therapy-Related AML by FISH	2011144	Arginine: Glycine Amidinotransferase ( <i>GATM</i> ) Deficiency Sequencing
2011906	Adrenoleukodystrophy, X-Linked ( <i>ABCD1</i> ) Sequencing and Deletion/Duplication	2006216	Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel, 7 Genes
2011902	Adrenoleukodystrophy, X-Linked ( <i>ABCD1</i> ) Sequencing	2011958	Ashkenazi Jewish ( <i>BRCA1</i> and <i>BRCA2</i> ) 3 Mutations
2011880	Adrenoleukodystrophy, X-Linked ( <i>ABCD1</i> ) Deletion/Duplication	0051415	Ashkenazi Jewish Panel (Bloom, Canavan, Familial Dysautonomia, Fanconi Anemia Group C, Gaucher, Mucopolidosis IV, Niemann-Pick Type A, and Tay-Sachs)
2011151	Agammaglobulinemia Panel, Sequencing (9 Genes) and Deletion/Duplication (6 Genes) Deletion/Duplication (6 Genes)	2008471	<i>ATP7A</i> -Related Copper Transport Disorders ( <i>ATP7A</i> ) Sequencing and Deletion/Duplication
2006230	Alagille Syndrome ( <i>JAG1</i> ) by Targeted Sequencing and Deletion/Duplication	2007872	<i>ATP7A</i> -Related Copper Transport Disorders ( <i>ATP7A</i> ) Sequencing
2006102	<i>ALK</i> Gene Rearrangements by FISH, Lung	2008443	<i>ATP7A</i> -Related Copper Transport Disorders ( <i>ATP7A</i> ) Deletion/Duplication
2011622	Alpha Globin ( <i>HBA1</i> and <i>HBA2</i> ) Deletion/Duplication	2001961	* <i>ATP7A</i> -Related Copper Transport Disorders ( <i>ATP7A</i> ) Sequencing, Familial Mutation
2011708	Alpha Globin ( <i>HBA1</i> and <i>HBA2</i> ) Sequencing and Deletion/Duplication	2006193	B-Cell Clonality Screening (IgH and IgK) by PCR
2001582	Alpha Thalassemia <i>HBA1</i> and <i>HBA2</i> Sequencing	2002298	<i>BCL6</i> Rearrangement 3q27, Chromosome FISH, Interphase
2001961	Alpha Thalassemia <i>HBA1</i> and <i>HBA2</i> Sequencing, Familial Mutation	2002298	<i>BCR-ABL1</i> Fusion, t(9;22)(q34;q11.2), Chromosome FISH, Interphase
0051495	Alpha Thalassemia <i>HBA1</i> and <i>HBA2</i> Gene Deletions <sup>‡</sup>	2008420	<i>BCR-ABL1</i> Mutation Analysis by Next Generation Sequencing
0051256	Alpha-1-Antitrypsin and A1A Genotype with Reflex to Phenotype	2005017	<i>BCR-ABL1</i> Major (p210), Quantitative
0051786	Alport Syndrome ( <i>COL4A5</i> ) Sequencing	2005016	<i>BCR-ABL1</i> Minor (p190), Quantitative
2001961	Alport Syndrome ( <i>COL4A5</i> ) Sequencing, Familial Mutation <sup>‡</sup>	2005010	<i>BCR-ABL1</i> , Qualitative, with Reflex to <i>BCR-ABL1</i> , Quantitative
2002394	Alport Syndrome, X-linked ( <i>COL4A5</i> ) Deletion/Duplication	0050578	Beta Globin ( <i>HBB</i> ) Sequencing <sup>‡</sup>
2002398	Alport Syndrome, X-linked ( <i>COL4A5</i> ) Sequencing and Deletion/Duplication	2001961	Beta Globin ( <i>HBB</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2011152	Amyotrophic Lateral Sclerosis (ALS) Panel, Sequencing and Deletion/Duplication, 11 Genes	0051421	Beta Globin Gene Mutations for <i>HbS</i> , <i>HbC</i> , and <i>HbE</i> by PCR <sup>‡</sup>
2005077	Angelman Syndrome and Prader-Willi Syndrome by Methylation <sup>‡</sup>	0051288	Beta-2-Adrenergic Receptor Haplotyping
2005564	Angelman Syndrome ( <i>UBE3A</i> ) Sequencing	0051700	Biotinidase Deficiency ( <i>BTD</i> ) 5 Mutations
		0051730	Biotinidase Deficiency ( <i>BTD</i> ) Sequencing



