Ambry Genetics®

Comprehensive Test Menu

Cancer

BRAIN TUMORS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Brain tumors, hereditary	BrainTumorNext®*: 29 genes	14-21 days	8847, 8847-R
Neurofibromatosis type 2 (NF2)	*NF2*	14-21 days	9024
Schwannomatosis	*SMARCB1*	14-21 days	7180

BREAST AND GYNECOLOGIC CANCER

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Ataxia-telangiectasia	*ATM*	14-21 days	9014
Breast cancer, hereditary	*BRCA1/BRCA2*	6-10 days	8838
	BRCA Ashkenazi Jewish 3-site mutation panel	6-10 days	5892
	BRCAplus®: 8 genes	7-10 days	8836
	BRCANext™*: 18 genes	14-21 days	8855, 8855-R
	BRCANext-*Expanded*™*: 23 genes	14-21 days	8860, 8860-R
CHEK2-related cancer	*CHEK2*	14-21 days	9016
Li-Fraumeni syndrome	*TP53*	14-21 days	2866
Ovarian, breast and uterine cancer, hereditary	BRCANext*: 18 genes	14-21 days	8855, 8855-R
	BRCANext-*Expanded**: 23 genes	14-21 days	8860, 8860-R
PALB2-associated cancer	*PALB2*	14-21 days	2366
PTEN-related disorders (Cowden syndrome, Proteus syndrome, macrocephaly and autism)	*PTEN*	14-21 days	2106

COMPREHENSIVE CANCER

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Breast, ovarian, colorectal, uterine, pancreatic, prostate, and other selected cancers	CancerNext®*: 36 genes	14-21 days	8824, 8824-R
	CancerNext-*Expanded*®*: 77 genes	14-21 days	8874, 8874-R
	CustomNext-*Cancer*®*: choose up to 91 genes	14-21 days	9510, 9510-R

ENDOCRINE TUMORS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Multiple endocrine neoplasia type 1 (MEN1)	*MEN1*	14-21 days	2646
Multiple endocrine neoplasia type 2 (MEN2) and familial medullary thyroid cancer (FMTC)	*RET*	14-21 days	2680
Paraganglioma (PGL) and pheochromocytoma (PCC), hereditary	PGLNext®*: 14 genes	14-21 days	5504, 5504-R

GASTROINTESTINAL CANCER

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Adenomatous polyposis	*APC, MUTYH*	14-21 days	8726
Colorectal cancer, hereditary	ColoNext®*: 20 genes	14-21 days	8822, 8822-R
Familial adenomatous polyposis (FAP)	*APC*	14-21 days	3040
Gastric cancer, hereditary diffuse (HDGC)	*CDH1*	10-21 days	4726
Juvenile polyposis syndrome (JPS)	*BMPR1A, SMAD4*	14-21 days	8604
Lynch syndrome (formerly hereditary non-polyposis colorectal cancer or HNPCC) - germline only	*MLH1, MSH2, MSH6, PMS2, EPCAM*	14-21 days	8517
MUTYH-associated polyposis (MAP)	*MUTYH*	14-21 days	4661
Pancreatic cancer, hereditary	PancNext®*: 13 genes	14-21 days	8042, 8042-R
Peutz-Jeghers syndrome (PJS)	*STK11*	14-21 days	2766

GENITOURINARY CANCER

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Birt-Hogg-Dubé syndrome (BHDS)	*FLCN*	14-21 days	5921
Hereditary leiomyomatosis and renal cell carcinoma (HLRCC)	*FH*	14-21 days	6301
Kidney cancer, hereditary	*RenalNext®*: 20 genes*	14-21 days	5900, 5900-R
Prostate cancer, hereditary	*ProstateNext®*: 14 genes*	14-21 days	8845, 8845-R
von Hippel-Lindau disease (VHL)	*VHL*	14-21 days	2606

SKIN CANCER/MELANOMA

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Malignant melanoma, hereditary	*CDKN2A, CDK4*	14-21 days	4708
	MelanomaNext®*: 9 genes	14-21 days	8849, 8849-R
Nevoid basal cell carcinoma syndrome (NBCCS)/ Gorlin syndrome	*PTCH1*	14-21 days	5684

