

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 <input type="checkbox"/> F <input type="checkbox"/> M
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:			Gender (optional) <input type="checkbox"/> Man <input type="checkbox"/> Woman <input type="checkbox"/> Nonbinary <input type="checkbox"/> Self-described
Address		State/Province	Country
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	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:
BILLING			
<input type="checkbox"/> INSURANCE BILLING (Include copy of both sides of insurance card)			
<input type="checkbox"/> INSTITUTIONAL BILLING			
Facility Name	<input type="checkbox"/> Send invoice to facility address above		
Address			
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. <input type="checkbox"/> 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:
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:

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: _____
See instructions on the Supplemental Information Page
 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
	<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® testing.

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)			
<input type="checkbox"/> <i>BRCA1/2</i> test				Polyposis test: <input type="checkbox"/> <i>APC/MUTYH</i>			
For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (Lynch)				<input type="checkbox"/> Other: _____			
Lynch Syndrome test: <input type="checkbox"/> <i>MLH1, MSH2, MSH6, PMS2, EPCAM</i>				<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)							
<input type="checkbox"/> +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>CHEK2</i>	9016		<input type="checkbox"/>	<i>MSH2</i> inversion	2226	Lynch syndrome
<input type="checkbox"/>	<i>DICER1</i>	5260		<input type="checkbox"/>	<i>MSH6</i>	8512	Lynch syndrome
<input type="checkbox"/>	<i>PALB2</i>	2366		<input type="checkbox"/>	<i>MUTYH</i>	4661	<i>MUTYH</i> -associated polyposis
<input type="checkbox"/>	<i>PTEN</i>	2106	<i>PTEN</i> -related disorders (including Cowden syndrome)	<input type="checkbox"/>	<i>PMS2</i>	4646	Lynch syndrome
<input type="checkbox"/>	<i>TP53</i>	2866	Li-Fraumeni syndrome	<input type="checkbox"/>	<i>STK11</i>	2766	Peutz-Jeghers syndrome
Endocrine Tumors				Genitourinary Cancer			
<input type="checkbox"/>	<i>MEN1</i>	2646	Multiple endocrine neoplasia type 1	<input type="checkbox"/>	<i>BAP1</i>	9044	
<input type="checkbox"/>	<i>RET</i> gene sequence	2680	Multiple endocrine neoplasia type 2	<input type="checkbox"/>	<i>FH</i>	6301	Hereditary leiomyomatosis and renal cell cancer
Gastrointestinal Cancer				<input type="checkbox"/>	<i>FLCN</i>	5921	Birt-Hogg-Dubé syndrome
<input type="checkbox"/>	<i>APC</i>	3040	Familial adenomatous polyposis	<input type="checkbox"/>	<i>VHL</i>	2606	Von-Hippel Lindau disease
<input type="checkbox"/>	<i>APC</i> and <i>MUTYH</i> concurrent	8726	Adenomatous polyposis	<input type="checkbox"/>	<i>TSC1</i> and <i>TSC2</i>	5904	Tuberous sclerosis complex
<input type="checkbox"/>	<i>BMPRI1</i> and <i>SMAD4</i> concurrent	8604	Juvenile polyposis syndrome	Skin Cancer/Melanoma			
<input type="checkbox"/>	<i>CDH1</i>	4726	Hereditary diffuse gastric cancer	<input type="checkbox"/>	<i>CDKN2A</i> and <i>CDK4</i> concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
<input type="checkbox"/>	<i>EPCAM del/dup</i>	8519	Lynch syndrome	<input type="checkbox"/>	<i>PTCH1</i>	5684	Gorlin syndrome
<input type="checkbox"/>	Lynch syndrome (concurrent)	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup</i>	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

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.

Previously Reported Variants*:

Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____

Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____

Gene: _____ Variant (c. and/or p.): _____ Testing Lab: _____ Ambry ID: _____

*See instructions for reporting of Previously Reported Variants on 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)
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Exome
REQUIRED: Select a Primary Test Order

<input type="checkbox"/>	ExomeNext® <input type="checkbox"/> Proband only <input type="checkbox"/> Duo <input type="checkbox"/> Trio	9900	Exome sequencing	<input type="checkbox"/>	ExomeNext-Rapid®	9999-R	Rapid trio exome sequencing including a defined list of established disease-causing variants in the mitochondrial DNA (mtDNA)
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ExomeNext Supplemental Test Options (Primary test order required. See descriptions for details.)

<input type="checkbox"/>	ACMG Secondary Findings* <input type="checkbox"/> Decline	9920	Analysis of genes included in the ACMG Recommended List of secondary findings. Secondary findings results are available free of charge for the proband and each family member who is fully sequenced as part of the duo/trio.	<input type="checkbox"/>	ExomeReveal® RNA Analysis	9990	RNA analysis is available with all ExomeNext orders except for ExomeNext-Rapid. EDTA and PAX-gene RNA tubes are required.
				<input type="checkbox"/>	Mito DNA	9900-M	Analysis of a defined list of established disease-causing variants in the mitochondrial DNA (mtDNA)

* Secondary Findings: Check "decline" to opt-out of the ACMG Recommended List of secondary findings. If left unchecked, secondary findings will be reported.

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	BMPRIA, 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				
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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			
<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"/>	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				

KNOWN VARIANT ANALYSIS (Please include a copy of relative's report)

Gene(s): _____ Variant(s) (c. and/or p.): _____ Relative Name: _____

Relationship to Relative: _____ Accession # (if tested at Ambry): _____

Positive control sample: will be provided already at Ambry not available

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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.

Known variant analysis should be accompanied by a copy of the original testing report or internal Ambry testing information (internal Ambry ID, Name/DOB). Please review information about positive controls and other specifics at ambrygen.com/knownvariantanalysis.

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. Reflex testing is no longer available for Oncology orders.

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.

Previously Reported Variants

ExomeNext® report comments about the presence or absence of a variant previously reported **in the patient** require the "Previously Reported 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

Previously reported variant requests without an internal Ambry reference ID or positive lab report will not receive a variant-specific report comment.

Variant-specific report comments about the presence or absence of a variant previously reported **in a family member** are not included in ExomeNext or Neurology panel reports.