## molecular genetic and oncology testing

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2001961	Biotinidase Deficiency ( <i>BTD</i> ) Sequencing, Familial Mutation, Targeted Sequencing	2009331	Cerebral Caverosus Malformation ( <i>CCM</i> ) Sequencing, 3 Genes
0055691	<i>BIRC2-MALT1 (API2-MALT1)</i> Translocation, t(11;18) by RT-PCR	2009326	Cerebral Caverosus Malformation ( <i>CCM</i> ) Panel, Sequencing and Deletion/Duplication, 3 Genes
0051433	Bloom ( <i>BLM</i> ) 2281del6/ins 7 Mutations <sup>‡</sup>	2003172	Cerebral Caverosus Malformation ( <i>CCM1, CCM2, and CCM3</i> ) Deletion/Duplication
2012222	Bone Marrow Failure Sequencing, 35 Genes	2001961	Cerebral Caverosus Malformation ( <i>CCM1, CCM2, and CCM3</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2002498	<i>BRAF</i> Codon 600 Mutation Detection by Pyrosequencing	2006222	CHARGE Syndrome ( <i>CHD7</i> ) Sequencing
0051750	<i>BRAF</i> Codon 600 Mutation Detection with Reflex to <i>MLH1</i> Promoter Methylation	2012155	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies, <i>PMP22</i> Deletion/Duplication with Reflex to Sequencing Panel
2007132	<i>BRAF</i> V600E Mutation Detection in Hairy Cell Leukemia by Real-Time PCR, Quantitative	2012151	Charcot-Marie-Tooth (CMT) and Related Hereditary Neuropathies Panel Sequencing
2012026	Breast and Ovarian Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 20 Genes	2012160	Charcot-Marie-Tooth Type 1A (CMT1A)/Hereditary Neuropathy with Liability to Pressure Palsies (HNPP), <i>PMP22</i> Deletion/Duplication
2011949	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing and Deletion/Duplication	2007545	Childhood-Onset Epilepsy Panel, Sequencing and Deletion/Duplication, 40 Genes
2011954	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Sequencing	2002292	Chromosome Analysis, Bone Marrow
2011915	Breast and Ovarian Hereditary Cancer Syndrome ( <i>BRCA1</i> and <i>BRCA2</i> ) Deletion/Duplication	2002300	Chromosome Analysis, Lymph Node
2006218	Brugada Syndrome Panel, 7 Genes	2002290	Chromosome Analysis, Leukemic Blood
2012032	Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes	2002296	Chromosome Analysis, Solid Tumor
2009323	Cardiomyopathy and Arrhythmia Panel, Sequencing, 85 Genes	2011130	Chromosome FISH, Amniotic Fluid with Reflex to Chromosome Analysis or Genomic Microarray
2010183	Cardiomyopathy and Arrhythmia Panel, Sequencing (85 Genes) and Deletion/Duplication (83 Genes)	2011131	Chromosome FISH, Chorionic Villus with Reflex to Chromosome Analysis or Genomic Microarray
0051453	Canavan Disease ( <i>ASPA</i> ) 4 Mutations <sup>‡</sup>	2006356	Chronic Granulomatous Disease ( <i>CYBB</i> Gene Scanning and <i>NCF1</i> Exon 2 GT Deletion) with Reflex to <i>CYBB</i> Sequencing
2004203	Carnitine Deficiency, Primary ( <i>SLC22A5</i> ) Sequencing and Deletion/Duplication	2006361	Chronic Granulomatous Disease, X-Linked ( <i>CYBB</i> ) Gene Scanning with Reflex to Sequencing
2004199	Carnitine Deficiency, Primary ( <i>SLC22A5</i> ) Deletion/Duplication	2006366	Chronic Granulomatous Disease ( <i>NCF1</i> ) Exon 2 GT Deletion
0051682	Carnitine Deficiency, Primary ( <i>SLC22A5</i> ) Sequencing	2001961	Chronic Granulomatous Disease ( <i>CYBB</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2001961	Carnitine Deficiency, Primary ( <i>SLC22A5</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>	2002295	Chronic Lymphocytic Leukemia (CLL) Panel by FISH
2006224	Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel, 3 Genes	2006261	Citrin Deficiency ( <i>SLC25A13</i> ) Sequencing
2011114	<i>CBFB-MYH11</i> inv(16) Detection, Quantitative	2001961	Citrin Deficiency ( <i>SLC25A13</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2004935	<i>CDKL5</i> -Related Disorders ( <i>CDKL5</i> ) Sequencing and Deletion/Duplication	2007069	Citrullinemia, Type I ( <i>ASS1</i> ) Sequencing
2004931	<i>CDKL5</i> -Related Disorders ( <i>CDKL5</i> ) Sequencing	2011157	Cobalamin/Propionate/Homocysteine Metabolism Related Disorders Panel, Sequencing (25 Genes) and Deletion / Duplication (24 Genes)
2004927	<i>CDKL5</i> -Related Disorders ( <i>CDKL5</i> ) Deletion and Duplication	2011616	Colon Cancer Gene Panel, Somatic
2004247	<i>CEBPA</i> Mutation Detection	2006220	Congenital Amegakaryocytic Thrombocytopenia (CAMT) Sequencing
2005018	Celiac Disease ( <i>HLA-DQA1*05, HLA-DQB1*02, and HLA-DQB1*03:02</i> ) Genotyping	2001933	Cystic Fibrosis ( <i>CFTR</i> ) 32 Mutations <sup>‡</sup>
2001961	<i>CDKL5</i> -Related Disorders Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>		