OTHER INDIVIDUAL HEREDITARY CANCER CONDITIONS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Li-Fraumeni syndrome	*TP53*	14-21 days	2866
Neurofibromatosis type 1 (NF1)	*NF1*	14-21 days	5704
Pleuropulmonary blastoma and *DICER1*- related disorders	*DICER1*	14-21 days	5260
PTEN-related disorders (Cowden syndrome, Proteus syndrome, macrocephaly and autism)	*PTEN*	14-21 days	2106
Retinoblastoma, hereditary	*RB1*	14-21 days	5426
Tuberous sclerosis complex (TSC)	*TSC1, TSC2*	14-21 days	5904

* Tests eligible for +RNAinisight(R)
+RNAinsight: Paired DNA/RNA analysis for up to 91 genes. Learn more at rnainsight.com

TESTS SUPPLEMENTED WITH +RNAINSIGHT®

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Paraganglioma (PGL) and pheochromocytoma (PCC), hereditary	PGLNext®: gene sequence and deletion/duplication +RNAinsight®	14-21 days	5504-R
Kidney cancer, hereditary	RenalNext®: gene sequence and deletion/duplication +RNAinsight®	14-21 days	5900-R
Pancreatic cancer, hereditary	*PancNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8042-R
Pancreatitis	*PancNext® plus Pancreatitis +RNAinsight®*	14-21 days	8064-R
Colorectal cancer, hereditary	*ColoNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8822-R
Breast, ovarian, colorectal, uterine, pancreatic, prostate, and other cancer	*CancerNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8824-R
Prostate cancer, hereditary	*ProstateNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8845-R
Brain tumors, hereditary	*BrainTumorNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8847-R
Malignant melanoma, hereditary	*MelanomaNext®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8849-R
Breast cancer, hereditary	*BRCANext™: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8855-R
Breast cancer, hereditary	*BRCANext-Expanded™: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8860-R
Breast, ovarian, colorectal, uterine, pancreatic, prostate, and other cancer	*CancerNext-Expanded®: gene sequence and deletion/duplication +RNAinsight®*	14-21 days	8874-R
Breast, ovarian, colorectal, uterine, pancreatic, prostate, and other cancer	*CustomNext: Cancer® +RNAinsight®*	14-21 days	9510-R
Breast, ovarian, colorectal, uterine, pancreatic, prostate, and other cancer	*Add-on testing CustomNext: Cancer® +RNAinsight®*	14-21 days	9510-R-A

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Cardiology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Arrhythmias and cardiomyopathy, inherited	CardioNext®: 92 genes	2-3 weeks	8911
Arrhythmias, inherited (long QT syndrome, Brugada syndrome, and others)	LongQTNext™: 17 genes	2-3 weeks	8890
	RhythmNext®: 42 genes	2-3 weeks	8900
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	ARVCNext™: 11 genes	2-3 weeks	8904
Cardiomyopathy, inherited	CMNext®: 56 genes	2-3 weeks	8887
Catecholaminergic polymorphic ventricular tachycardia (CPVT)	CPVTNext®: 4 genes	2-3 weeks	8902
Dilated cardiomyopathy (DCM)	DCMNext®: 37 genes	2-3 weeks	8884
Hereditary transthyretin amyloidosis (hATTR)	*TTR*	2-3 weeks	1560
Familial hypercholesterolemia (FH), Familial chylomicronemia syndrome (FCS), and Sitosterolemia	FHNext®: *APOB, LDLR, LDLRAP1, PCSK9* and *SLC01B1* (c.521T>C)	2-3 weeks	8680
	FCSNext: 5 genes	2-3 weeks	8920
	Sitosterolemia: *ABCG5, ABCG8*	2-3 weeks	8930
Custom cardiology panel	CustomNext-*Cardio*®: Choose up to 167 genes	2-3 weeks	9520

CONNECTIVE TISSUE RELATED DISORDERS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Noonan syndrome and other RASopathies	NoonanNext™: 18 genes	2-3 weeks	8402
Marfan syndrome, thoracic aortic aneurysms/ dissections and related disorders	*FBN1* reflex TAADNext®	2-3 weeks	8783
	TAADNext: 35 genes	2-3 weeks	8789

