

**Gene Sequence Analysis (unless otherwise indicated)**

Test Code	Test Name	CPT Codes
<b>Angelman/Prader Willi</b>		
<b>8520</b>	<b>Ambry SEQUENCE™: Angelman Syndrome</b> ( <i>SNRPN, UBE3A</i> )	83891, 83892, 83894x12, 83898x11, 83904x26, 83909x26, 83912x2
2400	Angelman Syndrome <i>UBE3A</i> -Related	83891, 83894x11, 83898x10, 83904x23, 83909x23, 83912
2420	Angelman Syndrome X-Linked ( <i>SLC9A6</i> )	83891, 83894x16, 83898x15, 83904x28, 83909x28, 83912
2440	Prader-Willi/Angelman Syndrome Methylation Analysis ( <i>SNRPN</i> )	83891, 83892, 83894x2, 83898, 83904x3, 83909x3, 83912
<b>Cancer</b>		
<b>8500</b>	<b>Ambry SEQUENCE™: HNPCC</b> Complete pathway, steps 1-3 ( <i>MLH1, MSH2, MSH6</i> )	83891, 83894x38, 83898x38, 83900 83901x33, 83904x48, 83909x49, 83912x4
8504	Step 1 only ( <i>MLH1, MSH2</i> )	83891, 83894x33, 83898x32, 83904x32, 83909x32, 83912x2
8506	Step 2 only ( <i>MLH1, MSH2 del/dup</i> )	83891, 83894, 83900, 83901x33, 83909, 83912
8502	HNPCC AMPLIFIED™ ( <i>MLH1, MSH2</i> )	83891, 83894x33, 83898x33, 83904x32, 83900x1, 83901x33, 83909x32, 83912x3
2200	HNPCC ( <i>MLH1</i> )	83891, 83894x17, 83898x16, 83904x16, 83909x16, 83912
2220	HNPCC ( <i>MSH2</i> )	83891, 83894x17, 83898x16, 83904x16, 83909x16, 83912
2240	HNPCC ( <i>MSH6</i> )	83891, 83894x17, 83898x16, 83904x16, 83909x16, 83912
2360	PALB2-Related Pancreatic Cancer	83891, 83898x15, 83904x30, 83909x30, 83912
2100	PTEN disorders ( <i>PTEN</i> )	83891, 83894x12, 83898x11, 83904x22, 83909x22, 83912
1685	Juvenile Polyposis ( <i>SMAD4</i> )	83891, 83894x11, 83898x10, 83904x15, 83909x15, 83912
2680	Multiple Endocrine Neoplasia Type 2 SEQUENCE™	83891, 83894x21, 83898x20, 83904x35, 83909x35, 83912
2684	Multiple Endocrine Neoplasia Type 2 Step 1 only (exons: 10,11, 13-16)	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
<b>Congenital Hyperinsulinism &amp; Diabetes</b>		
<b>8080</b>	<b>Ambry SEQUENCE™: Congenital Hyperinsulinism</b> If positive after 1st gene analyzed ( <i>ABCC8</i> ) If positive after both genes analyzed ( <i>ABCC8, KCNJ11</i> )	83891, 83894x35, 83898x35, 83904x35, 83909x35, 83912 83891, 83894x40, 83898x39, 83904x43, 83909x43, 83912x2
<b>8062</b>	<b>Ambry SEQUENCE™: Neonatal Diabetes</b> If positive after 1st gene analyzed ( <i>KCNJ11</i> ) If positive after 2nd gene analyzed ( <i>KCNJ11, INS</i> ) If positive after 3rd gene analyzed ( <i>KCNJ11, INS, ABCC8</i> )	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912 83891, 83894x8, 83898x6, 83904x12, 83909x12, 83912x2 83891, 83894x43, 83898x41, 83904x47, 83909x47, 83912x3
<b>MODY - Maturity Onset Diabetes of the Young</b>		
<b>9000</b>	<b>Ambry SEQUENCE™: MODY</b> If positive after 1st gene analyzed ( <i>TCF1</i> ) If positive after 2nd gene analyzed ( <i>TCF1, GCK</i> ) If positive after 3rd gene analyzed ( <i>TCF1, GCK, HNF4A</i> )	83891, 83894x10, 83898x9, 83904x20, 83909x20, 83912 83891, 83894x21, 83898x19, 83904x40, 83909x40, 83912x2 83891, 83894x32, 83898x29, 83904x60, 83909x60, 83912x3
