

To submit an order via email, please send the completed test requisition form to info@ambrygen.com

PATIENT INFORMATION					
Name (Last, First, MI)			Sex at Birth <input type="checkbox"/> F <input type="checkbox"/> M	Date of Birth (MM/DD/YY)	MRN
Ethnicity: <input type="checkbox"/> African American <input type="checkbox"/> Asian <input type="checkbox"/> White <input type="checkbox"/> Hispanic <input type="checkbox"/> Portuguese <input type="checkbox"/> Native American <input type="checkbox"/> Other:					Ashkenazi Jewish <input type="checkbox"/> Yes <input type="checkbox"/> No
Address		City		State	Zip
Phone			Email		
SPECIMEN INFORMATION (For phlebotomy service, select all services you are requesting)					
Type(s) <input type="checkbox"/> Blood (EDTA preferred) <input type="checkbox"/> Saliva <input type="checkbox"/> Buccal Swab* <input type="checkbox"/> DNA <input type="checkbox"/> Cord Blood** <input type="checkbox"/> Other**:			<input type="checkbox"/> Personal history of allogenic bone marrow or peripheral stem cell transplant*		
Collection Date	Specimen ID			Medical Record #	
<p>* 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</p> <p>** Fetal specimens, cord blood and POC will have maternal cell contamination studies added for a charge. Maternal and fetal specimen required. Please see bottom of page 5 for Maternal Cell Contamination sample submission test codes.</p> <p>^ Buccal swab sample available for chromosomal microarray (SNP array, familial targeted microarray), CustomNext-Neuro, epilepsy, ExomeNext, fragile X syndrome, hereditary neuropathy (familial transthyretin amyloidosis), HHTNext, migraine (familial hemiplegic migraine), and neurodevelopmental disorder tests. Buccal swab samples from patients with a history of allogenic bone marrow or stem cell transplant should not be used for genetic testing. For these patients, an alternative specimen (e.g. cultured fibroblasts) is required. Testing on buccal swab samples from patients with active hematological disease is not recommended. An alternative specimen (e.g. cultured fibroblasts) is recommended. Please see ambrygen.com/specimen-requirements for details.</p>					
Phlebotomy Services Request: <input type="checkbox"/> Phlebotomy draw <input type="checkbox"/> Insurance preverification first <input type="checkbox"/> Send blood kit to patient† <input type="checkbox"/> Send saliva kit to patient <input type="checkbox"/> Send pediatric saliva kit to patient					
† As the patient's clinician, I am unaware of any potential for complication or difficulty in drawing blood for the listed patient(s). I understand that the phlebotomist has full authority to refuse to draw any patient if the safety of the phlebotomist and/or patient(s) are in question.					
INDICATION(S) FOR TESTING					
ICD-10 code(s):					
Will the medical management change depending on the results of the test? <input type="checkbox"/> Yes <input type="checkbox"/> No					
Was genetic counseling completed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown Date Genetic Counseling was Performed: _____					
PRENATAL SAMPLES ONLY					
Sample type: <input type="checkbox"/> Direct CVS <input type="checkbox"/> Cultured CVS <input type="checkbox"/> Cultured amnio <input type="checkbox"/> POC <input type="checkbox"/> Cultured POC				Gestational age at sample collection	
ORDERING LICENSED PROVIDER/SENDING FACILITY (Each listed person will receive a copy of the report)					
Facility Name (Facility Code)		Address		City	State /Country Zip Phone
Ordering Licensed Provider Name (Last, First)(Code)		NPI#	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, PRE-TEST GENETIC COUNSELING, AND MEDICAL NECESSITY FOR GENETIC TESTING					
The undersigned person (or representative thereof) ensures he/she is a licensed medical professional authorized to order genetic testing and confirms that the patient has given appropriate consent. I confirm that testing is medically necessary and that test results may impact medical management for the patient. 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. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the attached letter of medical necessity.					
Signature Required for Processing Medical Professional Signature:				Date:	
INSURANCE BILLING (Include copy of both sides of insurance card)			INSTITUTIONAL BILLING		
Patient Relation to Policy Holder? <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Child		Name and DOB of Policy Holder (if not self)		Facility Name <input type="checkbox"/> Send invoice to facility address above	
Insurance Company		Policy #	HMO Auth #	Address	
Out of Pocket: In many cases, we will start testing immediately (may vary by test or for patients with government insurance). We will attempt to contact the patient if the estimated out-of-pocket costs are > USD \$100.			Contact Name		
Special Billing Notes:			Phone Number		Email/Fax
			<input type="checkbox"/> PATIENT PAYMENT		<input type="checkbox"/> Check (Payable to Ambry Genetics) <input type="checkbox"/> Credit Card (Call 949-900-5795)
<p>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.</p> <p>For patient payment by credit card: I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above. In order to expedite consideration for eligibility for Ambry's Patient Assistance Program, please provide the total annual gross household income: \$ _____ and the number of family members in the household supported by the listed income: _____. I authorize Ambry Genetics Corporation to verify the above information for the sole purpose of assessing financial need, including the right to seek supporting documentation.</p>					
For NY Residents: <input type="checkbox"/> I am a New York resident and I give Ambry Genetics permission to store my sample for longer than 60 days. NOTE: If left blank, consent is interpreted as "NO".					
Signature Required For Insurance/Self-Pay Patients and NY Sample Storage Consent:				Date:	

