

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 (MM/DD/YY)	Sex Assigned at Birth <input type="checkbox"/> F <input type="checkbox"/> M	Gender (optional) <input type="checkbox"/> Man <input type="checkbox"/> Woman <input type="checkbox"/> Nonbinary <input type="checkbox"/> Self-described
Genetic Ancestry: <input type="checkbox"/> Ashkenazi Jewish <input type="checkbox"/> Asian <input type="checkbox"/> Black/African American <input type="checkbox"/> French Canadian/Cajun <input type="checkbox"/> Hispanic/Latino <input type="checkbox"/> Mediterranean <input type="checkbox"/> Middle Eastern <input type="checkbox"/> Native American <input type="checkbox"/> Pacific Islander <input type="checkbox"/> Portuguese <input type="checkbox"/> White <input type="checkbox"/> Unknown <input type="checkbox"/> Other:				MRN
Address		City	State	Zip
Mobile #	Email			

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

<input type="checkbox"/> Personal history of allogenic bone marrow or peripheral stem cell transplant	
Specimen ID	Medical Record #

* 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.

 Collection Assistance: Phlebotomy draw** Send saliva kit to patient Send buccal swab kit to patient | Insurance preverification first (available for ExomeNext and SNP array only)

** 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
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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

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: _____

<input type="checkbox"/> INSURANCE BILLING (Include copy of both sides of insurance card)		<input type="checkbox"/> 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
Special Billing Notes:		Contact Name	
		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)	

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.

For NY Residents:
 I understand that New York State law requires Ambry Genetics to destroy my sample at the end of the testing process or not more than sixty days after the sample was taken. By checking this box, I agree that Ambry Genetics will instead retain my sample for at least 6 months after the testing above has been completed, and may (a) retain and use samples and health information for an indefinite period of time in accordance with applicable law; and (b) de-identify such samples and information and use and share the resulting de-identified samples and information in accordance with applicable law.

Patient Signature (I agree to terms above): _____

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 Tumor is MSI-High or IHC-Abnormal

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: <input type="checkbox"/> Fallopian tube <input type="checkbox"/> Primary peritoneal
Ovarian		
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

 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: _____
 My patient is the most informative family member available for testing. The affected relative and all intervening relatives are either deceased or unwilling/unavailable for testing.

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
	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/> Deceased <input type="checkbox"/> Declines <input type="checkbox"/> No Contact
	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/> Deceased <input type="checkbox"/> Declines <input type="checkbox"/> No Contact
	<input type="checkbox"/>	<input type="checkbox"/>			<input type="checkbox"/> Deceased <input type="checkbox"/> Declines <input type="checkbox"/> No Contact

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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. 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 <input type="checkbox"/> <i>BRCA1/2</i> test	For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (polyposis) Polyposis test: <input type="checkbox"/> <i>APC/MUTYH</i>
For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (Lynch) Lynch Syndrome test: <input type="checkbox"/> <i>MLH1, MSH2, MSH6, PMS2, EPCAM</i>	<input type="checkbox"/> Other: _____ <input type="checkbox"/> 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
<input type="checkbox"/>	8857	BRCANext®	Breast & gynecologic cancer test	<input type="checkbox"/>	8821	ColoNext®	Colorectal cancer & polyposis test
Add on: <input type="checkbox"/> Limited Evidence				Add on: <input type="checkbox"/> Limited Evidence			
<input type="checkbox"/>	8836	BRCAPlus®	STAT breast management test	<input type="checkbox"/>	9511	CustomNext-Cancer® Notes: _____	Custom test Gene content is required. Use CustomNext-Cancer supplemental form for guidance.
<input type="checkbox"/>	8824	CancerNext®	Pan-cancer test				
<input type="checkbox"/>	8875	CancerNext-Expanded®	Pan-cancer test				
Add on: <input type="checkbox"/> Limited Evidence							
Add on: <input type="checkbox"/> Pancreatitis							

