

COLLECTION DATE (REQUIRED)

If date of collection is not provided, three calendar days before specimen receipt will be used (for specimens stored longer than 30 days, the day of archive retrieval will be used as the date of service)

PATIENT INFORMATION

Legal Name (Last, First, MI)		Date of Birth (Month/DD/YYYY)	Sex Assigned at Birth ☐ F ☐ M	Gender (optional) ☐ Man ☐ Woman ☐ Nonbinary ☐ Self-described
Genetic Ancestry: ☐ Ashkenazi Jewish ☐ Asian ☐ Black/African American ☐ French Canadian/Cajun ☐ Hispanic/Latino ☐ Mediterranean ☐ Middle Eastern ☐ Native American ☐ Pacific Islander ☐ Portuguese ☐ White ☐ Unknown ☐ Other:				MRN/National ID
Address			State/Province	Country
Mobile #		Email		

SPECIMEN INFORMATION* (Please see ambrygen.com/specimen-requirements for details)

☐ Personal history of allogenic bone marrow or peripheral stem cell transplant	
Specimen ID	Specimen Type

* Fetal specimens, cord blood, buccal samples, and POC samples are not accepted from International clients.

INDICATION(S) FOR TESTING

ICD-10 code(s):

ORDERING LICENSED PROVIDER/SENDING FACILITY (Each listed person will receive a copy of the report)

Facility Name (Facility Code)		Address	
State /Province		Country	Phone
Ordering Licensed Provider Name (Last, First)(Code)	NPI# (US only)	Phone	Fax/Email

ADDITIONAL RESULTS RECIPIENTS

Genetic Counselor or Other Medical Provider Name (Last, First) (Code)	Phone/Fax/Email
Genetic Counselor or Other Medical Provider Name (Last, First) (Code)	Phone/Fax/Email

CONFIRMATION OF INFORMED CONSENT AND MEDICAL NECESSITY FOR GENETIC TESTING

I confirm that the genetic test ordered is medically appropriate. All information on this TRF is true to the best of my knowledge. I also confirm that the patient has consented to proceed with genetic testing, including the transfer and processing of their sample and personal/sensitive information in the United States. I agree to allow Ambry Genetics to facilitate the provision of pre-test genetic counseling services by a third-party service, as required by the patient's insurance provider.

Signature Required for Processing Medical Professional Signature:	Date:
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BILLING

☐ INSURANCE BILLING (Include copy of both sides of insurance card)	
☐ INSTITUTIONAL BILLING	
Facility Name	☐ Send invoice to facility address above
Address	
Contact Name	
Phone Number	Email/Fax

☐ PATIENT PAYMENT	☐ Check (Payable to Ambry Genetics)	☐ Credit Card (Call 949-900-5795)
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Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. For direct insurance billing: I authorize my insurance benefits to be paid directly to Ambry Genetics Corporation (Ambry), authorize Ambry to release medical information concerning my testing to my insurer, to be my designated representative for purposes of appealing any denial of benefits as needed and to request additional medical records for this purpose. I understand that I am financially responsible for any amounts not covered by my insurer and responsible for sending Ambry money received from my health insurance company.

☐ I agree to be contacted regarding future research studies for which I may be a candidate. Any future research projects will be subject to a separate informed consent process and participation is voluntary. Learn more about Ambry's privacy practices at <https://www.ambrygen.com/legal/notice-of-privacy-practices>.

For patient payment by credit card: I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above.

Patient Signature (I agree to terms above):	Date:
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Patient Consent To Testing
 I acknowledge and agree that my health care provider has ordered genetic testing for processing at Ambry Genetics, a laboratory based in the United States and subject to U.S. based privacy laws, including the Health Insurance Portability and Accountability Act (HIPAA). I have provided my health care provider with the appropriate consent acknowledgment and documentation as required under the applicable laws in my country of residence for my genetic information to be sent to and processed in the United States. I have reviewed Ambry's Notice of Privacy Practices and understand and agree to the use of my health information in accordance with such privacy practices. I understand that I can exercise my rights to privacy consistent with HIPAA and Ambry's privacy notices.