\* Multiple Tests Available ‡ Fetal Testing Available

## molecular genetic and oncology testing

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2001968	Cystic Fibrosis ( <i>CFTR</i> ) 32 Mutations with Reflex to Sequencing	2001961	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI ( <i>PLOD1</i> ) Sequencing, Familial Mutation <sup>‡</sup>
2001967	Cystic Fibrosis ( <i>CFTR</i> ) 32 Mutations with Reflex to Sequencing and Reflex to Deletion/Duplication	2010193	Endocrine Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 13 Genes
2001969	Cystic Fibrosis ( <i>CFTR</i> ) 32 Mutations, Atypical	2002378	Eosinophilia Panel by FISH
0056003	Cystic Fibrosis ( <i>CFTR</i> ) 5T Mutation	2007914	<i>EPOR</i> Mutation Detection by Sequencing
0051642	Cystic Fibrosis ( <i>CFTR</i> ) Deletion/Duplication	2008603	<i>ERBB2</i> ( <i>HER2/neu</i> ) Gene Amplification by FISH, Tissue
0051110	Cystic Fibrosis ( <i>CFTR</i> ) Sequencing	2002298	<i>ETV6-RUNX1</i> ( <i>TEL-AML1</i> ) Fusion, t(12;21)(p13;q22), Chromosome FISH, Interphase
2001961	Cystic Fibrosis ( <i>CFTR</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>	0051220	Ewing Sarcoma by RT-PCR
0056006	Cystic Fibrosis Cis-Trans ( <i>CFTR</i> ) R117H and 5T Mutations	2007225	<i>EWSR1</i> (22q12) Gene Rearrangement by FISH
0051640	Cystic Fibrosis Sequencing with Reflex to Deletion/Duplication	2002298	<i>EWSR1</i> (22q12) Rearrangement, Chromosome FISH, Interphase
0050098	Cystic Fibrosis, 3199del6 Only	2006336	Exome Sequencing Symptom-Guided Analysis, Patient Only
0051104	Cytochrome P450 2C19 ( <i>CYP2C19</i> ) 9 Mutations	2006332	Exome Sequencing with Symptom-Guided Analysis
0051103	Cytochrome P450 2C9 ( <i>CYP2C9</i> ) 2 Mutations	2006342	Exome Sequencing Control, Family Member
0051232	Cytochrome P450 2D6 ( <i>CYP2D6</i> ) 14 Mutations, 7 Gene Duplication	2007543	Expanded Carrier Screening Panel, 100-Plus Disorders
2006325	Cytogenomic SNP Microarray, Oncology	2007531	Expanded Carrier Screening Panel, 100-Plus Disorders with Fragile X
2010229	Cytogenomic Molecular Inversion Probe Array, FFPE Tissue, Oncology	2008800	Expanded Hearing Loss Panel, Sequencing (56 Genes)
2002298	<i>DDIT3</i> ( <i>CHOP</i> ) (12q13) Gene Rearrangement, Chromosome FISH, Interphase	2008803	Expanded Hearing Loss Panel, Sequencing (56 Genes) and Deletion/Duplication (53 Genes)
2006236	Diamond-Blackfan Anemia ( <i>RPL11</i> ) Sequencing	0097720	Factor V Leiden ( <i>F5</i> ), R506Q Mutation
2006234	Diamond-Blackfan Anemia ( <i>RPL5</i> ) Sequencing	2001549	Factor V, R2 Mutation
2006238	Diamond-Blackfan Anemia ( <i>RPS19</i> ) Sequencing	2003220	Factor XIII ( <i>F13A1</i> ), V34L Mutation
2012166	Dihydropyrimidine Dehydrogenase ( <i>DPYD</i> ), 3 Mutations	2004920	Familial Adenomatous Polyposis ( <i>APC</i> ) Deletion and Duplication
2006226	Dilated Cardiomyopathy (DCM)/Left Ventricular Noncompaction (LVNC) Panel, 27 Genes	2004863	Familial Adenomatous Polyposis ( <i>APC</i> ) Sequencing
2011235	Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication <sup>‡</sup>	2001961	Familial Adenomatous Polyposis ( <i>APC</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2011241	Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication with Reflex to Sequencing	2004915	Familial Adenomatous Polyposis Panel: <i>APC</i> Sequencing, <i>APC</i> Deletion/Duplication, and <i>MYH 2</i> Mutations
2011153	Duchenne/Becker Muscular Dystrophy (DMD) Sequencing	2002658	Familial Mediterranean Fever ( <i>MEFV</i> ) Sequencing
0051463	Dysautonomia, Familial ( <i>IKBKAP</i> ) 2 Mutations <sup>‡</sup>	0051468	Fanconi Anemia Group C ( <i>FANCC</i> ) 2 Mutations <sup>‡</sup>
2006244	Dyskeratosis Congenita, Autosomal ( <i>TERC</i> ) Sequencing	2006069	Febrile Seizures Panel
2006228	Dyskeratosis Congenita, X-linked ( <i>DKC1</i> ) Sequencing	0051752	FG Syndrome, <i>FGS1</i> ( <i>MED12</i> ), R961W Mutation
2008605	<i>EGFR</i> Gene Amplification by FISH	2007883	Filaggrin ( <i>FLG</i> ) 2 mutations
2002440	<i>EGFR</i> Mutation Detection by Pyrosequencing	2005400	<i>FLT3</i> Mutation Detection by PCR
2005559	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI ( <i>PLOD1</i> ) Sequencing and Deletion/Duplication	2011806	<i>FLT3</i> Signal Ratio Mutation Detection by PCR
2005555	Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI ( <i>PLOD1</i> ) Deletion/Duplication	2002298	<i>FOXO1</i> ( <i>FKHR</i> ) (13q13) Gene Rearrangement, Chromosome FISH, Interphase
		2001497	<i>FOXO1</i> ( <i>FKHR</i> ) (13q14) Gene Rearrangement by FISH



## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2002662	Freeman-Sheldon Syndrome ( <i>MYH3</i> ) Sequencing Exon 17	2001961	Hemophilia B ( <i>F9</i> ) Sequencing, Familial Mutation, Targeted Sequencing†
0051176	Galactosemia ( <i>GALT</i> ) 9 Mutations‡	2012052	Hereditary Hemolytic Anemia Sequencing, 28 Genes
0051175	Galactosemia ( <i>GALT</i> ) Enzyme Activity and 9 Mutations	0051348	Hereditary Hemorrhagic Telangiectasia ( <i>ACVRL1</i> and <i>ENG</i> ) Deletion/Duplication
2006697	Galactosemia ( <i>GALT</i> ) Sequencing	0051381	Hereditary Hemorrhagic Telangiectasia ( <i>ACVRL1</i> and <i>ENG</i> ) Sequencing
2001961	Galactosemia ( <i>GALT</i> ) Sequencing, Familial Mutation, Targeted Sequencing†	0051382	Hereditary Hemorrhagic Telangiectasia ( <i>ACVRL1</i> and <i>ENG</i> ) Sequencing and Deletion/Duplication
2002674	Gastrointestinal Stromal Tumor Mutation ( <i>C-KIT</i> )	2001961	Hereditary Hemorrhagic Telangiectasia ( <i>ACVRL1</i> and <i>ENG</i> ) Sequencing, Familial Mutation, Targeted Sequencing†
0051438	Gaucher ( <i>GBA</i> ) 8 Mutations‡	2011461	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHA</i> ) Sequencing
0051476	Glaucoma, Primary Congenital ( <i>CYP1B1</i> ) Sequencing	2007108	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> ) Sequencing and Deletion/Duplication
2001961	Glaucoma, Primary Congenital ( <i>CYP1B1</i> ) Sequencing, Familial Mutation, Targeted Sequencing	2007117	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHC</i> ) Sequencing and Deletion/Duplication
2011465	<i>GLI3</i> -Related Disorders ( <i>GLI3</i> ) Sequencing and Deletion/Duplication	2007122	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHD</i> ) Sequencing and Deletion/Duplication
2011470	<i>GLI3</i> -Related disorders ( <i>GLI3</i> ) Sequencing	2007113	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> , <i>SDHC</i> , and <i>SDHD</i> ) Deletion/Duplication
2011424	<i>GLI3</i> -Related Disorders ( <i>GLI3</i> ) Deletion/Duplication	2007167	Hereditary Paraganglioma-Pheochromocytoma ( <i>SDHB</i> , <i>SDHC</i> , and <i>SDHD</i> ) Sequencing and Deletion/Duplication Panel
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency ( <i>G6PD</i> ) Sequencing	2005408	Hereditary Persistence of Fetal Hemoglobin (HPFH) 8 Mutations
0051684	Glucose-6-Phosphate Dehydrogenase ( <i>G6PD</i> ) 2 Mutations, African Alleles	2012049	HLA-B*15:02 Genotyping, Carbamazepine Hypersensitivity
2011140	Guanidinoacetate Methyltransferase ( <i>GAMT</i> ) Deficiency Sequencing	2002429	HLA-B*5701 Genotyping
2001992	Hearing Loss, Nonsyndromic Panel ( <i>GJB2</i> ) Sequencing, ( <i>GJB6</i> ) 2 Deletions and Mitochondrial DNA 2 Mutations	2011264	HLA Class I (ABC) by Next Generation Sequencing
0051374	Hearing Loss, Nonsyndromic, Connexin 26 ( <i>GJB2</i> ) Sequencing	2011272	HLA Class II ( <i>DRB1</i> and <i>DQB1</i> ) by Next Generation Sequencing
2001961	Hearing Loss, Nonsyndromic, Connexin 26 ( <i>GJB2</i> ) Sequencing, Familial Mutation, Targeted Sequencing	0051650	HNPCC/Lynch Syndrome ( <i>MLH1</i> ) Sequencing and Deletion/Duplication
2001956	Hearing Loss, Nonsyndromic, Connexin 30 ( <i>GJB6</i> ) 2 Deletions	0051654	HNPCC/Lynch Syndrome ( <i>MSH2</i> ) Sequencing and Deletion/Duplication
2002044	Hearing Loss, Nonsyndromic, Mitochondrial DNA 2 Mutations	0051656	HNPCC/Lynch Syndrome ( <i>MSH6</i> ) Sequencing and Deletion/Duplication
0055656	Hemochromatosis, Hereditary ( <i>HFE</i> ) 3 Mutations	0051737	HNPCC/Lynch Syndrome ( <i>PMS2</i> ) Sequencing and Deletion/Duplication
2005792	Hemoglobin Evaluation Reflexive Cascade	2006892	HNPCC/Lynch Syndrome ( <i>PMS2</i> ) Sequencing
2004686	Hemoglobin Lepore ( <i>HBD-HBB</i> Fusion) 3 Mutations	2001728	HNPCC/Lynch Syndrome Deletion/Duplication— <i>MLH1</i> , <i>MSH2</i> , <i>MSH6</i> , or <i>PMS2</i>
2001759	Hemophilia A ( <i>F8</i> ) 2 Inversions‡	2001961	HNPCC/Lynch Syndrome, Targeted Sequencing, Familial Mutation, Targeted Sequencing
2001614	Hemophilia A ( <i>F8</i> ) 2 Inversions with Reflex to Sequencing and Reflex to Deletion/Duplication	0040018	Huntington Disease ( <i>HD</i> ) Mutation with Reflex to Southern Blot
2001751	Hemophilia A ( <i>F8</i> ) Deletion/Duplication	2011154	Hyper IgM Syndrome Panel, Sequencing (12 Genes) and Deletion/Duplication (10 Genes)
2001747	Hemophilia A ( <i>F8</i> ) Sequencing		
2001961	Hemophilia A ( <i>F8</i> ) Sequencing, Familial Mutation, Targeted Sequencing†		
2001578	Hemophilia B ( <i>F9</i> ) Sequencing		
2010494	Hemophilia B ( <i>F9</i> ) Sequencing and Deletion/Duplication		
2010499	Hemophilia B ( <i>F9</i> ) Deletion/Duplication		