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.)			
<input type="checkbox"/>	<i>ATM</i>	9014	Ataxia-telangiectasia	<input type="checkbox"/>	<i>MLH1</i>	8508	Lynch syndrome
<input type="checkbox"/>	<i>BRCA1/2</i>	8838	Hereditary breast and ovarian cancer	<input type="checkbox"/>	<i>MSH2 + EPCAM del/dup</i>	8510	Includes <i>MSH2</i> inversion
<input type="checkbox"/>	<i>BRCA1/2</i> Ashkenazi Jewish 3-site mutation panel	5892		<input type="checkbox"/>	<i>MSH2</i> inversion	2226	Lynch syndrome
<input type="checkbox"/>	<i>CHEK2</i>	9016		<input type="checkbox"/>	<i>MSH6</i>	8512	Lynch syndrome
<input type="checkbox"/>	<i>DICER1</i>	5260		<input type="checkbox"/>	<i>MUTYH</i>	4661	<i>MUTYH</i> -associated polyposis
<input type="checkbox"/>	<i>PALB2</i>	2366		<input type="checkbox"/>	<i>PMS2</i>	4646	Lynch syndrome
<input type="checkbox"/>	<i>PTEN</i>	2106	<i>PTEN</i> -related disorders (including Cowden syndrome)	<input type="checkbox"/>	<i>STK11</i>	2766	Peutz-Jeghers syndrome
<input type="checkbox"/>	<i>TP53</i>	2866	Li-Fraumeni syndrome	Genitourinary Cancer			
Endocrine Tumors				<input type="checkbox"/>	<i>BAP1</i>	9044	
<input type="checkbox"/>	<i>MEN1</i>	2646	Multiple endocrine neoplasia type 1	<input type="checkbox"/>	<i>FH</i>	6301	Hereditary leiomyomatosis and renal cell cancer
<input type="checkbox"/>	<i>RET</i> gene sequence	2680	Multiple endocrine neoplasia type 2	<input type="checkbox"/>	<i>FLCN</i>	5921	Birt-Hogg-Dubé syndrome
Gastrointestinal Cancer				<input type="checkbox"/>	<i>VHL</i>	2606	Von-Hippel Lindau disease
<input type="checkbox"/>	<i>APC</i>	3040	Familial adenomatous polyposis	<input type="checkbox"/>	<i>TSC1</i> and <i>TSC2</i>	5904	Tuberous sclerosis complex
<input type="checkbox"/>	<i>APC</i> and <i>MUTYH</i> concurrent	8726	Adenomatous polyposis	Skin Cancer/Melanoma			
<input type="checkbox"/>	<i>BMPRIA</i> and <i>SMAD4</i> concurrent	8604	Juvenile polyposis syndrome	<input type="checkbox"/>	<i>CDKN2A</i> and <i>CDK4</i> concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
<input type="checkbox"/>	<i>CDH1</i>	4726	Hereditary diffuse gastric cancer	<input type="checkbox"/>	<i>PTCH1</i>	5684	Gorlin syndrome
<input type="checkbox"/>	<i>EPCAM</i> del/dup	8519	Lynch syndrome	Other Hereditary Cancer Testing			
<input type="checkbox"/>	Lynch syndrome (concurrent)	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM</i> del/dup	<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"/>	FHNext®	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 FHNext, which has been associated in medical literature with statin-induced myopathies			
Arrhythmia Panels				<input type="checkbox"/>	FCSNext (Familial Chylomicronemia Syndrome)	8920	<i>APOA5, APOC2, GPIIIBP1, 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							
For Reflex or Concurrent Testing:							
Test 1: _____		<input type="checkbox"/> Reflex to	Test 2: _____		<input type="checkbox"/> Reflex to	Test 3: _____	
		<input type="checkbox"/> Concurrent with			<input type="checkbox"/> Concurrent with		
See Reflex or Concurrent Testing section of the Supplemental Information page.							
Chromosomal Microarray							
<input type="checkbox"/>	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)	<input type="checkbox"/>	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							
<input type="checkbox"/>	ExomeNext®-Proband	9993	Proband only exome sequencing	<input type="checkbox"/>	ExomeNext-Trio	9995	Trio exome sequencing
<input type="checkbox"/>	ExomeNext-Proband plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing	<input type="checkbox"/>	ExomeNext-Trio plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing
<input type="checkbox"/>	ExomeNext-Duo	9991	Duo exome sequencing	<input type="checkbox"/>	ExomeNext-Rapid®	9999R	Rapid Trio exome sequencing plus mtDNA sequencing (Institutional billing or patient payment only)
<input type="checkbox"/>	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.							
<input type="checkbox"/> Opt-out: I choose to decline the ACMG Recommended List of secondary findings							
ExomeNext Supplemental Test Options							
<input type="checkbox"/>	ExomeReveal™	9990	RNA analysis available with all ExomeNext orders except for ExomeNext-Rapid, EDTA and PAXgene RNA tubes required				
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"/>	Multiple endocrine neoplasia type I	2646	<i>MEN1</i>	<input type="checkbox"/>	Neurofibromatosis type 1	5704	<i>NF1</i>
				<input type="checkbox"/>	von-Hippel Lindau disease	2606	<i>VHL</i>