Patient Signature (I agree to terms above):	Date:
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PLEASE SUBMIT THE FOLLOWING WITH THE TRF:

1. Clinic Notes 2. Pedigree 3. Insurance Card and Authorization Documents

CLINICAL HISTORY
PLEASE ATTACH PEDIGREE /CLINICAL CONSULTATION NOTES, IF AVAILABLE

Birth and Neonatal History ☐ Not Applicable	Developmental History ☐ Not Applicable
Gestational age at birth: _____ Birth weight: _____ Head circumference at birth (if available): _____ ☐ Congenital anomalies, explain: _____ ☐ Positive newborn screen, explain: _____	Developmental delay: ☐ yes ☐ no ☐ unknown Delay prior to seizure onset: ☐ yes ☐ no ☐ unknown ☐ N/A Type of delay (choose all that apply): ☐ motor ☐ language ☐ global Intellectual disability: ☐ yes ☐ no ☐ unknown Regression or plateau: ☐ yes ☐ no ☐ unknown Does patient meet DSM-V diagnostic criteria for an autism spectrum disorder?: ☐ yes ☐ no ☐ unknown
Seizure History ☐ Not Applicable	Cardiac History ☐ Not Applicable
Age at first unprovoked seizure (first seizure without fever or other acute metabolic or structural cause): _____ Seizure types (choose all that apply): ☐ Infantile/epileptic spasms ☐ Myoclonic ☐ Generalized tonic clonic ☐ Tonic ☐ Typical absence ☐ Focal seizures ☐ Atonic ☐ Atypical absence Are seizures: ☐ refractory ☐ well-controlled Has this patient been diagnosed with an epilepsy syndrome? ☐ yes ☐ no ☐ unknown If yes, please specify: _____	Sudden cardiac arrest ☐ Y ☐ N (if yes): # Episodes: _____ Age first incident: _____ Syncope ☐ Y ☐ N If yes, # Episodes: _____ Age first incident: _____ History of cardiomyopathy ☐ Y ☐ N Age at dx: _____ Cardiomyopathy type: _____ History of Arrhythmia ☐ Y ☐ N Age at dx: _____ Arrhythmia type: _____ ☐ Congenital heart defect _____
Pulmonology History ☐ Not Applicable	Other History ☐ Not Applicable
☐ Positive newborn screen ☐ CBAVD ☐ Meconium ileus ☐ Infections: _____ ☐ Sweat chloride: _____mmol/L ☐ Sweat chloride: ☐ <40 ☐ 40-60 ☐ >60 ☐ Pancreatic insufficiency IRT level: _____ ☐ Respiratory distress, explain: _____ ☐ Respiratory assistance devices: _____ ☐ Ultrasound findings: _____	☐ Hearing problems: _____ ☐ Vision problems: _____ ☐ Migraine: _____ ☐ Psychiatric: _____ ☐ Hematological: _____ ☐ Suspected genetic condition: _____ ☐ Other clinical findings: _____

Cancer History ☐ Not Applicable Metastatic: ☐ Yes ☐ No Tumor is ☐ MSI-High or ☐ IHC-Abnormal

Cancer/Tumor	Age at Dx	Pathology and Other Info
Brain		
Breast		Type: ER ☐ (+) ☐ (-) ☐ unk PR ☐ (+) ☐ (-) ☐ unk HER2/neu ☐ (+) ☐ (-) ☐ unk
2nd primary breast		Type: ER ☐ (+) ☐ (-) ☐ unk PR ☐ (+) ☐ (-) ☐ unk HER2/neu ☐ (+) ☐ (-) ☐ unk
Colorectal		Location: _____
Ovarian		☐ Fallopian tube ☐ Primary peritoneal
Melanoma/Skin		
Prostate		Gleason Score: _____
Uterine		
Hematologic*		Type: ☐ Allogenic bone marrow or peripheral stem cell transplant*
Other Cancer		Type: _____
GI polyps		☐ Adenomatous Polyp #: ☐ 1 ☐ 2-5 ☐ 6-9 ☐ 10-19 ☐ 20-99 ☐ 100+ ☐ Other type: _____ Polyp #: ☐ 1 ☐ 2-5 ☐ 6-9 ☐ 10-19 ☐ 20-99 ☐ 100+

*Blood or saliva from patients with active/recent hematological disease will undergo additional review and may not be accepted in some cases. For these, cultured fibroblasts or fresh/fresh frozen normal tissue are preferred. See ambrygen.com/specimen-requirements for details.