<b>MODY - Neonatal Diabetes &amp; Congenital Hyperinsulinism</b>		
1380	( <i>ABCC8</i> )	83891, 83894x35, 83898x35, 83904x35, 83909x35, 83912
1370	Hyperinsulinism-Hyperammonemia ( <i>GLUD1</i> )	83891, 83894x13, 83898x12, 83904x24, 83909x24, 83912
1620	Insulin ( <i>INS</i> )	83891, 83894x3, 83898x2, 83904x4, 83909x4, 83912
1360	( <i>KCNJ11</i> )	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912
1480	MODY1 ( <i>HNF4A</i> )	83891, 83894x11, 83898x10, 83904x20, 83909x20, 83912
1340	MODY2/Neonatal Diabetes ( <i>GCK</i> )	83891, 83894x11, 83898x10, 83904x20, 83909x20, 83912
1420	MODY3 ( <i>TCF1</i> )	83891, 83894x10, 83898x9, 83904x20, 83909x20, 83912
1400	MODY4/Neonatal Diabetes ( <i>IPF1</i> )	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912
1500	MODY5 ( <i>TCF2</i> )	83891, 83894x11, 83898x10, 83904x20, 83909x20, 83912
8300	MODY 1-3 Deletion/Duplication ( <i>HNF4A, GCK, TCF1</i> )	83891, 83894, 83900, 83901x40, 83909, 83912
1510	RCAD/MODY5 AMPLIFIED™ ( <i>TCF2</i> )	83891, 83894x11, 83898x10, 83900, 83901x8, 83904x20, 83909x21, 83912
1510	RCAD/MODY5 Deletion/Duplication ( <i>TCF2</i> )	83891, 83894, 83900, 83901x8, 83909, 83912

**Gene Sequence Analysis (unless otherwise indicated)**

Test Code	Test Name	CPT Codes
<b>Chromosomal Microarray Analysis (aCGH)</b>		
6000	Ambry CMA: 105K Array Oligo	83891, 83892x2, 83894, 88386x4, 83912
<b>Cystic Fibrosis (CFTR)</b>		
1002	508 FIRST™ (CFTR)	83891, 83892, 83894x2, 83898, 83912
1006	CF AMPLIFIED™ (CFTR)	83891, 83894x2, 83898x77, 83900, 83901x27, 83903x29, 83904, 83909, 83912x2
1000	CF Gene Sequence Analysis (CFTR)	83891, 83894x2, 83898x82, 83903x29, 83904, 83909, 83912
1004	CF Deletion/Duplication (CFTR)	83891, 83894, 83900, 83901x27, 83909, 83912
1010	TG Repeat (CFTR)	83891, 83894, 83898, 83904x2, 83909x2, 83912
<b>Diamond-Blackfan Anemia</b>		
<b>8540</b>	<b>Ambry SEQUENCE™: Diamond-Blackfan Anemia</b> (Complete pathway, steps 1-3)	83891, 83894x38, 83898x37, 83904x74, 83909x74, 83912x7
8542	Step 1 only (RSP19)	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
8544	Step 2 only (RPL5, RPL11, RPL35A)	83891, 83894x17, 83898x16, 83904x32, 83909x32, 83912x3
8545	Step 1 and step 2	83891, 83894x23, 83898x22, 83904x44, 83909x44, 83912x4
8547	Step 2 reflex to Step 3	83891, 83894x32, 83898x31, 83904x62, 83909x62, 83912x6
2560	RPS19-Related DBA	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
2460	RPL5-Related DBA	83891, 83894x8, 83898x7, 83904x14, 83909x14, 83912
2480	RPL11-Related DBA	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
2500	RPL35A-Related DBA	83891, 83894x4, 83898x3, 83904x6, 83909x6, 83912
2580	RPS24-Related DBA	83891, 83894x6, 83898x5, 83904x10, 83909x10, 83912
2540	RPS17-Related DBA	83891, 83894x6, 83898x5, 83904x10, 83909x10, 83912
2520	RPS7-Related DBA	83891, 83894x6, 83898x5, 83904x10, 83909x10, 83912
<b>Dyskeratosis Congenita (DC)</b>		
<b>Ambry SEQUENCE™: Dyskeratosis Congenita</b>		
8160	Complete pathway, Steps 1-3 (DKC1, TINF2, TERC, NHP2, NOP10, TERT)	83891, 83894x39, 83898x38, 83904x75, 83909x76, 83912x3
