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Comprehensive Test Menu | December 2018

Cancer

BRAIN TUMORS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Brain tumors, hereditary	BrainTumorNext: 27 genes	14-21 days	8847
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
	BreastNext: 17 genes	14-21 days	8820
CHEK2-related cancer	*CHEK2*	14-21 days	9016
Li-Fraumeni syndrome	*TP53*	14-21 days	2866
Ovarian and uterine cancer, hereditary	GYNplus: 13 genes	14-21 days	8835
Ovarian, breast and uterine cancer, hereditary	OvaNext: 25 genes	14-21 days	8830
Ovarian cancer, paired tumor/germline	TumorNext-*HRD*	21-28 days	9811
	TumorNext-*BRCA*	21-28 days	9810
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 and other cancer	CancerNext: 34 genes	14-21 days	8824
	CancerNext-*Expanded*: 67 genes	14-21 days	8874
	CustomNext-*Cancer*: choose up to 81 genes	14-21 days	9510

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: 12 genes	14-21 days	5504

Additional testing options like Gene Sequence Analysis, Deletion/Duplication Analysis, or Single Site Analysis may be available. Various reflex options between panels may also be available. Please see ambrygen.com for a complete list of genes and other details.

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GASTROINTESTINAL CANCER

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Adenomatous polyposis	*APC, MUTYH*	14-21 days	8726
Colorectal cancer, hereditary	ColoNext: 17 genes	14-21 days	8822
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
Lynch syndrome, paired tumor/germline	TumorNext-*Lynch*	21-28 days	8980
MUTYH-associated polyposis (MAP)	*MUTYH*	14-21 days	4661
Pancreatic cancer, hereditary	PancNext: 13 genes	14-21 days	8042
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: 18 genes	14-21 days	5900
Prostate cancer, hereditary	ProstateNext: 14 genes	14-21 days	8845
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: 8 genes	14-21 days	8849
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*	10-21 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

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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
	CustomNext-*Cardio*: Choose up to 167 genes	2-3 weeks	9520
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: 7 genes	2-3 weeks	8884
Familial transthyretin amyloidosis	*TTR*	2-3 weeks	1560
	HCMNext: 30 genes	2-3 weeks	8936
	HCMNext reflex	2-3 weeks	8883
Familial hypercholesterolemia (FH) and Other Lipid Disorders	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

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
	FBN1 reflex TAADNext	2-3 weeks	8783
Marfan syndrome, thoracic aortic aneurysms/ dissections and related disorders	TAADNext: 35 genes	2-3 weeks	8789

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Clinical Genomics

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Karyotype	High resolution	1-3 weeks	3660
	High resolution, rule out mosaic	1-3 weeks	3662
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-Trio	6-8 weeks	9995
	ExomeNext-Trio plus mtDNA	6-8 weeks	9996
	ExomeNext-*Select*: Choose up to 500 genes	6-8 weeks	9500
	ExomeNext-*Rapid*	8 days (verbal) 14 days (full report)	9999R
	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: 12 genes	14-21 days	5504
von Hippel-Lindau disease (VHL)	*VHL*	14-21 days	2606

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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
Peutz-Jeghers syndrome	*STK11*	10-21 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
CHARGE syndrome	*CHD7*	2-4 weeks	2380
Cornelia de Lange syndrome	CdLSNext: *NIPBL, SMC1A, HDAC8, RAD21, SMC3*	2-3 weeks	7040
Noonan syndrome	NoonanNext: 18 genes	2-3 weeks	8402

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Neurology

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Comprehensive neurology testing	CustomNext-*Neuro*: Choose up to 196 genes	2-3 weeks*	9540

**Orders with >140 genes will have a TAT of 4-6 weeks*

EPILEPSY

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Comprehensive epilepsy testing	EpilepsyNext: 100 genes	2-3 weeks	7019
Rapid epilepsy testing	EpiRapid: 16 genes	10-14 days	7033
	EpiRapid reflex EpilepsyNext	2-3 weeks	7034
Familial Hemiplegic Migraine	FHM: 4 genes	2-3 weeks	7035
Febrile seizures	EpiFirst-*Fever*: 13 genes	2-3 weeks	7011
Infantile spasms	EpiFirst-*IS*: 17 genes	2-3 weeks	7013
	Tuberous sclerosis complex: *TSC1, TSC2*	2-3 weeks	5904
Non-lesional focal epilepsy	EpiFirst-*Focal*: 11 genes	2-3 weeks	7017

HEREDITARY NEUROPATHY

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Familial transthyretin amyloidosis	*TTR*	2-3 weeks	1560

NEUROCUTANEOUS/NEURO-ONCOLOGY DISORDERS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Ataxia-telangiectasia	*ATM*	2-3 weeks	9014
Brain tumors, hereditary	BrainTumorNext: 27 genes	2-3 weeks	8847
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

Additional testing options like Gene Sequence Analysis, Deletion/Duplication Analysis, or Single Site Analysis may be available. Various reflex options between panels may also be available. Please see ambrygen.com for a complete list of genes and other details.

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NEURODEVELOPMENTAL DISORDERS

Disease/Condition	Test Name/Gene(s)	TAT	Test Code
Autism spectrum disorders	AutismNext: 48 genes	2-3 weeks	7024
	PTEN	1-3 weeks	2106
Autism spectrum disorders, epilepsy, intellectual disability	Neurodevelopment-*Expanded:* 196 genes	4-6 weeks	7028
Fragile X syndrome (Trinucleotide repeat analysis)	*FMR1*	1-2 weeks	4544
Intellectual disability	IDNext: 140 genes	2-3 weeks	7027
Rett syndrome	*MECP2*	2-3 weeks	2026

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

Vascular

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

Other

Test Name/Gene(s)	Test Code
MCC for amniotic fluid culture or CVS	1260
MCC reference for maternal blood sample	1262

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