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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							
<b>REQUIRED: Select a Primary Test Order</b>							
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				Skin Cancer/Melanoma			
<input type="checkbox"/>	<i>APC</i>	3040	Familial adenomatous polyposis	<input type="checkbox"/>	<i>CDKN2A</i> and <i>CDK4</i> concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
<input type="checkbox"/>	<i>APC</i> and <i>MUTYH</i> concurrent	8726	Adenomatous polyposis	<input type="checkbox"/>	<i>PTCH1</i>	5684	Gorlin syndrome
<input type="checkbox"/>	<i>BMPRI1</i> and <i>SMAD4</i> concurrent	8604	Juvenile polyposis syndrome	Other Hereditary Cancer Testing			
<input type="checkbox"/>	<i>CDH1</i>	4726	Hereditary diffuse gastric cancer	<input type="checkbox"/>	<i>NF1</i>	5704	Neurofibromatosis type 1
<input type="checkbox"/>	<i>EPCAM del/dup</i>	8519	Lynch syndrome	<input type="checkbox"/>	<i>NF2</i>	9024	Neurofibromatosis type 2
<input type="checkbox"/>	Lynch syndrome (concurrent)	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup</i>	<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 <a href="http://ambrygen.com/hereditary-cancer-single-gene-tests">ambrygen.com/hereditary-cancer-single-gene-tests</a> 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
<b>CARDIOLOGY</b>							
<b>Comprehensive Cardiovascular Panels</b>				<b>Familial Hypercholesterolemia</b>			
<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, TAA, HHT, Noonan, and lipidemias. Required: completed CustomNext-Cardio supplemental form. <a href="http://ambrygen.com/forms">ambrygen.com/forms</a>	<input type="checkbox"/> Check this box if you would like to have the <i>SLC10B1</i> c.521T>C polymorphism reported with FHNext, which has been associated in medical literature with statin-induced myopathies			
<b>Arrhythmia Panels</b>				<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	<b>Aneurysms and Related Disorders</b>			
<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
<b>Cardiomyopathy Panels</b>				<input type="checkbox"/>	Marfan reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext
<input type="checkbox"/>	HCMNext®	8936	30 genes for hypertrophic cardiomyopathy	<b>Hereditary Hemorrhagic Telangiectasia (HHT)</b>			
<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	<b>Noonan Syndrome</b>			
<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	<b>Other</b>			
				<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 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"/>	Peutz-Jeghers syndrome	2766 STK11

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

NEUROLOGY	
<input type="checkbox"/>	<b>Opt-in to Reporting of Variants of Unknown Significance (VUS)</b> 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"/>	<b>Parental samples provided for cosegregation</b> 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"/>	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: <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](http://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](http://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](http://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](http://ambrygen.com/knownvariantanalysis).

### 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.