8163	Steps 1 & 2 (DKC1, TINF2, TERC, NHP2, NOP10)	83891, 83894x20, 83898x19, 83904x38, 83909x38, 83912x2
8162	Step 1 Only: (DKC1, TINF2, TERC)	83891, 83894x18, 83898x17, 83904x 34, 83909x34, 83912
8164	Step 2 Only: (NHP2, NOP10)	83891, 83894x3, 83898x2, 83904x4, 83909x4, 83912
2140	Step 3 Only: (TERT)	83891, 83894x20, 83898x19, 83904x38, 83909x38, 83912
1960	DKC1- Related Dyskeratosis Congenita	83891, 83894x16, 83898x15, 83904x30, 83909x30, 83912
1980	TINF2- Related Dyskeratosis Congenita	83891, 83894x2, 83898, 83904x2, 83909x2, 83912
2120	TERC- Related Dyskeratosis Congenita	83891, 83894x2, 83898, 83904x2, 83909x2, 83912
<b>Gastroenterology</b>		
8020	Pancreatitis Panel (CFTR, PRSS1, SPINK1)	83891, 83894x2, 83898x94, 83903x44, 83904x2, 83909x2, 83912x3
8040	Pancreatitis AMPLIFIED™ (Panel + CFTR Del/Dup)	83891, 83894x2, 83898x86, 83900, 83901x27, 83903x41, 83904x3, 83909x3, 83912x4
1100	PRSS1	83891, 83894, 83898x17, 83903x7, 83904, 83909, 83912
1120	SPINK1	83891, 83894, 83898x14, 83903x8, 83904, 83909, 83912
1660	Chymotrypsin C-Related Pancreatitis (CTRC)	83891, 83894x8, 83898x7, 83904x14, 83909x14, 83912
1840	Wilson Disease (ATP7B)	83891, 83894x26, 83898x26, 83904x33, 83909x33, 83912
<b>Genetics</b>		
1640	Alagille AMPLIFIED™: (JAG1)	83891, 83894x26, 83898x25, 83900, 83901x24, 83904x48, 83909x48, 83912x2
1643	Alagille Gene Sequence Analysis (JAG1)	83891, 83894x26, 83898x25, 83904x48, 83909x48, 83912
1641	Alagille Deletion/Duplication (JAG1)	83891, 83894, 83900, 83901x24, 83909, 83912
1320	Aminoglycoside-Related Hearing Loss (MT-RNR1)	83891, 83894x4, 83898x3, 83904x6, 83909x6, 83912
1800	Ashkenazi Jewish FlexPanel	83891, 83894, 83898x30, 83904x30, 83909x30, 83912

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<b>Genetics</b>		
1040	Beta Thalassemia Plus ( <i>HBB</i> )	83891, 83894x3, 83898x2, 83904x8, 83909x8, 83912
1220	Canavan AMPLIFIED™ ( <i>ASPA</i> )	83891, 83894x7, 83898x6, 83900, 83901x4, 83904x12, 83909x13, 83912x2
2380	CHARGE Syndrome ( <i>CHD7</i> )	83891, 83894x39, 83898x38, 83904x60, 83909x45, 83912
1720	Fabry Disease ( <i>GLA</i> )	83891, 83894x9, 83898x8, 83904x16, 83909x16, 83912
2620	Familial Hypocalciuric Hypercalcemia ( <i>CASR</i> )	83891, 83894x10, 83898x9, 83904x18, 83909x18, 83912
1820	Gaucher Disease ( <i>GBA</i> )	83891, 83894x15, 83898x14, 83904x20, 83909x20, 83912
1600	Glutaric Acidemia Type 1 ( <i>GCDH</i> )	83891, 83894x9, 83898x8, 83904x16, 83909x16, 83912
1800	Glycogen Storage Disease Type Ia ( <i>G6PC</i> )	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
1900	Glycogen Storage Disease Type Ib ( <i>SLC37A4</i> )	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
2746	Hereditary Angioedema AMPLIFIED™ ( <i>SERPING1</i> )	83891, 83894x8, 83898x7, 83904x15, 83900, 83901x6, 83909x16, 83912x2
2740	Hereditary Angioedema ( <i>SERPING1</i> )	83891, 83894x8, 83898x7, 83904x15, 83909x15, 83912
2744	Hereditary Angioedema Deletion/Duplication ( <i>SERPING1</i> )	83891, 83894, 83900, 83901x6, 83909, 83912