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GASTROENTEROLOGY						
<input type="checkbox"/>	CFTR gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Juvenile polyposis syndrome	8604 BMPR1A, SMAD4
<input type="checkbox"/>	Hirschsprung disease (RET-related)	2680	RET gene sequence	<input type="checkbox"/>	Pancreatitis	8022 CFTR, CPA1, CTRC, PRSS1, SPINK1
<input type="checkbox"/>				<input type="checkbox"/>	Peutz-Jeghers syndrome	2766 STK11

HEMATOLOGY/ONCOLOGY			
<input type="checkbox"/>	Shwachman-Diamond syndrome	1440	SBDS

NEUROLOGY	
<input type="checkbox"/>	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.
<input type="checkbox"/>	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: _____	<input type="checkbox"/> Reflex to	Test 2: _____	<input type="checkbox"/> Reflex to
	<input type="checkbox"/> Concurrent with		<input type="checkbox"/> 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			
<input type="checkbox"/>	EpilepsyNext®	6864	124 genes for epilepsy	<input type="checkbox"/>	AutismNext®	6863	72 genes for non-syndromic autism spectrum disorders and/or intellectual disability
<input type="checkbox"/>	EpilepsyNext-Expanded™	6865	>890 genes associated with seizures, primarily with neonatal to childhood onset	<input type="checkbox"/>	Autism, macrocephaly	2106	PTEN
Hereditary Neuropathy				<input type="checkbox"/>	Fragile X syndrome	4544	FMR1 repeat expansion analysis and methylation studies
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	TTR	<input type="checkbox"/>	NeurodevelopmentNext™	6861	202 genes known to cause developmental delay, intellectual disability and/or autism spectrum disorders

Neurocutaneous/Neuro-Oncology Disorders							
<input type="checkbox"/>	Ataxia-telangiectasia	9014	ATM	<input type="checkbox"/>	Neurofibromatosis 2	9024	NF2
<input type="checkbox"/>	HHTNext®	8672	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4	<input type="checkbox"/>	Nevoid basal cell carcinoma syndrome/Gorlin syndrome	5684	PTCH1
<input type="checkbox"/>	Legius syndrome	5724	SPRED1	<input type="checkbox"/>	Tuberous sclerosis complex	5904	TSC1, TSC2
<input type="checkbox"/>	Li-Fraumeni syndrome	2866	TP53	<input type="checkbox"/>	von Hippel-Lindau disease	2606	VHL
<input type="checkbox"/>	Neurofibromatosis 1	5704	NF1				

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"/>	CFTR gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Telomere-related pulmonary fibrosis	8140	TERT, TERC
				Respiratory Distress Syndrome			
<input type="checkbox"/>				<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)

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Electronic ordering is also available via AmbryPort®. Please visit <https://portal.ambrygen.com/login> to log in to the online portal.

Hereditary Cancer Multi-Gene Tests

For current hereditary cancer panel gene content, please visit www.ambrygen.com/providers/oncology/test-menu (linked to QR code below).



Scan for current hereditary cancer panel gene content

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.

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.

Testing on buccal swab samples is available for hereditary cancer testing, ExomeNext, chromosomal microarray, epilepsy and neurodevelopmental disorder panels, fragile X syndrome, hereditary neuropathy (familial transthyretin amyloidosis), and HHT next. 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.

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).

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.