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

Cancer History Not Applicable Metastatic: Yes No

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

Previously Detected Alteration(s): _____ Gene Name: _____ Testing Lab: _____
 Patient previously tested at Ambry? Yes No Family previously tested at Ambry? Yes No
 Name: _____ DOB: _____ Relation: _____

FAMILY HISTORY (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: _____

Relation to patient	Maternal	Paternal	Disease	Dx age
	<input type="checkbox"/>	<input type="checkbox"/>		
	<input type="checkbox"/>	<input type="checkbox"/>		
	<input type="checkbox"/>	<input type="checkbox"/>		
	<input type="checkbox"/>	<input type="checkbox"/>		

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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.

For multiple test orders, testing will be run concurrently (multiple tests initiated at the same time) unless otherwise specified. To order reflexive testing (second test starts pending first test outcome), please clearly indicate the order of reflexive tests in the notes section or next to the test check box. For reflex test orders, any positive findings (pathogenic/likely pathogenic) in the first test will be reported out to the clinician, and the requested second test will be canceled; all other findings will automatically reflex (including VUS).

CANCER TEST ORDERS			
REQUIRED: Select a Primary Test Order		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.)	
For Patients Meeting <i>BRCA1/2</i> Testing Criteria		<input type="checkbox"/> BrainTumorNext®	8847 29 gene brain tumor test
<input type="checkbox"/> <i>BRCA1/2</i> test		<input type="checkbox"/> BRCANext™	8855 18 gene breast cancer test
For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (Lynch)		<input type="checkbox"/> BRCANext-Expanded™	8860 23 gene breast cancer test
Lynch Syndrome test: <input type="checkbox"/> <i>MLH1, MSH2, MSH6, PMS2, EPCAM</i>		<input type="checkbox"/> BRCAplus®	8836 8 gene breast cancer test
For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (polyposis)		<input type="checkbox"/> CancerNext®	8824 36 gene cancer test
Polyposis test: <input type="checkbox"/> <i>APC/MUYTH</i>		<input type="checkbox"/> CancerNext-Expanded®	8874 77 gene cancer test
<input type="checkbox"/> Other: _____		<input type="checkbox"/> ColoNext®	8822 20 gene colorectal cancer test
<input type="checkbox"/> None of the above (patient does not meet any genetic testing criteria)		<input type="checkbox"/> CustomNext-Cancer®	9510 up to 91 gene custom test**
Other Supplemental Test Options (Select if applicable)			
<input type="checkbox"/> +RNAinsight® (Not available with BRCAplus, pancreatitis panel, or STAT orders; PAXgene® tube required for RNA)		<input type="checkbox"/> MelanomaNext®	8849 9 gene melanoma test
		<input type="checkbox"/> PancNext®	8042 13 gene pancreatic cancer test
		<input type="checkbox"/> Pancreatitis panel	8022 6 gene pancreatitis test
		<input type="checkbox"/> PGLNext®	5504 14 gene PGL/PCC test
		<input type="checkbox"/> ProstateNext®	8845 14 gene prostate cancer test
		<input type="checkbox"/> RenalNext®	5900 20 gene renal cancer test