\* Multiple Tests Available ‡ Fetal Testing Available

## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2002298	Hyperdiploidy with Trisomy 4 and 10, Chromosome FISH, Interphase	0051644	Kell Antigen Genotyping ( <i>KEL1-KEL2</i> )
0051367	Hypochondropiasia ( <i>FGFR3</i> ) 2 Mutations	0040137	<i>KIT</i> (D816V) Mutation by PCR
2006444	<i>IDH1</i> and <i>IDH2</i> Mutation Analysis, Exon 4	2002437	<i>KIT</i> Mutations in AML by Fragment Analysis and Sequencing
2001536	<i>IGH-BCL2</i> Fusion, t(14;18) by FISH	2002695	<i>KIT</i> Mutations, Melanoma
2002298	<i>IGH-BCL2</i> Fusion, t(14;18)(q32;q21), Chromosome FISH, Interphase	0040248	<i>KRAS</i> Mutation Detection
0055557	<i>IGH-CCND1</i> ( <i>BCL1-JH</i> ) Translocation, t(11;14) by PCR	2001932	<i>KRAS</i> Mutation Detection with Reflex to <i>BRAF</i> Codon 600 Mutation Detection
2007226	<i>IGH-CCND1</i> Fusion, t(11;14) by FISH	2008347	Legius Syndrome ( <i>SPRED1</i> ) Sequencing and Deletion/Duplication
2002298	<i>IGH-CCND1</i> Fusion, t(11;14)(q13;q32), Chromosome FISH, Interphase	2002945	Legius Syndrome ( <i>SPRED1</i> ) Sequencing
2001538	<i>IGH-MYC</i> Fusion, t(8;14) by FISH	2008373	Legius Syndrome ( <i>SPRED1</i> ) Deletion/Duplication
2002298	<i>IGH</i> Rearrangement, Chromosome FISH, Interphase	2001961	Legius Syndrome ( <i>SPRED1</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2002298	<i>IGH</i> Rearrangement 14q32, Chromosome FISH, Interphase	2004543	<i>LMNA</i> -Related Disorders ( <i>LMNA</i> ) Sequencing
0040227	<i>IGHV</i> Mutation Analysis by Sequencing	2004539	<i>LMNA</i> -Related Disorders ( <i>LMNA</i> ) Deletion/Duplication
2007535	Infantile Epilepsy Panel, Sequence Analysis and Exon-Level Deletion/Duplication, 38 Genes	2001961	<i>LMNA</i> -Related Disorders ( <i>LMNA</i> ), Familial Mutation, Targeted Sequencing†
2006274	Inherited Insulin Resistance Syndromes ( <i>INSR</i> ) Sequencing	2002705	Loeys-Dietz Syndrome ( <i>TGFBR1</i> and <i>TGFBR2</i> ) Sequencing
2001961	Inherited Insulin Resistance Syndromes ( <i>INSR</i> ) Sequencing, Familial Mutation, Targeted Sequencing	2006232	Long QT Syndrome (LQTS) Panel, 12 Genes
2006344	Inosine Triphosphate ( <i>ITPA</i> ) and Interleukin 28 B ( <i>IL28B</i> )-Associated Variants, 4 SNPs	2011612	Lung Cancer Comprehensive Mutation and Translocation Panel by Next Generation Sequencing
2004680	Interleukin 28 B ( <i>IL28B</i> )-Associated Variants, 2 SNPs	2008894	Lung Cancer Limited Panel
2002357	<i>JAK2</i> Exon 12 Mutation Analysis by PCR	2008895	Lung Cancer Limited Panel with <i>KRAS</i>
0051245	<i>JAK2</i> Gene, V617F Mutation, Qualitative	2002650	Lymphoma (Aggressive) Panel by FISH
0040168	<i>JAK2</i> Gene, V617F Mutation, Quantitative	2005584	Marfan Syndrome, <i>FBN1</i> Sequencing and Deletion/Duplication
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	2005589	Marfan Syndrome, <i>FBN1</i> Sequencing
2012085	<i>JAK2</i> Gene, V617F Mutation, Qualitative with Reflex to <i>JAK2</i> Exon 12 Mutation Analysis by PCR	2005580	Marfan Syndrome, <i>FBN1</i> Deletion/Duplication
2004984	Juvenile Polyposis Syndrome ( <i>BMPR1A</i> ) Deletion/Duplication	2001961	Marfan Syndrome, <i>FBN1</i> Sequencing, Familial Mutation, Targeted Sequencing†
2004988	Juvenile Polyposis Syndrome ( <i>BMPR1A</i> ) Sequencing	2003016	<i>MDM2</i> Gene Amplification by FISH
2004992	Juvenile Polyposis Syndrome ( <i>BMPR1A</i> ) Sequencing and Deletion/Duplication	0051205	Medium Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADM</i> ) 2 Mutations
2001961	Juvenile Polyposis ( <i>BMPR1A</i> ) Sequencing, Familial Mutation, Targeted Sequencing†	0051758	Medium Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADM</i> ) Sequencing
2001976	Juvenile Polyposis ( <i>SMAD4</i> ) Deletion/Duplication	2001961	Medium Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADM</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2001971	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing and Deletion/Duplication	2010209	Melanoma Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 6 Genes
0051510	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing	0055655	Methylenetetrahydrofolate Reductase ( <i>MTHFR</i> ) 2 Mutations
2001961	Juvenile Polyposis ( <i>SMAD4</i> ) Sequencing, Familial Mutation, Targeted Sequencing†	0051740	Microsatellite Instability (MSI), HNPCC/Lynch Syndrome by PCR

## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2002327	Mismatch Repair by IHC with Reflex to <i>BRAF</i> Codon 600 Mutation and <i>MLH1</i> Promoter Methylation	0051458	Niemann-Pick, Type A ( <i>SMPD1</i> ) 4 Mutations <sup>‡</sup>
2006050	Mitochondrial Disorders (108 Nuclear Genes) Sequencing	2007537	Non-Invasive Prenatal Testing for Fetal Aneuploidy
2006065	Mitochondrial Disorders (mtDNA) Sequencing	2004189	Noonan Syndrome ( <i>PTPN11</i> ) Sequencing with Reflex to <i>SOS1</i> Sequencing
2006878	Mitochondrial Disorders (108 Nuclear Genes) Sequencing and Deletion/Duplication	0051805	Noonan Syndrome ( <i>PTPN11</i> ) Sequencing
2006872	Mitochondrial Disorders (mtDNA) Sequencing and Deletion/Duplication	2004195	Noonan Syndrome ( <i>SOS1</i> ) Sequencing
2006054	Mitochondrial Disorders Panel (mtDNA and 108 Nuclear Genes) Sequencing and Deletion/Duplication	2001961	Noonan Syndrome ( <i>SOS1</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2006061	Mitochondrial Genome (mtDNA and 108 Nuclear Genes) Deletion/Duplication	0040174	<i>NPM1</i> Mutation by PCR and Fragment Analysis
2002499	<i>MLH1</i> Promoter Methylation, Paraffin	2003123	<i>NRAS</i> Mutation Detection by Pyrosequencing
2002298	<i>MLL</i> Rearrangement 11q23, Chromosome FISH, Interphase	2004896	Ornithine Transcarbamylase Deficiency ( <i>OTC</i> ) Sequencing and Deletion/Duplication
0051755	Molar Pregnancy, 16 DNA Markers	2004901	Ornithine Transcarbamylase Deficiency ( <i>OTC</i> ) Sequencing
2005545	<i>MPL</i> Codon 515 Mutation Detection by Pyrosequencing, Quantitative	2004892	Ornithine Transcarbamylase Deficiency ( <i>OTC</i> ) Deletion/Duplication
0051448	Mucopolipidosis Type IV ( <i>MCOLN1</i> ) 2 Mutations <sup>‡</sup>	2001961	Ornithine Transcarbamylase Deficiency ( <i>OTC</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2005346	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Deletion/Duplication	2002016	Pancreatitis, Hereditary ( <i>PRSS1</i> ) Sequencing
2005359	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing	2001961	Pancreatitis, Hereditary ( <i>PRSS1</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2005360	Multiple Endocrine Neoplasia ( <i>MEN1</i> ) Sequencing and Deletion/Duplication	2002012	Pancreatitis, Idiopathic ( <i>SPINK1</i> ) Sequencing
0051390	Multiple Endocrine Neoplasia Type 2 (MEN2), <i>RET</i> Gene Mutations by Sequencing	2001961	Pancreatitis, Idiopathic ( <i>SPINK1</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2001961	Multiple Endocrine Neoplasia Type 2 ( <i>RET</i> ) Sequencing, Familial Mutation, Targeted Sequencing	2001961	Pancreatitis ( <i>CTRC</i> ), Familial Mutation, Targeted Sequencing
2002294	Multiple Myeloma Panel by FISH	2010703	Pancreatitis ( <i>CTRC</i> ) Sequencing
2006307	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations with Reflex to Sequencing	2002528	Pancreatobiliary FISH
2006191	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) Sequencing	0040113	<i>PAX-FOXO1</i> and <i>PAX7-FOXO1</i> Translocation by RT-PCR
2004911	<i>MUTYH</i> -Associated Polyposis ( <i>MUTYH</i> ) 2 Mutations	2002298	<i>PDGFRA</i> , Chromosome FISH, Interphase
2002345	<i>MYC</i> (8q24) Gene Rearrangement by FISH	2002298	<i>PDGFRA-FIP1L1</i> Fusion ( <i>CHIC2</i> Deletion), Chromosome FISH, Interphase
2002298	<i>MYC</i> Rearrangement 8q24, Chromosome FISH, Interphase	2002298	<i>PDGFRB</i> , Chromosome FISH, Interphase
2007227	<i>MYCN</i> ( <i>N-MYC</i> ) Gene Amplification by FISH	2002298	<i>PDGFRB</i> Rearrangement 5q33.1, Chromosome FISH, Interphase
2002709	Myelodysplastic Syndrome (MDS) Panel by FISH	2007366	Periodic Fever Syndromes Deletion/Duplication, 6 Genes
2011117	Myeloid Malignancies Mutation Panel by Next Generation Sequencing	2007370	Periodic Fever Syndromes Panel, Sequencing (7 Genes) and Deletion/Duplication, (6 Genes)
2012182	Myeloid Malignancies Somatic Mutation and Copy Number Analysis Panel	2007376	Periodic Fever Syndromes Sequencing, 7 Genes
2002360	Myeloproliferative Disorders Panel by FISH	2008398	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing and Deletion/Duplication
2005023	Narcolepsy ( <i>HLA-DQB1*06:02</i> ) Genotyping	2008394	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing
2007159	Neurofibromatosis Type 1 ( <i>NF1</i> ) Sequencing	2008377	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Deletion/Duplication
2007154	Neurofibromatosis Type 1 ( <i>NF1</i> ) Sequencing and Deletion/Duplication	2001961	Peutz-Jeghers Syndrome ( <i>STK11</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>