PREVIOUS TEST HISTORY (Please include copy of test results if performed at another laboratory) ☐ Limited family history

 Known Familial Variant: ☐ Family ☐ Self Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____
 Patient previously tested at Ambry? ☐ Yes ☐ No Family previously tested at Ambry? ☐ Yes ☐ No
 Name: _____ DOB: _____ Relation: _____

FAMILY MEMBER INFORMATION (Completion of this section is required for orders including parental samples)

 Mother - Name: _____ DOB: _____ ☐ unaffected ☐ affected, list symptoms/dx: _____ Dx age: _____
 Father - Name: _____ DOB: _____ ☐ unaffected ☐ affected, list symptoms/dx: _____ Dx age: _____

Relationship to Patient	Mat	Pat	Age at Dx	Family Testing and Cancer Type Details	Reason relative has not been tested
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact

Comprehensive Test Requisition Form - Page 3 of 7

If this TRF is sent to Ambry without or ahead of the sample, it will be treated as a preverification. If test ordered is different than the test preverified, we will honor what is on the TRF order form with the sample. Preverification will only be performed for ExomeNext or SNP Array testing.

Concurrent Testing: There is no action needed on your part if this is your desired strategy.

☐ **Reflex Testing:** Please select this option if you wish to have testing performed in a reflex manner, and indicate the order of testing below:

Test 1: _____ Test 2: _____

See Reflex or Concurrent Testing section of the Supplemental Information page for more information.

CANCER TEST ORDERS
Primary Test Order

! REQUIRED: Select a Primary Test Order

For Patients Meeting <i>BRCA1/2</i> Testing Criteria	For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (polyposis)
☐ <i>BRCA1/2</i> test	Polyposis test: ☐ <i>APC/MUTYH</i>
For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (Lynch)	☐ Other: _____
Lynch Syndrome test: ☐ <i>MLH1, MSH2, MSH6, PMS2, EPCAM</i>	☐ None of the above (patient does not meet any genetic testing criteria)

Select an Optional Supplemental Test (Per payer policy, all tests in this section will be processed and billed separately; tests may be performed as a reflex.)

Order	Test Code	Test Name	Description	Order	Test Code	Test Name	Description
☐	8857	BRCANext®	19 gene breast & gynecologic cancer test	☐	8821	ColoNext®	21 gene colorectal cancer & polyposis test
		Add on: ☐ Limited Evidence (Additional 7 genes)				Add on: ☐ Limited Evidence (Additional 5 genes)	
☐	8836	BRCAPlus®	13 gene STAT breast management test				
☐	8824	CancerNext®	40 gene pan-cancer test				
☐	8875	CancerNext-Expanded®	77 gene pan-cancer test	☐	9511	CustomNext-Cancer®	up to 90 gene custom test
		Add on: ☐ Limited Evidence (Additional 8 genes)				Notes: _____	Gene content is required. Use CustomNext-Cancer supplemental form for guidance.
		Add on: ☐ Pancreatitis (Additional 5 genes)				_____	

Other Supplemental Test Options (Select if applicable)

☐ +RNAinsight® (Not available with BRCAPlus, or STAT orders; PAXgene® tube required for RNA)

Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
Breast and/or Ovarian Cancer				Gastrointestinal Cancer (Cont.)			
☐	<i>ATM</i>	9014	Ataxia-telangiectasia	☐	<i>MLH1</i>	8508	Lynch syndrome
☐	<i>BRCA1/2</i>	8838	Hereditary breast and ovarian cancer	☐	<i>MSH2 + EPCAM</i> del/dup	8510	Includes <i>MSH2</i> inversion
☐	<i>BRCA1/2</i> Ashkenazi Jewish 3-site mutation panel	5892		☐	<i>MSH2</i> inversion	2226	Lynch syndrome
☐	<i>CHEK2</i>	9016		☐	<i>MSH6</i>	8512	Lynch syndrome
☐	<i>DICER1</i>	5260		☐	<i>MUTYH</i>	4661	<i>MUTYH</i> -associated polyposis
☐	<i>PALB2</i>	2366		☐	<i>PMS2</i>	4646	Lynch syndrome
☐	<i>PTEN</i>	2106	<i>PTEN</i> -related disorders (including Cowden syndrome)	☐	<i>STK11</i>	2766	Peutz-Jeghers syndrome
☐	<i>TP53</i>	2866	Li-Fraumeni syndrome	Genitourinary Cancer			
Endocrine Tumors				☐	<i>BAP1</i>	9044	
☐	<i>MEN1</i>	2646	Multiple endocrine neoplasia type 1	☐	<i>FH</i>	6301	Hereditary leiomyomatosis and renal cell cancer
☐	<i>RET</i> gene sequence	2680	Multiple endocrine neoplasia type 2	☐	<i>FLCN</i>	5921	Birt-Hogg-Dubé syndrome
Gastrointestinal Cancer				☐	<i>VHL</i>	2606	Von-Hippel Lindau disease
☐	<i>APC</i>	3040	Familial adenomatous polyposis	☐	<i>TSC1</i> and <i>TSC2</i>	5904	Tuberous sclerosis complex
☐	<i>APC</i> and <i>MUTYH</i> concurrent	8726	Adenomatous polyposis	Skin Cancer/Melanoma			
☐	<i>BMPRI1</i> and <i>SMAD4</i> concurrent	8604	Juvenile polyposis syndrome	☐	<i>CDKN2A</i> and <i>CDK4</i> concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
☐	<i>CDH1</i>	4726	Hereditary diffuse gastric cancer	☐	<i>PTCH1</i>	5684	Gorlin syndrome
☐	<i>EPCAM</i> del/dup	8519	Lynch syndrome	Other Hereditary Cancer Testing			
☐	Lynch syndrome (concurrent)	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM</i> del/dup	☐	<i>NF1</i>	5704	Neurofibromatosis type 1
				☐	<i>NF2</i>	9024	Neurofibromatosis type 2
				☐	<i>RB1</i>	5426	Hereditary retinoblastoma
				☐	<i>SMARCB1</i>	7180	Schwannomatosis
Other Single Syndrome Orders				Please visit ambrygen.com/hereditary-cancer-single-gene-tests for details.			
☐				Test Code(s): _____ Gene/Test Name(s): _____			