1940	Hunter Syndrome ( <i>IDS</i> )	83891, 83894x10, 83898x9, 83904x20, 83909x20, 83912
2160	Hurler Syndrome ( <i>IDUA</i> )	83891, 83894x12, 83898x11, 83904x22, 83909x22, 83912
2040	Infantile Spasms ( <i>CDKL5</i> )	83891, 83894x23, 83898x22, 83904x43, 83909x43, 83912
1260	Maternal Cell Contamination	83891, 83894x8, 83898x7, 83904x14, 83909x14, 83912
2640	Multiple Endocrine Neoplasia Type1 Syndrome ( <i>MEN1</i> )	83891, 83894, 83898x7, 83909x2, 83912
1860	Niemann-Pick Disease Types A & B ( <i>SMPD1</i> )	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
1760	PKU ( <i>PAH</i> gene for Phenylketonuria)	83891, 83894x14, 83898x13, 83904x21, 83909x21, 83912
1740	Pompe Disease ( <i>GAA</i> )	83891, 83894x19, 83898x18, 83904x34, 83909x34, 83912
1508	Renal Cysts AMPLIFIED™ ( <i>TCF2</i> )	83891, 83894x11, 83898x10, 83900, 83901x8, 83904x20, 83909x21, 83912
1506	Renal Cysts Deletion/Duplication ( <i>TCF2</i> )	83891, 83894, 83900, 83901x8, 83909x1, 83912
1506	Renal Cysts Deletion/Duplication ( <i>TCF2</i> )	83891, 83894, 83900, 83901x8, 83909x1, 83912
2700	<i>RET</i> - related Hirschsprung SEQUENCE™	83891, 83894x21, 83898x20, 83904x35, 83909x35, 83912
2704	<i>RET</i> - related Hirschsprung SEQUENCE™ Step 1	83891, 83894x7, 83898x6, 83904x12, 83909x12, 83912
1440	Shwachman-Diamond Syndrome ( <i>SBDS</i> )	83891, 83894x5, 83898x4, 83904x10, 83909x10, 83912
2180	Smith-Lemli-Opitz Syndrome ( <i>DHCR7</i> )	83891, 83894x8, 83898x7, 83904x14, 83909x14, 83912
1240	Tay-Sachs Plus ( <i>HEXA</i> )	83891, 83894x16, 83898x15, 83904x28, 83909x28, 83912
1560	Transthyretin Amyloidosis ( <i>TTR</i> )	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912
2600	Von Hippel-Lindau Disease ( <i>VHL</i> )	83891, 83894x4, 83898x3, 83904x6, 83909x6, 83912
1700	Warfarin Sensitivity ( <i>CYP2C9</i> & <i>VKORC1</i> SNP Analysis)	83891, 83894x4, 83898x3, 83904x6, 83909x6, 83912
2040	X-Linked Retardation ( <i>CDKL5</i> )	83891, 83894x23, 83898x22, 83904x43, 83909x43, 83912
<b>Hereditary Hemorrhagic Telangiectasia (HHT)</b>		
1680	HHT AMPLIFIED™: ( <i>ACVRL1, ENG</i> )	83891, 83894x25, 83898x24, 83900, 83901x27, 83904x29, 83909x29, 83912x3
1683	HHT Gene Sequence Analysis ( <i>ACVRL1, ENG</i> )	83891, 83894x25, 83898x24, 83904x29, 83909x29, 82912x2
1681	HHT Deletion/Duplication: ( <i>ACVRL1, ENG</i> )	83891, 83894, 83900, 83901x27, 83909, 83912
1684	SMAD4-Related HHT ( <i>SMAD4</i> exons 8-11) (Reflex to exons 1-7)	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912 83891, 83894x11, 83898x10, 83904x15, 83904x15, 83909x15, 83912
1689	HHT ( <i>ENG</i> )	83891, 83894x16, 83898x15, 83904x20, 83909x20, 83912
1690	HHT ( <i>ACVRL1</i> )	83891, 83894x10, 83898x9, 83904x18, 83909x18, 83912

**Gene Sequence Analysis (unless otherwise indicated)**

Test Code	Test Name	CPT Codes
<b>Noonan / LEOPARD Syndrome</b>		
<b>8400</b>	<b>Ambry SEQUENCE: Noonan Syndrome</b> ( <i>PTPN11, SOS1, RAF1, KRAS</i> )	83891, 83894x48, 83898x47, 83904x94, 83909x94, 83912x4