Order	Test Name	Test Code	Description
Breast and/or Ovarian Cancer			
<input type="checkbox"/>	<i>ATM</i>	9014	Ataxia-telangiectasia
<input type="checkbox"/>	<i>BRCA1/2</i>	8838	Hereditary breast and ovarian cancer
<input type="checkbox"/>	<i>BRCA1/2</i> Ashkenazi Jewish 3-site mutation panel	5892	
<input type="checkbox"/>	<i>CHEK2</i>	9016	
<input type="checkbox"/>	<i>DICER1</i>	5260	
<input type="checkbox"/>	<i>PALB2</i>	2366	
<input type="checkbox"/>	<i>PTEN</i>	2106	<i>PTEN</i> -related disorders (including Cowden syndrome)
<input type="checkbox"/>	<i>TP53</i>	2866	Li-Fraumeni syndrome
Endocrine Tumors			
<input type="checkbox"/>	<i>MEN1</i>	2646	Multiple endocrine neoplasia type 1
<input type="checkbox"/>	<i>RET</i> gene sequence	2680	Multiple endocrine neoplasia type 2
Gastrointestinal Cancer			
<input type="checkbox"/>	<i>APC</i>	3040	Familial adenomatous polyposis
<input type="checkbox"/>	<i>APC</i> and <i>MUTYH</i> concurrent	8726	Adenomatous polyposis
<input type="checkbox"/>	<i>BMPRIA</i> and <i>SMAD4</i> concurrent	8604	Juvenile polyposis syndrome
<input type="checkbox"/>	<i>CDH1</i>	4726	Hereditary diffuse gastric cancer
<input type="checkbox"/>	<i>EPCAM</i> del/dup	8519	Lynch syndrome
<input type="checkbox"/>	Lynch syndrome (concurrent)	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM</i> del/dup

Order	Test Name	Test Code	Description
Gastrointestinal Cancer (Cont.)			
<input type="checkbox"/>	<i>MLH1</i>	8508	Lynch syndrome
<input type="checkbox"/>	<i>MSH2 + EPCAM</i> del/dup	8510	Includes <i>MSH2</i> inversion
<input type="checkbox"/>	<i>MSH2</i> inversion	2226	Lynch syndrome
<input type="checkbox"/>	<i>MSH6</i>	8512	Lynch syndrome
<input type="checkbox"/>	<i>MUTYH</i>	4661	<i>MUTYH</i> -associated polyposis
<input type="checkbox"/>	<i>PMS2</i>	4646	Lynch syndrome
<input type="checkbox"/>	<i>STK11</i>	2766	Peutz-Jeghers syndrome
Genitourinary Cancer			
<input type="checkbox"/>	<i>BAP1</i>	9044	
<input type="checkbox"/>	<i>FH</i>	6301	Hereditary leiomyomatosis and renal cell cancer
<input type="checkbox"/>	<i>FLCN</i>	5921	Birt-Hogg-Dubé syndrome
<input type="checkbox"/>	<i>VHL</i>	2606	Von-Hippel Lindau disease
<input type="checkbox"/>	<i>TSC1</i> and <i>TSC2</i>	5904	Tuberous sclerosis complex
Skin Cancer/Melanoma			
<input type="checkbox"/>	<i>CDKN2A</i> and <i>CDK4</i> concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
<input type="checkbox"/>	<i>PTCH1</i>	5684	Gorlin syndrome
Other Hereditary Cancer Testing			
<input type="checkbox"/>	<i>NF1</i>	5704	Neurofibromatosis type 1
<input type="checkbox"/>	<i>NF2</i>	9024	Neurofibromatosis type 2
<input type="checkbox"/>	<i>RB1</i>	5426	Hereditary retinoblastoma
<input type="checkbox"/>	<i>SMARCB1</i>	7180	Schwannomatosis
Other Single Syndrome Orders			
<input type="checkbox"/>	Please visit ambrygen.com/hereditary-cancer-single-gene-tests for details.		
	Test Code(s): _____ Gene/Test Name(s): _____		