Comprehensive Test Requisition Form - Page 4 of 7

Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description				
CARDIOLOGY											
Comprehensive Cardiovascular Panels				Familial Hypercholesterolemia							
☐	CardioNext®	8911	92 genes for hereditary cardiomyopathies and arrhythmias	☐	FHNext®	8680	4 genes (<i>APOB, LDLR, LDLRAP1, PCSK9</i>)				
☐	CustomNext-Cardio®	9520	Up to 167 genes related to hereditary cardiomyopathies, arrhythmias, TAA, HHT, Noonan, and lipidemias. Required: completed CustomNext-Cardio supplemental form. ambrygen.com/forms	☐ Check this box if you would like to have the <i>SLC01B1</i> c.521T>C polymorphism reported with FHNext, which has been associated in medical literature with statin-induced myopathies							
Arrhythmia Panels				☐	FCSNext (Familial Chylomicronemia Syndrome)	8920	<i>APOA5, APOC2, GPIHBP1, LMF1, LPL</i>				
☐	LongQTNext™	8890	17 genes for long QT, Brugada and short QT syndromes	☐	Sitosterolemia	8930	<i>ABCG5, ABCG8</i>				
☐	RhythmNext®	8900	42 genes for long QT syndrome, Brugada and short QT syndromes, CPVT and ARVC	Aneurysms and Related Disorders							
☐	CPVTNext®	8902	4 genes for catecholaminergic polymorphic ventricular tachycardia	☐	TAADNext®	8789	35 genes for thoracic aortic aneurysms/dissections, Marfan syndrome, Ehlers-Danlos and related disorders				
Cardiomyopathy Panels				☐	Marfan reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext				
☐	HCMNext®	8936	30 genes for hypertrophic cardiomyopathy	Hereditary Hemorrhagic Telangiectasia (HHT)							
☐	HCMNext Reflex	8883	<i>MYBPC3, MYH7</i> reflex to HCMNext	☐	HHTNext®	8672	<i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>				
☐	DCMNext®	8884	37 genes for dilated cardiomyopathy	Noonan Syndrome							
☐	CMNext®	8887	56 genes for hereditary cardiomyopathy	☐	NoonanNext™	8402	18 genes for RASopathies				
☐	ARVCNext™	8904	11 genes for arrhythmogenic right ventricular cardiomyopathy	Other							
				☐	Transthyretin amyloidosis	1560	<i>TTR</i>				
CLINICAL GENOMICS											
For Reflex or Concurrent Testing:											
Test 1: _____ ☐ Reflex to _____ Test 2: _____ ☐ Reflex to _____ Test 3: _____ ☐ Concurrent with _____ ☐ Concurrent with _____											
See Reflex or Concurrent Testing section of the Supplemental Information page.											
Chromosomal Microarray											
☐	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)	☐	Familial targeted microarray	5495	Paid option. Only available following SNP Array (5490) completed at Ambry. Incidental findings unrelated to the variant(s) detected in the proband will NOT be reported. Name of proband tested at Ambry: _____				
Exome											
! REQUIRED: Select a Primary Test Order											
☐	ExomeNext®-Proband	9993	Proband only exome sequencing	☐	ExomeNext-Trio	9995	Trio exome sequencing				
☐	ExomeNext-Proband plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing	☐	ExomeNext-Trio plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing				
☐	ExomeNext-Duo	9991	Duo exome sequencing	☐	ExomeNext-Rapid®	9999R	Rapid Trio exome sequencing plus mtDNA sequencing (Institutional billing or patient payment only)				
☐	ExomeNext-Duo plus mtDNA	9992	Duo exome sequencing plus mtDNA sequencing								
If ordering ExomeNext/ExomeNext-Rapid, please complete:											
Secondary Findings Report: Check below to opt-out of the ACMG Recommended List of secondary findings. If left unchecked, secondary findings will be reported.											
☐ Opt-out: I choose to decline the ACMG Recommended List of secondary findings											
ExomeNext Supplemental Test Options											
☐	ExomeReveal™	9990	RNA analysis available with all ExomeNext orders except for ExomeNext-Rapid, EDTA and PAXgene RNA tubes required								
ENDOCRINOLOGY											
☐	Hereditary leiomyomatosis renal cell carcinoma	6301	<i>FH</i>	☐	Multiple endocrine neoplasia type 2 and familial medullary thyroid cancer (FMTC)	2680	<i>RET</i> gene sequence				
☐	Multiple endocrine neoplasia type I	2646	<i>MEN1</i>	☐	Neurofibromatosis type 1	5704	<i>NF1</i>				
				☐	von-Hippel Lindau disease	2606	<i>VHL</i>				