Clinical Genomics

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Chromosomal microarray analysis	SNP Array	2-3 weeks	5490
	Familial Targeted Microarray	2-3 weeks	5495
Exome sequencing	ExomeNext®-*Proband*	6-8 weeks	9993
	ExomeNext-*Proband* plus mtDNA	6-8 weeks	9994
	ExomeNext-*Duo*	6-8 weeks	9991
	ExomeNext-*Duo* plus mtDNA	6-8 weeks	9992
	ExomeNext-*Trio*	6-8 weeks	9995
	ExomeNext-*Trio* plus mtDNA	6-8 weeks	9996
	ExomeNext-*Rapid*®	14 days (preliminary) 18 days (full report)	9999R
	ExomeNext-*Select:* Choose up to 500 genes	2-4 weeks	9500
	Exome sequencing only - Raw data	4-6 weeks	9997
	Exome sequencing only - Raw data + Filtered Variant List	4-6 weeks	9998

Endocrinology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Hereditary leiomyomatosis and renal cell carcinoma	*FH*	14-21 days	6301
Maturity-onset diabetes of the young (MODY)	*HNF1A, HNF4A, HNF1B, GCK, PDX1*	4-5 weeks	8310
Multiple endocrine neoplasia type I (MEN1)	*MEN1*	14-21 days	2646
Multiple endocrine neoplasia type 2 (MEN2) and familial medullary thyroid cancer (FMTC)	*RET*	14-21 days	2680
Neurofibromatosis type 1 (NF1)	*NF1*	14-21 days	5704
Paraganglioma (PGL) and pheochromocytoma (PCC), hereditary	PGLNext*: 14 genes	14-21 days	5504, 5504-R
von Hippel-Lindau disease (VHL)	*VHL*	14-21 days	2606

Gastroenterology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Cystic fibrosis	508 FIRST®: deltaF508 mutation, reflex to *CFTR* Gene Sequence Analysis and Deletion/ Duplicaton Analysis	5-13 days	1002
	CFTR Gene Sequence Analysis and Deletion/ Duplication Analysis (concurrent)	5-13 days	1007
Hirschsprung disease (*RET*-related)	*RET*	14-21 days	2680
Juvenile polyposis syndrome (JPS)	*BMPR1A, SMAD4*	14-21 days	8604
Pancreatitis	*CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*	14-28 days	8022
	PancNext* plus Pancreatitis: 19 genes	14-21 days	8064, 8064-R
Peutz-Jeghers syndrome	*STK11*	14-21 days	2766

Hematology/Oncology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Diamond-Blackfan anemia	DBANext: 11 genes	14-28 days	8550
Dyskeratosis congenita	DCNext: 7 genes	14-28 days	8161
Shwachman-Diamond syndrome (SDS)	*SBDS*	14-28 days	1440

Multiple Congenital Anomalies

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Noonan syndrome	NoonanNext: 18 genes	2-3 weeks	8402

Neurology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Comprehensive neurology testing: intellectual disability, epilepsy, and autism spectrum disorders	CustomNext-*Neuro*™: Choose up to 500 genes from the neurology menu	2-4 weeks	9545

EPILEPSY

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Comprehensive epilepsy testing	EpilepsyNext®: 124 genes	2-4 weeks	6864
	EpilepsyNext-*Expanded*™: >890 genes	6-8 weeks	6865
Rapid epilepsy testing	EpiRapid®: 22 genes	10-14 days	6862
Familial hemiplegic migraine	FHM: 7 genes	2-4 weeks	6866

NEUROCUTANEOUS/NEURO-ONCOLOGY DISORDERS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Ataxia-telangiectasia	*ATM*	2-3 weeks	9014
Brain tumors, hereditary	BrainTumorNext*: 29 genes	2-3 weeks	8847, 8847-R
Hereditary hemorrhagic telangiectasia (HHT)	HHTNext®: 6 genes	2-3 weeks	8672
Legius syndrome	*SPRED1*	2-3 weeks	5724
Li-Fraumeni syndrome	*TP53*	2-3 weeks	2866
Neurofibromatosis 1 (NF1)	*NF1*	2-3 weeks	5704
Neurofibromatosis 2 (NF2)	*NF2*	2-3 weeks	9024
Nevoid basal cell carcinoma syndrome (NBCCS)/Gorlin syndrome	*PTCH1*	2-3 weeks	5684
Schwannamatosis	*SMARCB1*	2-3 weeks	7180
Tuberous sclerosis complex (TSC)	*TSC1 , TSC2*	2-3 weeks	5904
von Hippel-Lindau disease (VHL)	*VHL*	2-3 weeks	2606