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Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
CARDIOLOGY							
Comprehensive Cardiovascular Panels				Familial Hypercholesterolemia			
<input type="checkbox"/>	CardioNext®	8911	92 genes for hereditary cardiomyopathies and arrhythmias	<input type="checkbox"/>	FHNNext®	8680	4 genes (<i>APOB, LDLR, LDLRAP1, PCSK9</i>)
<input type="checkbox"/>	CustomNext-Cardio®	9520	Up to 167 genes related to hereditary cardiomyopathies, arrhythmias, TAAD, HHT, Noonan, and lipidemias. Required: completed CustomNext-Cardio supplemental form. ambrygen.com/forms	<input type="checkbox"/> Check this box if you would like to have the <i>SLC01B1</i> c.521T>C polymorphism reported with FHNNext, which has been associated in medical literature with statin-induced myopathies			
Arrhythmia Panels				<input type="checkbox"/>	FCSNext (Familial Chylomicronemia Syndrome)	8920	<i>APOA5, APOC2, GPIHBP1, LMF1, LPL</i>
<input type="checkbox"/>	LongQTNext™	8890	17 genes for long QT, Brugada and short QT syndromes	<input type="checkbox"/>	Sitosterolemia	8930	<i>ABCG5, ABCG8</i>
<input type="checkbox"/>	RhythmNext®	8900	42 genes for long QT syndrome, Brugada and short QT syndromes, CPVT and ARVC	Aneurysms and Related Disorders			
<input type="checkbox"/>	CPVTNext®	8902	4 genes for catecholaminergic polymorphic ventricular tachycardia	<input type="checkbox"/>	TAADNext®	8789	35 genes for thoracic aortic aneurysms/dissections, Marfan syndrome, Ehlers-Danlos and related disorders
Cardiomyopathy Panels				<input type="checkbox"/>	Marfan reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext
<input type="checkbox"/>	HCMNext®	8936	30 genes for hypertrophic cardiomyopathy	Hereditary Hemorrhagic Telangiectasia (HHT)			
<input type="checkbox"/>	HCMNext Reflex	8883	<i>MYBPC3, MYH7</i> reflex to HCMNext	<input type="checkbox"/>	HHTNext®	8672	<i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>
<input type="checkbox"/>	DCMNext®	8884	37 genes for dilated cardiomyopathy	Noonan Syndrome			
<input type="checkbox"/>	CMNext®	8887	56 genes for hereditary cardiomyopathy	<input type="checkbox"/>	NoonanNext™	8402	18 genes for RASopathies
<input type="checkbox"/>	ARVCNext™	8904	11 genes for arrhythmogenic right ventricular cardiomyopathy	Other			
				<input type="checkbox"/>	Transthyretin amyloidosis	1560	<i>TTR</i>
CLINICAL GENOMICS							
<input type="checkbox"/>	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)	<input type="checkbox"/>	ExomeNext-Trio	9995	Trio exome sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	Familial targeted microarray	5495	Paid option. Only available following SNP Array (5490) completed at Ambyr. Incidental findings unrelated to the variant(s) detected in the proband, will NOT be reported. Name of proband tested at Ambyr: _____	<input type="checkbox"/>	ExomeNext-Trio plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	ExomeNext®-Proband	9993	Proband only exome sequencing	<input type="checkbox"/>	ExomeNext-Rapid®	9999R	<input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	ExomeNext-Proband plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing	Must be ordered through AP*			
<input type="checkbox"/>	ExomeNext-Duo	9991	Duo exome sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies	<input type="checkbox"/>	ExomeNext-Select	9500	Up to 500 gene custom exome sequencing test
<input type="checkbox"/>	ExomeNext-Duo plus mtDNA	9992	Duo exome sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies	*AP is AmbyrPort®, our online portal ambrygen.com/ap			
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. Secondary findings are not available for ExomeNext-Select orders. <input type="checkbox"/> Opt-out: I choose to decline the ACMG Recommended List of secondary findings							
ENDOCRINOLOGY							
<input type="checkbox"/>	Hereditary leiomyomatosis renal cell carcinoma	6301	<i>FH</i>	<input type="checkbox"/>	Multiple endocrine neoplasia type 2 and familial medullary thyroid cancer (FMTC)	2680	<i>RET</i> gene sequence
<input type="checkbox"/>	Maturity-onset diabetes of the young	8310	<i>HNF1A, HNF4A, HNF1B, GCK, PDX1</i>	<input type="checkbox"/>	Neurofibromatosis type 1	5704	<i>NF1</i>
<input type="checkbox"/>	Multiple endocrine neoplasia type I	2646	<i>MEN1</i>	<input type="checkbox"/>	PGLNext®	5504	14 gene PGL/PCC test
				<input type="checkbox"/>	von-Hippel Lindau disease	2606	<i>VHL</i>
GASTROENTEROLOGY							
<input type="checkbox"/>	<i>CFTR</i> gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Juvenile polyposis syndrome	8604	<i>BMPR1A, SMAD4</i>
<input type="checkbox"/>	Hirschsprung disease (<i>RET</i> -related)	2680	<i>RET</i> gene sequence	<input type="checkbox"/>	Pancreatitis	8022	<i>CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1</i>
				<input type="checkbox"/>	Peutz-Jeghers syndrome	2766	<i>STK11</i>
HEMATOLOGY/ONCOLOGY							
<input type="checkbox"/>	DBANext	8550	11 genes for Diamond-Blackfan anemia	<input type="checkbox"/>	Shwachman-Diamond syndrome	1440	<i>SBDS</i>
<input type="checkbox"/>	DCNext	8161	7 genes for dyskeratosis congenita				
MULTIPLE CONGENITAL ANOMALIES				NEPHROLOGY			
<input type="checkbox"/>	NoonanNext™	8402	18 genes for Noonan syndrome and RASopathies	<input type="checkbox"/>	NephrolithiasisNext	9580	30 genes for nephrolithiasis and nephrocalcinosis