\* Multiple Tests Available ‡ Fetal Testing Available

## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2004510	<i>PIK3CA</i> Mutation Detection	2007091	Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Sequencing and Deletion/Duplication, 53 Genes
2004980	Plasminogen Activator Inhibitor-1, PAI-1 ( <i>SERPINE1</i> ) Genotyping	2007095	Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Deletion/Duplication, 53 Genes
*	Platelet Antigen Genotyping: HPA 1, 2, 3, 4, 5, 6, and 15 (ordered individually or as panel)	0051614	Rett Syndrome ( <i>MECP2</i> ) Sequencing and Deletion/Duplication
2002363	<i>PML-RARa</i> Translocation by FISH	0051618	Rett Syndrome ( <i>MECP2</i> ) Deletion and Duplication
2002871	<i>PML-RARa</i> Translocation, t(15;17) by RT-PCR, Quantitative	0051378	Rett Syndrome ( <i>MECP2</i> ) Sequencing
2012250	Polycystic Kidney Disease, Autosomal Dominant ( <i>PKD1</i> and <i>PKD2</i> ) Sequencing and Deletion/Duplication	2001961	Rett Syndrome ( <i>MECP2</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>
2012255	Polycystic Kidney Disease, Autosomal Dominant ( <i>PKD1</i> and <i>PKD2</i> ) Sequencing	*	Rh Genotyping: C/c, D, and E/e (ordered individually)
2012246	Polycystic Kidney Disease, Autosomal Dominant ( <i>PKD1</i> and <i>PKD2</i> ) Deletion/Duplication	2008418	<i>ROS1</i> by FISH
2007539	Prenatal Carrier Screening Panel, 85 Disorders	2002298	<i>RUNX1-RUNX1T1 (AML1-ETO)</i> Fusion, t(8;21)(q22;q22), Chromosome FISH, Interphase
2007541	Prenatal Carrier Screening Panel, 85 Disorders with Fragile X	2010138	<i>RUNX1-RUNX1T1 (AML1-ETO)</i> t(8;21) Detection, Quantitative
2011156	Primary Antibody Deficiency Panel, Sequencing (35 Genes) and Deletion/Duplication (26 Genes)	2006240	Shwachman-Diamond Syndrome ( <i>SBDS</i> ) Sequencing
0051476	Primary Congenital Glaucoma ( <i>CYP1B1</i> ) Sequencing	2005896	<i>SCN1A</i> -Related Seizure Disorders ( <i>SCN1A</i> ), Sequencing and Deletion/Duplication
2007533	Progressive Myoclonic Epilepsy Panel, Sequence Analysis and Exon-Level Deletion/Duplication, 12 Genes	2003243	Septin 9 ( <i>SEPT9</i> ) Methylated DNA Detection by Real-Time PCR
0056060	Prothrombin ( <i>F2</i> ) G20210A Mutation	2006242	Short QT Syndrome Panel, 3 Genes
2002726	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Deletion/Duplication	2011457	Smith-Lemli-Opitz Syndrome ( <i>DHCR7</i> ) Sequencing <sup>‡</sup>
2002722	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing	2012015	Skeletal Dysplasia Panel, Sequencing (39 Genes) and Deletion/Duplication (35 Genes) <sup>‡</sup>
2002470	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing and Deletion/Duplication	2012018	Skeletal Dysplasia Panel, Sequencing 39 Genes
2001961	<i>PTEN</i> -Related Disorders ( <i>PTEN</i> ) Sequencing, Familial Mutation, Targeted Sequencing	2012007	Skeletal Dysplasia Panel, Deletion/Duplication 35 Genes
2003405	Pulmonary Arterial Hypertension ( <i>BMPR2</i> ) Sequencing and Deletion/Duplication	2007991	Solid Tumor Mutation Panel by Next Generation Sequencing
2003410	Pulmonary Arterial Hypertension ( <i>BMPR2</i> ) Sequencing	2007222	<i>SS18 (SYT)</i> (18q11) Gene Rearrangement by FISH
2003401	Pulmonary Arterial Hypertension ( <i>BMPR2</i> ) Deletion/Duplication	2002298	<i>SS18 (SYT)</i> (18q11) Gene Rearrangement, Chromosome FISH, Interphase
2001961	Pulmonary Arterial Hypertension ( <i>BMPR2</i> ) Sequencing, Familial Mutation, Targeted Sequencing <sup>‡</sup>	0040114	<i>SYT-SSX</i> , t(18;X) Translocations by RT-PCR
2002730	<i>RASA1</i> -Related Disorders ( <i>RASA1</i> ) Sequencing	2007569	TACI-Associated Common Variable Immunodeficiency ( <i>TNFRSF13B</i> ) Sequencing
2007852	<i>RASA1</i> -Related Disorders ( <i>RASA1</i> ) Sequencing and Deletion/Duplication	2001961	*TACI-Associated Common Variable Immunodeficiency ( <i>TNFRSF13B</i> ) Sequencing, Familial Mutation, Targeted Sequencing
2007830	<i>RASA1</i> -Related Disorders ( <i>RASA1</i> ) Deletion/Duplication	2008409	T-Cell Clonality by Next Generation Sequencing
2001961	<i>RASA1</i> -Related Disorders ( <i>RASA1</i> ) Sequencing, Familial Mutation, Targeted Sequencing	0055567	T-Cell Clonality Screening by PCR
2010214	Renal Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 15 Genes	0051428	Tay-Sachs ( <i>HEXA</i> ) 7 Mutations <sup>‡</sup>
2007085	Retinitis Pigmentosa/Leber Congenital Amaurosis Panel, Sequencing and Deletion/Duplication, 53 Genes	2002298	<i>TCF3 (E2A)</i> Rearrangement 19p13, Chromosome FISH, Interphase
		0051506	Thanatophoric Dysplasia, Types I and II ( <i>FGFR3</i> ) 13 Mutations <sup>‡</sup>
		2012233	Thiopurine Methyltransferase ( <i>TPMT</i> ) Genotyping, 4 Variants