Comprehensive Test Requisition Form - Page 5 of 7

GASTROENTEROLOGY							
☐	CFTR gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status	☐	Juvenile polyposis syndrome	8604	BMPR1A, SMAD4
☐	Hirschsprung disease (RET-related)	2680	RET gene sequence	☐	Pancreatitis	8022	CASR, CFTR, CPA1, CTSC, PRSS1, SPINK1
☐				☐	Peutz-Jeghers syndrome	2766	STK11
HEMATOLOGY/ONCOLOGY							
☐	Shwachman-Diamond syndrome	1440	SBDS				
NEUROLOGY							
☐ Opt-in to Reporting of Variants of Unknown Significance (VUS) For patients undergoing an epilepsy or neurodevelopmental disorder panel, checking this box indicates that VUS identified on the test(s) ordered below will be reported for this patient. If you do not check this box, VUS will NOT be reported.							
☐ Parental samples provided for cosegregation Cosegregation testing of family members is available for the following panels: EpilepsyNext, EpilepsyNext-Expanded, AutismNext, NeurodevelopmentNext							
For Reflex or Concurrent Testing: Test 1: _____ ☐ Reflex to _____ Test 2: _____ ☐ Reflex to _____ Test 3: _____ ☐ Concurrent with _____ ☐ Concurrent with _____ See Reflex or Concurrent Testing section of the Supplemental Information page.							
Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
Epilepsy				Neurodevelopmental Disorders			
☐	EpilepsyNext®	6864	124 genes for epilepsy	☐	AutismNext®	6863	72 genes for non-syndromic autism spectrum disorders and/or intellectual disability
☐	EpilepsyNext-Expanded™	6865	>890 genes associated with seizures, primarily with neonatal to childhood onset	☐	Autism, macrocephaly	2106	PTEN
Hereditary Neuropathy				☐	Fragile X syndrome	4544	FMR1 repeat expansion analysis and methylation studies
☐	Familial transthyretin amyloidosis	1560	TTR	☐	NeurodevelopmentNext™	6861	202 genes known to cause developmental delay, intellectual disability and/or autism spectrum disorders
Neurocutaneous/Neuro-Oncology Disorders							
☐	Ataxia-telangiectasia	9014	ATM	☐	Neurofibromatosis 2	9024	NF2
☐	HHTNext®	8672	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4	☐	Nevoid basal cell carcinoma syndrome/Gorlin syndrome	5684	PTCH1
☐	Legius syndrome	5724	SPRED1	☐	Tuberous sclerosis complex	5904	TSC1, TSC2
☐	Li-Fraumeni syndrome	2866	TP53	☐	von Hippel-Lindau disease	2606	VHL
☐	Neurofibromatosis 1	5704	NF1				
PULMONOLOGY							
Congenital Central Hypoventilation Syndrome				Primary Ciliary Dyskinesia			
☐	Congenital central hypoventilation syndrome	1580	PHOX2B gene sequence	☐	PCDNext®	8122	21 genes for primary ciliary dyskinesia ☐ Report poly T/TG status
Cystic Fibrosis				Pulmonary Fibrosis			
☐	CFTR gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status	☐	Telomere-related pulmonary fibrosis	8140	TERT, TERC
				Respiratory Distress Syndrome			
				☐	Surfactant dysfunction (respiratory distress syndrome)	8100	ABCA3, SFTPB, SFTPC gene sequence
VASCULAR							
☐	HHTNext®	8672	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4	☐	TAADNext®	8789	35 genes for thoracic aortic aneurysms
☐	Marfan syndrome reflex to TAADNext	8783	FBN1 reflex to TAADNext				
SPECIFIC SITE ANALYSIS (Please include a copy of relative's report)							
Gene(s): _____ Mutation(s): _____ Relative Name: _____							
Relationship to Relative: _____ Accession # (if tested at Ambry): _____							
Positive control sample: ☐ will be provided ☐ already at Ambry ☐ not available							