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NEUROLOGY							
<input type="checkbox"/> Opt-in to Reporting of Variants of Unknown Significance (VUS) For patients undergoing an epilepsy, neurodevelopmental disorder, or familial hemiplegic migraine 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.							
Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
Comprehensive Neuro Testing				Neurodevelopmental Disorders			
Must order through Ambry-Port	CustomNext-Neuro™	9545	Customizable test of up to 500 genes from neuro menu <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	AutismNext®	6863	72 genes for non-syndromic autism spectrum disorders and/or intellectual disability <input type="checkbox"/> Check if parental samples are included
	Epilepsy			<input type="checkbox"/>	Autism, macrocephaly	2106	PTEN
	<input type="checkbox"/>	EpiRapid®	6862	22 epilepsy genes with treatment associations <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Fragile X syndrome	4544
<input type="checkbox"/>	EpilepsyNext®	6864	124 genes for epilepsy <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	NeurodevelopmentNext™	6861	202 genes known to cause developmental delay, intellectual disability and/or autism spectrum disorders <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	EpilepsyNext-Expanded™	6865	>890 genes associated with seizures, primarily with neonatal to childhood onset <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	NeurodevelopmentNext-Expanded	6860	>1,400 genes associated with neonatal to childhood onset developmental delay, seizures, intellectual disability, developmental regression, autism spectrum disorders <input type="checkbox"/> Check if parental samples are included
Hereditary Neuropathy				Neurocutaneous/Neuro-Oncology Disorders			
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	TTR	<input type="checkbox"/>	Ataxia-telangiectasia	9014	ATM
Migraine				<input type="checkbox"/>	BrainTumorNext®	8847	29 genes for brain tumors
<input type="checkbox"/>	Familial hemiplegic migraine	6866	ATP1A2, CACNA1A, PRRT2, SCN1A <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	HHTNext®	8672	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4
Note: Gene lists for NeurodevelopmentNext-Expanded and EpilepsyNext-Expanded are regularly updated 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				<input type="checkbox"/>	Legius syndrome	5724	SPRED1
For Reflex or Concurrent Testing: Test 1: _____ <input type="checkbox"/> Reflex to _____ Test 2: _____ <input type="checkbox"/> Concurrent with _____				<input type="checkbox"/>	Li-Fraumeni syndrome	2866	TP53
				<input type="checkbox"/>	Neurofibromatosis 1	5704	NF1
				<input type="checkbox"/>	Neurofibromatosis 2	9024	NF2
				<input type="checkbox"/>	Nevoid basal cell carcinoma syndrome/Gorlin syndrome	5684	PTCH1
				<input type="checkbox"/>	Tuberous sclerosis complex	5904	TSC1, TSC2
				<input type="checkbox"/>	von Hippel-Lindau disease	2606	VHL
PULMONOLOGY							
Congenital Central Hypoventilation Syndrome				Primary Ciliary Dyskinesia			
<input type="checkbox"/>	Congenital central hypoventilation syndrome	1580	PHOX2B gene sequence	<input type="checkbox"/>	PCDNext®	8122	21 genes for primary ciliary dyskinesia <input type="checkbox"/> Report poly T/TG status
Cystic Fibrosis				Pulmonary Fibrosis			
<input type="checkbox"/>	508 FIRST®	1002	CFTR deltaF508 mutation analysis with reflex to CFTR gene sequence and deletion/duplication <input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Telomere-related pulmonary fibrosis	8140	TERT, TERC
				Respiratory Distress Syndrome			
<input type="checkbox"/>	CFTR gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Surfactant dysfunction (respiratory distress syndrome)	8100	ABCA3, SFTPB, SFTPC gene sequence
VASCULAR							
<input type="checkbox"/>	HHTNext®	8672	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4	<input type="checkbox"/>	TAADNext®	8789	35 genes for thoracic aortic aneurysms
<input type="checkbox"/>	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: <input type="checkbox"/> will be provided <input type="checkbox"/> already at Ambry <input type="checkbox"/> not available							
FOR PRENATAL SPECIMENS, POC OR CORD BLOOD: MATERNAL CELL CONTAMINATION ANALYSIS REQUIRED							
Both maternal and fetal specimens are required.							
<input type="checkbox"/> 1260 MCC for fetal specimen or cord blood <input type="checkbox"/> 1262 MCC Reference for maternal blood sample (No Charge)							