## molecular genetic and oncology testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
0056200	Thrombotic Risk, DNA Panel	2001961	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing, Familial Mutation, Targeted Sequencing†
2006385	Thrombotic Risk, Reflexive Panel	2002988	von Hippel-Lindau (VHL) Deletion/Duplication
0030133	Thrombotic Risk, Inherited Etiologies (Most Common) with Reflex to Factor V Leiden	2002970	von Hippel-Lindau (VHL) Sequencing
2002298	Trisomy 8, Chromosome FISH, Interphase	2002965	von Hippel-Lindau (VHL) Sequencing and Deletion/Duplication
2002298	Trisomy 9, Chromosome FISH, Interphase	2005480	von Willebrand Disease, Type 2A (VWF) Sequencing Exon 28 with Reflex to 9 Exons
2002298	Trisomy 12, Chromosome FISH, Interphase	2005486	von Willebrand Disease, Type 2B (VWF) Sequencing
0050547	Twin Zygosity Testing	2005490	von Willebrand Disease, Type 2M (VWF) Sequencing
0051332	UDP-Glucuronosyltransferase 1A1 (UGT1A1) Genotyping	2005494	von Willebrand Disease, Type 2N (VWF) Sequencing
2001181	UroVysion FISH	2005476	von Willebrand Disease, Platelet Type (GP1BA) 4 Mutations
2007384	Vascular Malformations Panel Sequencing and Deletion/Duplication, 14 Genes	2001961	von Willebrand Disease, Type 2A, 2B, 2M, and 2N (VWF) Sequencing, Familial Mutation, Targeted Sequencing†
2007390	Vascular Malformations Panel Sequencing, 14 Genes	0051370	Warfarin Sensitivity (CYP2C9 and VKORC1) 3 Mutations
2007380	Vascular Malformations Panel Deletion/Duplication, 14 Genes	2004358	Warfarin Genotyping Plus
2004212	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing and Deletion/Duplication	2005766	WT1 Mutations by Sequencing
2002001	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing	2006352	X-Chromosome Inactivation Analysis
2004208	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Deletion/Duplication	2001778	Y Chromosome Microdeletion

## molecular infectious disease—viral testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2007192	Adenovirus, Quantitative PCR	0060041	Herpes Simplex Virus by PCR
2007473	Adenovirus, Qualitative, PCR	2010095	Herpes Simplex Virus (HSV-1/HSV-2) Subtype by PCR
0090067	BK Virus, Quantitative PCR	0060071	Herpesvirus 6 (HHV6) (A and B), Quantitative PCR
2002304	BK Virus, Quantitative PCR, Blood	2008440	Herpesvirus 8 (HHV-8) DNA, Quantitative Real-Time PCR
2002310	BK Virus, Quantitative PCR, Urine	2002996	Herpes Virus 8 DNA, Qualitative Real-Time PCR
0060040	Cytomegalovirus by PCR	2011942	High Risk by PCR, SurePath
2004760	Cytomegalovirus Antiviral Drug Resistance by Sequencing	0060784	Human Metapneumovirus by RT-PCR
0060031	Cytomegalovirus by PCR, Whole Blood or Bone Marrow	2011937	Human Papillomavirus (HPV) 16 and 18 Genotype by PCR, SurePath
0051813	Cytomegalovirus, Quantitative PCR	2007894	Human Papillomavirus (HPV) Genotypes 16 and 18/45 by Transcription-Mediated Amplification (TMA), ThinPrep
2006966	Cytomegalovirus, Quantitative PCR with Reflex to Drug Resistance Testing by Sequencing	0065999	Human Papillomavirus (HPV), High Risk by Hybrid Capture, Cervical Brush
2005730	Enterovirus and Parechovirus Detection by RT-PCR	0060744	Human Papillomavirus (HPV), High Risk by Hybrid Capture, SurePath
0050249	Enterovirus Detection by RT-PCR	2008404	Human Papillomavirus (HPV), High Risk by Hybrid Capture, ThinPrep
0050246	Epstein-Barr Virus by PCR	2011947	Human Papillomavirus (HPV), High Risk by PCR, ThinPrep
0051352	Epstein-Barr Virus, Quantitative PCR		
0051353	Epstein-Barr Virus, Quantitative PCR, Whole Blood		

\* Multiple Tests Available ‡ Fetal Testing Available

## molecular infectious disease—viral testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2007890	Human Papillomavirus (HPV), High Risk by Transcription-Mediated Amplification (TMA) with Reflex to HPV Genotypes 16 and 18/45 by TMA, ThinPrep	0051281	Norovirus Group 1 and 2 Detection by RT-PCR
2011933	Human Papillomavirus (HPV), High Risk with 16 and 18 Genotype by PCR, SurePath	2006247	Parainfluenza 1–4 by RT-PCR
2011940	Human Papillomavirus (HPV), High Risk with 16 and 18 Genotype by PCR, ThinPre	2005731	Parechovirus Detection by RT-PCR
2008788	Influenza A Virus H1/H3 Subtyping by PCR with Reflex to H1N1 (2009) Oseltamivir Resistance by Sequencing	0060043	Parvovirus B 19 by PCR
2007469	Influenza A Virus H1/H3 Subtyping by Real-Time RT-PCR	0060028	Parvovirus B 19 by PCR, Bone Marrow
0060764	Influenza B by PCR (Orderable Test: Respiratory Virus Mini Panel)	2002565	Respiratory Viruses DFA with Reflex to Respiratory Virus Mini Panel by RT-PCR
2002643	Influenza Virus A and B DFA with Reflex to Respiratory Virus Mini Panel by PCR	0060764	Respiratory Virus Mini Panel by RT-PCR
0099169	JC Virus by PCR	2007805	Respiratory Virus Panel by PCR
2009387	Mumps Virus RNA Qualitative, Real-Time PCR	0060042	Varicella-Zoster Virus by PCR
		2007063	Viral Meningitis Panel by PCR, Cerebrospinal Fluid
		2007062	Viral Meningoencephalitis Panel by PCR, Cerebrospinal Fluid
		0050229	West Nile Virus RNA by RT-PCR