Note: The gene list for EpilepsyNext-*Expanded* is updated annually due to proactive review of current literature using an internal, peer-reviewed clinical validity scheme (Smith ED, Radtke K, Rossi M, *et al.* 2017 Human mutation 38(5):600-608). The patient's test report will include a list of genes evaluated. For up-to-date gene lists, visit ambrygen.com

NEURODEVELOPMENTAL DISORDERS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Autism spectrum disorders and/or intellectual disability (non-syndromic)	AutismNext®: 72 genes	2-4 weeks	6863
	PTEN	2-3 weeks	2106
Developmental delay, intellectual disability, and/or autism spectrum disorders	NeurodevelopmentNext: 202 genes	2-4 weeks	6861
Fragile X syndrome (trinucleotide repeat analysis)	*FMR1*	1-2 weeks	4544

Prenatal Testing*

Test Name	Test Code
MCC for amniotic fluid culture or CVS	1260
Cultured amniocytes or CVS samples	1262

Pulmonology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Congenital central hypoventilation syndrome (CCHS)	*PHOX2B*	2-4 weeks	1580
Cystic fibrosis	508 FIRST: deltaF508 mutation, reflex to *CFTR* Gene Sequence Analysis and Deletion/ Duplicaton Analysis	5-13 days	1002
	CFTR Gene Sequence Analysis and Deletion/ Duplication Analysis (concurrent)	5-13 days	1007
Primary ciliary dyskinesia (PCD)	PCDNext®: 21 genes	4-5 weeks	8122
Surfactant dysfunction (respiratory distress syndrome)	*ABCA3, SFTPB, SFTPC*	5-14 days	8100
Telomere-related pulmonary fibrosis	*TERT, TERC*	2-4 weeks	8140

Reproductive Health

CARRIER SCREENING

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Cystic Fibrosis and Spinal Muscular Atrophy	CF + SMA: *CFTR, SMN1*	10-14 days	9082
Ashkenazi Jewish related disorders	Ashkenazi Jewish: 48 gene panel including *CFTR* and *SMN1*	10-14 days	9083
FMR1 related disorders	Fragile X: *FMR1* repeat analysis	10-14 days	9084-A
Pan-ethnic carrier screening based on ACMG/ACOG guidance	Guidelines-Based panel: 163 genes	10-14 days	9085-A
Pan-ethnic carrier screening based on ACMG/ACOG guidance and more rare, severe disorders	Comprehensive panel: 418 genes	10-14 days	9086

* Note: Prenatal testing is available on a case-by-case basis upon staff review.

NON-INVASIVE PRENATAL TESTING

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Common chromosomal aneuploidies	Chromosomes 21, 18 and 13	5-7 days	
	Singleton Pregnancies		9080
	Twin Pregnancies		9081
Sex Chromosomes aneuploidies & fetal sex	Sex Chromosomes (X and Y)	5-7 days	Add-on
Autosomal aneuploidy, sex chromosome aneuploidy & fetal sex (singleton pregnancies only)	All Chromosomes	5-7 days	Add-on
Microdeletion syndrome (singleton pregnancies only)	- DiGeorge syndrome (22q11.2) - 1p36 Deletion syndrome - Prader-Willi/Angelman Syndrome (15q11.2) - Cri du Chat (5p15.2) - Wolf-Hirschhorn (4p16.3)	*5-7 days*	Add-on

Vascular

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Hereditary hemorrhagic telangiectasia (HHT)	HHTNext: 6 genes	2-3 weeks	8672
Marfan syndrome, thoracic aortic aneurysms/ dissections and related disorders	*FBN1* reflex TAADNext	2-3 weeks	8783
	TAADNext: 35 genes	2-3 weeks	8689

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