Hereditary Cancer Multi-Gene Tests

TEST NAME	TEST CODE	GENES
Pan-cancer		
CancerNext® (40 genes)	8824	APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, FH, FLCN, GREM1, HOXB13, MBD4, MET, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RPS20, SMAD4, STK11, TP53, TSC1, TSC2, VHL
CancerNext-Expanded® (77 genes or up to 90 genes w/ add-ons)	8875	AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, CTNNA1, DDX41, DICER1, EGFR, EPCAM, ETV6, FH, FLCN, GATA2, GREM1, HOXB13, KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, RPS20, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WT1 Optional Add-on 1 - Limited Evidence Genes (8 genes): ATRIP, EGLN1, KIF1B, MLH3, PALLD, RAD51B, RNF43, TERT Optional Add-on 2 - Pancreatitis Genes (5 genes): CFTR, CPA1, CTSC, PRSS1, SPINK1
STAT Breast Management		
BRCAPlus® (13 genes)	8836	ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53
Breast & gynecologic		
BRCANext® (19 genes or up to 26 genes w/ add-on)	8857	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53 Optional Add-on - Limited Evidence Genes (7 genes): ATRIP, CDC73, FH, NTHL1, POLD1, POLE, RAD51B
Colorectal & polyposis		
ColoNext® (21 genes or up to 26 genes w/ add-on)	8821	APC, AXIN2, BMPR1A, CDH1, EPCAM, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RPS20, SMAD4, STK11, TP53 Optional Add-on - Limited Evidence Genes (5 genes): ATM, CHEK2, CTNNA1, MLH3, RNF43
Customizable		
CustomNext-Cancer® (up to 90 genes) Required: complete CustomNext-Cancer supplemental form. ambrygen.com/forms	9511	To order all genes on Ambry's oncology menu, please order CancerNext-Expanded. AIP, ALK, APC, ATM, ATRIP, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CFTR, CHEK2, CPA1, CTNNA1, CTSC, DICER1, DDX41, EGFR, EGLN1, EPCAM, ETV6, FH, FLCN, GATA2, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PALLD, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POT1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD51B, RAD51C, RAD51D, RB1, RET, RNF43, RPS20, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, STK11, SUFU, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WT1 For Medicare Patients: At a minimum, the following core genes must be included in the panel to ensure Medicare coverage: APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, TP53.
Syndrome specific		
Adenomatous polyposis	8726	APC, MUTYH
BRCA1/2-associated hereditary breast and ovarian cancer (HBOC)	8838	BRCA1, BRCA2
Lynch syndrome	8517	MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup

Specimen Requirements

Blood/saliva from patients with a history of allogenic bone marrow or stem cell transplant cannot be used for genetic testing. Blood/saliva from patients with active hematological disease is not recommended. An alternative specimen may be needed. Please see ambrygen.com/specimen-requirements for details.

Specific site analysis for variants identified at an external laboratory must be accompanied by a copy of the original testing report. A positive control from a known positive family member is recommended (required for prenatal testing).

Please note that Ambry cannot guarantee the viability of your specimen for testing at our laboratory, given the logistics of international specimen transfer. Testing may not be completed on specimens of inadequate quality due to specimen transfer issues not under Ambry's control, including, but not limited to, delays at customs, or other transfer-related delays. Ambry or your health care provider will reach out to you in such a case to rearrange for a specimen collection and transfer for completion of the ordered test.

Reflex or Concurrent Testing

Concurrent testing is when multiple tests are initiated at the same time. When multiple tests are ordered on the same test requisition form, testing will be run concurrently unless otherwise specified.

Reflex testing is when a subsequent test is initiated pending the outcome of the initial test. Reflex testing may result in delayed reporting of results.

For reflex test orders:

- Any diagnostic finding at any step will result in cancellation of any subsequent reflex tests.
- Non-diagnostic findings (including VUS or Uncertain results) will automatically reflex to the subsequent test.
- Secondary findings results do not impact whether a subsequent test is initiated or canceled.

When ordering STAT panels (such as BRCAplus®), the results of the STAT panel will be prioritized and reported with a shorter turnaround time, even if the tests were run concurrently.

Known Familial Variants

Variant-specific report comments about the presence or absence of known familial variant(s) require the "Known Familial Variant" section of this form to be completed accurately, including an internal Ambry reference ID and/or a copy of the positive family member's lab report. Acceptable types of Ambry identifiers include:

- Accession number
- Order number
- Name and date of birth

Variant requests without an internal Ambry reference ID or positive family member's lab report will not receive a variant-specific report comment.

Variant-specific report comments are not included in ExomeNext or Neurology panel reports.