Hereditary Cancer Multi-Gene Tests

Test Name	Test Code	Genes
Adenomatous polyposis	8726	<i>APC, MUTYH</i>
BrainTumorNext® (29 genes)	8847	<i>AIP, ALK, APC, CDKN1B, CDKN2A, DICER1, EPCAM, LZTR1, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL</i>
BRCANext™ (18 genes)	8855	<i>ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53</i>
BRCANext-Expanded™ (23 genes)	8860	<i>ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53</i>
BRCApplus® (8 genes)	8836	<i>ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, TP53</i>
CancerNext® (36 genes)	8824	<i>APC, ATM, AXIN2, BARD1, BRCA1, BRCA2, BRIP1, BMPR1A, CDH1, CDK4, CDKN2A, CHEK2, DICER1, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RECQL, SMAD4, SMARCA4, STK11, TP53</i>
CancerNext-Expanded® (77 genes)	8874	<i>AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, BMPR1A, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EGLN1, EPCAM, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MIF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RECQL, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2</i>
ColoNext® (20 genes)	8822	<i>APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53</i>
CustomNext-Cancer® (up to 91 genes) Required: complete CustomNext-Cancer supplemental form. ambyr.com/forms	9510	<i>AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, BMPR1A, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CFTR, CHEK2, CPA1, CTNNA1, CTRC, DICER1, EGFR, EGLN1, EPCAM, FAM175A(ABRAXAS1)†, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MIF, MLH1, MLH3†, MRE11A†, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PALLD†, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50†, RAD51C, RAD51D, RB1, RECQL, RET, RINT1†, RPS20†, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, STK11, SUFU, TERT†, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2</i>
HBOC	8838	<i>BRCA1, BRCA2</i>
Lynch syndrome/HNPCC	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup</i>
MelanomaNext® (9 genes)	8849	<i>BAP1, BRCA2, CDK4, CDKN2A, MIF, POT1, PTEN, RB1, TP53</i>
PancNext® (13 genes)	8042	<i>APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53</i>
Pancreatitis panel (6 genes)	8022	<i>CASR, CFTR, CPA1, PRSS1, SPINK1, CTRC</i>
PGLNext® (14 genes)	5504	<i>EGLN1, FH, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL</i>
ProstateNext® (14 genes)	8845	<i>ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, RAD51D, TP53</i>
RenalNext® (20 genes)	5900	<i>BAP1, CHEK2, EPCAM, FH, FLCN, MET, MIF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL</i>

† Limited evidence gene