## molecular infectious disease—viral testing: hepatitis and HIV

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2004722	Hepatitis B Virus DNA Quantitative, Real-Time PCR with Reflex to Genotype	0092050	Human Immunodeficiency Virus (HIV) Phenotype Comprehensive
0056025	Hepatitis B Virus DNA Quantitative, Real-Time PCR	0093061	Human Immunodeficiency Virus 1 (HIV-1) by Qualitative PCR
2001567	Hepatitis B Virus Genotype	2010797	Human Immunodeficiency Virus 1 (HIV-1) by Quantitative PCR with Reflex to HIV PhenoSense GT
0055593	Hepatitis C Virus Genotyping	2004457	Human Immunodeficiency Virus 1 Integrase Inhibitory Resistance by Sequencing
2010793	Hepatitis C Virus (HCV) by Quantitative PCR with Reflex to HCV High-Resolution Genotype by Sequencing	0055598	Human Immunodeficiency Virus 1 RNA Quantitative Real-Time PCR
2006898	Hepatitis C Virus High-Resolution Genotyping	2002689	Human Immunodeficiency Virus 1 RNA Quantitative Real-Time PCR with Reflex to Genotype
0098264	Hepatitis C Virus RNA Qualitative PCR	0055670	Human Immunodeficiency Virus 1, Genotyping
0098268	Hepatitis C Virus RNA Quantitative Real-Time PCR	2004680	Interleukin 28 B ( <i>IL28B</i> )-Associated Variants, 2 SNPs
2002685	Hepatitis C Virus RNA Quantitative Real-Time PCR with Reflex to Genotype	0093370	Trofile Co-Receptor Tropism
2009256	HIV1 Genotype and Integrase Inhibitor Resistance by Sequencing	2004747	Trofile DNA Co-Receptor Tropism Assay
0092399	HIV PhenoSense GT		
2004331	Human Immunodeficiency Virus GenoSure MG		



## molecular infectious disease—bacterial testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
0060738	Acid-Fast Bacillus (AFB) Culture and AFB Stain with Reflex to <i>Mycobacterium tuberculosis</i> Amplified Direct Detection	2002838	<i>Clostridium difficile</i> toxin B gene (tcdB) by PCR
2008462	Antimicrobial Susceptibility—Carbapenem Resistance Confirmation by PCR	0065153	Vaginal Pathogens DNA Direct Probes
0060211	Antimicrobial Susceptibility— <i>mecA</i> Gene by PCR	0056105	<i>Legionella</i> Species by PCR
0060182	Bacterial Strain Characterization by Pulsed-Field Gel Electrophoresis	0060999	<i>Mycobacterium avium intracellulare</i> by DNA Probe (Orderable Test: AFB Identification)
0093057	<i>Bartonella</i> Species by PCR	0060999	<i>Mycobacterium chelonae-abscessus</i> Identification by PCR (Orderable Test: AFB Identification)
0060762	<i>Bartonella</i> DNA Detection by PCR, Whole Blood	2010775	<i>Mycobacterium tuberculosis</i> Complex Detection and Rifampin Resistance by PCR
0065078	<i>Bordetella pertussis</i> by PCR	0060771	<i>Mycobacterium tuberculosis</i> Complex Speciation
0065080	<i>Bordetella pertussis/parapertussis</i> by PCR	0060999	<i>Mycobacterium tuberculosis</i> by DNA Probe (Orderable Test: AFB Identification)
0055570	<i>Borrelia</i> species DNA Detection by PCR (Lyme Disease)	0060256	<i>Mycoplasma pneumoniae</i> by PCR
0060715	<i>Chlamydia pneumoniae</i> by PCR	0060244	<i>Neisseria gonorrhoeae</i> by Transcription-Mediated Amplification (TMA)
0060774	<i>Chlamydia trachomatis</i> and <i>Neisseria gonorrhoeae</i> by Transcription-Mediated Amplification (TMA), M4/UTM	0060720	Organism Identification by 16S rDNA Sequencing
2001551	<i>Chlamydia trachomatis</i> and <i>Neisseria gonorrhoeae</i> by Transcription-Mediated Amplification (TMA), SurePath	0060705	<i>Streptococcus</i> Group B by PCR
0060734	<i>Chlamydia trachomatis</i> and <i>Neisseria gonorrhoeae</i> by Transcription-Mediated Amplification (TMA), ThinPrep	2011172	Urogenital <i>Ureaplasma</i> and <i>Mycoplasma</i> Species by PCR
0060243	<i>Chlamydia trachomatis</i> by Transcription-Mediated Amplification (TMA)	0065153	Vaginal Pathogens DNA Direct Probes

## molecular infectious disease—other testing

TEST #	TEST NAME/DESCRIPTION	TEST #	TEST NAME/DESCRIPTION
2008665	<i>Babesia</i> Species by PCR	2011626	Microsporidia by PCR
0062224	<i>Blastomyces dermatitidis</i> Identification by DNA Probe	2006258	Sexually Transmitted Disease Panel 1
0065153	<i>Candida</i> species DNA Probe (Orderable Test: Vaginal Pathogens DNA Direct Probes)	2006254	<i>Pneumocystis jirovecii</i> by PCR
0062225	<i>Coccidioides immitis</i> Identification by DNA Probe	2009226	<i>Pneumocystis jirovecii</i> DFA with Reflex to <i>Pneumocystis jirovecii</i> by PCR
2007862	<i>Ehrlichia</i> and <i>Anaplasma</i> Species by Real-Time PCR	0055591	<i>Toxoplasma gondii</i> by PCR
0060756	Fungal Identification by ITS rDNA Sequencing	2008670	Tick-Borne Disease Panel by PCR, Blood
2011660	Gastrointestinal Parasite and Microsporidia by PCR	2005506	<i>Trichomonas vaginalis</i> by Amplified Detection
0062226	<i>Histoplasma capsulatum</i> Identification by DNA Probe	0065153	<i>Trichomonas vaginalis</i> DNA Probe (Orderable Test: Vaginal Pathogens DNA Direct Probes)
2004963	Malaria Detection and Speciation, Qualitative by Real-Time PCR		

\* Multiple Tests Available ‡ Fetal Testing Available



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