2284	Step 1 only ( <i>PTPN11</i> )	83891, 83894x15, 83898x14, 83904x28, 83909x28, 83912
8420	Step 2 only ( <i>SOS1, partial RAF1, KRAS</i> )	83891, 83894x34, 83898x33, 83904x66, 83909x66, 83912x3
<b>8440</b>	<b>Ambry SEQUENCE: Noonan/LEOPARD Syndrome</b> <i>PTPN11, RAF1, reflex to SOS1, KRAS</i> )	83891, 83894x48, 83898x47, 83904x94, 83909x94, 83912x4
8460	Step 1 <i>PTPN11</i> and partial <i>RAF1</i>	83891, 83894x19, 83898x18, 83904x36, 83909x36, 83912x2
8430	Step 2 <i>SOS1</i> and <i>KRAS</i>	83891, 83894x30, 83898x29, 83904x58, 83909x58, 83912x2
2280	<i>PTPN11</i> -Related Noonan/LEOPARD Disorders	83891, 83894x15, 83898x14, 83904x28, 83909x28, 83912
2320	<i>RAF1</i> -Related Noonan/LEOPARD Disorders	83891, 83894x15, 83898x14, 83904x30, 83909x30, 83912
2300	<i>SOS1</i> -Related Noonan Disorders	83891, 83894x25, 83898x24, 83904x28, 83909x48, 83912
2340	<i>KRAS</i> -Related Noonan Disorders	83891, 83894x6, 83898x5, 83904x10, 83909x10, 83912
8460	LEOPARD Syndrome ( <i>PTPN11, RAF1</i> )	83891, 83894x19, 83898x18, 83904x36, 83909x36, 83912x2
<b>Pulmonology</b>		
1300	<i>ABCA3</i>	83891, 83894x31, 83898x30, 83904x40, 83909x40, 83912
1140	Alpha-1 Antitrypsin Deficiency ( <i>SERPINA1</i> )	83891, 83894x5, 83898x4, 83904x10, 83909x10, 83912
1580	Congenital Central Hypoventilation Syndrome ( <i>PHOX2B</i> )	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912
8140	IPF Telomerase ( <i>TERT, TERC</i> )	83891, 83894x21, 83898x20, 83904x40, 83909x40, 83912x2
8120	PCD 61 ( <i>DNAH5 &amp; DNAI1</i> Mutation Panel)	83891, 83894, 83898x20, 83904x20, 83909x20, 83912
1540	Pulmonary Arterial Hypertension ( <i>PAH AMPLIFIED™-BMPR2</i> )	83891, 83894x17, 83898x16, 83900, 83901x13, 83904x33, 83909x33, 83912x2
1541	PAH Deletion/Duplication ( <i>BMPR2</i> )	83891, 83894, 83900, 83901x13, 83909, 83912
1160	Surfactant Protein B ( <i>SFTPB</i> )	83891, 83894x11, 83898x10, 83904x20, 83909x20, 83912
1180	Surfactant Protein C ( <i>SFTPC</i> )	83891, 83894x6, 83898x5, 83904x11, 83909x11, 83912
<b>Rett Syndrome</b>		
8200	Ambry SEQUENCE™: Rett Syndrome ( <i>MECP2, CDKL5</i> )	83891, 83894x32, 83898x30, 83900, 83901x12, 83904x59, 83909x60, 83912x3
2026	<i>MECP2</i> AMPLIFIED™	83891, 83894x9, 83898x8, 83900, 83901x12, 83904x16, 83909x17, 83912x2
2020	<i>MECP2</i> Gene Sequence Analysis	83891, 83894x9, 83898x8, 83904x16, 83909x16, 83912
2022	<i>MECP2</i> Deletion/Duplication	83891, 83894, 83900, 83901x12, 83909, 83912
2040	<i>CDKL5</i> Gene Sequence (Atypical Rett Syndrome)	83891, 83894x23, 83898x22, 83904x43, 83909x43, 83912
<b>Specific Mutation Analysis</b>		
call	Specific mutation analysis (1 mutation)	83891, 83894x2, 83898, 83904x2, 83909x2, 83912
call	Specific mutation analysis (1 entire exon)	83891, 83894x2, 83898, 83904x2, 83909x2, 83912
call	Specific mutation analysis (2 mutation)	83891, 83894x4, 83898x2, 83904x4, 83909x4, 83912
call	Specific mutation analysis (3 mutation)	83891, 83894x4, 83898x3, 83904x6, 83909x6, 83912
call	Specific mutation analysis (4 mutation)	83891, 83894x5, 83898x4, 83904x8, 83909x8, 83912