

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			
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"/> Asian <input type="checkbox"/> Ashkenazi Jewish <input type="checkbox"/> Black/African American <input type="checkbox"/> White <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"/> Unknown <input type="checkbox"/> Other:			
Address		City	State      Zip
Phone		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: <input type="checkbox"/> Phlebotomy draw** <input type="checkbox"/> Send saliva kit to patient   <input type="checkbox"/> 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
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
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)	
<b>Patient Acknowledgement:</b> 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. <b>For patient payment by credit card:</b> 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.			
<b>For NY Residents:</b> <input type="checkbox"/> By checking this box, I agree that Ambry Genetics will retain my sample for 6 months after the testing above has been completed. By not checking this box, I understand that under New York State law, Ambry Genetics must discard my sample after the longer of (a) testing completion and (b) 60 days after the Date of Collection above.			
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**

<b>Birth and Neonatal History</b> <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: _____	<b>Developmental History</b> <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
<b>Seizure History</b> <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: _____	<b>Cardiac History</b> <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 _____
<b>Pulmonology History</b> <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: _____	<b>Other History</b> <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](http://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: \_\_\_\_\_

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

## Comprehensive Test Requisition Form - Page 3 of 6

If this TRF is sent to Ambyr 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.

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			
<b>REQUIRED: Select a Primary Test Order</b>		<b>Select an Optional Supplemental Test</b> (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/MUTYH</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^^
		Notes: _____	
<b>Other Supplemental Test Options (Select if applicable)</b>		<input type="checkbox"/> MelanomaNext®	8849 9 gene melanoma test
<input type="checkbox"/> +RNAinsight® (Not available with BRCAplus, pancreatitis panel, or STAT orders; PAXgene® tube required for RNA)		<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 <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): _____		

## Comprehensive Test Requisition Form - Page 4 of 6

Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
<b>CARDIOLOGY</b>							
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. <a href="http://ambrygen.com/forms">ambrygen.com/forms</a>	<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			
<b>CLINICAL GENOMICS</b>							
<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"/>	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"/>	ExomeNext®-Proband	9993	Proband only exome sequencing	<input type="checkbox"/>	ExomeNext-Rapid®	9999R	
<input type="checkbox"/>	ExomeNext-Proband plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing	Must be ordered through AP* *AP is AmbyrPort®, our online portal <a href="http://ambrygen.com/ap">ambrygen.com/ap</a>			
<input type="checkbox"/>	ExomeNext-Duo	9991	Duo exome sequencing	If ordering ExomeNext/ExomeNext-Rapid, please complete: <b>Secondary Findings Report:</b> 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"/>	ExomeNext-Duo plus mtDNA	9992	Duo exome sequencing	<input type="checkbox"/> Opt-out: I choose to decline the ACMG Recommended List of secondary findings			
<b>ENDOCRINOLOGY</b>							
<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
<b>GASTROENTEROLOGY</b>							
<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>
<b>HEMATOLOGY/ONCOLOGY</b>							
<input type="checkbox"/>	DBANext	8550	11 genes for Diamond-Blackfan anemia	<input type="checkbox"/>	Peutz-Jeghers syndrome	2766	<i>STK11</i>
<input type="checkbox"/>	DCNext	8161	7 genes for dyskeratosis congenita	<input type="checkbox"/>	Shwachman-Diamond syndrome	1440	<i>SBDS</i>
<b>MULTIPLE CONGENITAL ANOMALIES</b>							
<input type="checkbox"/>	NoonanNext™	8402	18 genes for Noonan syndrome and RASopathies				

## Comprehensive Test Requisition Form - Page 5 of 6

**NEUROLOGY**

**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	<i>PTEN</i>
<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	<i>FMR1</i> repeat expansion analysis and methylation studies
<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	Neurocutaneous/Neuro-Oncology Disorders			
Hereditary Neuropathy				<input type="checkbox"/>	Ataxia-telangiectasia	9014	<i>ATM</i>
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	<i>TTR</i>	<input type="checkbox"/>	BrainTumorNext®	8847	29 genes for brain tumors
Migraine				<input type="checkbox"/>	HHTNext®	8672	<i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>
<input type="checkbox"/>	Familial hemiplegic migraine	6866	<i>ATP1A2, CACNA1A, PRRT2, SCN1A</i> <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Legius syndrome	5724	<i>SPRED1</i>
<b>Note:</b> Gene lists for EpilepsyNext-Expanded are updated annually 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 <a href="http://ambrygen.com">ambrygen.com</a>				<input type="checkbox"/>	Li-Fraumeni syndrome	2866	<i>TP53</i>
<b>For Reflex or Concurrent Testing:</b>				<input type="checkbox"/>	Neurofibromatosis 1	5704	<i>NF1</i>
Test 1: _____ <input type="checkbox"/> Reflex to _____ Test 2: _____ <input type="checkbox"/> Concurrent with _____				<input type="checkbox"/>	Neurofibromatosis 2	9024	<i>NF2</i>
				<input type="checkbox"/>	Nevoid basal cell carcinoma syndrome/ Gorlin syndrome	5684	<i>PTCH1</i>
				<input type="checkbox"/>	Tuberous sclerosis complex	5904	<i>TSC1, TSC2</i>
				<input type="checkbox"/>	von Hippel-Lindau disease	2606	<i>VHL</i>

**PULMONOLOGY**

Order	Test Name	Test Code	Description	Order	Test Name	Test Code	Description
Congenital Central Hypoventilation Syndrome				Primary Ciliary Dyskinesia			
<input type="checkbox"/>	Congenital central hypoventilation syndrome	1580	<i>PHOX2B</i> 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	<i>CFTR</i> deltaF508 mutation analysis with reflex to <i>CFTR</i> gene sequence and deletion/duplication <input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Telomere-related pulmonary fibrosis	8140	<i>TERT, TERC</i>
<input type="checkbox"/>	<i>CFTR</i> gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	Respiratory Distress Syndrome			
<input type="checkbox"/>				<input type="checkbox"/>	Surfactant dysfunction (respiratory distress syndrome)	8100	<i>ABCA3, SFTPB, SFTPC</i> gene sequence

**VASCULAR**

<input type="checkbox"/>	HHTNext®	8672	<i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>	<input type="checkbox"/>	TAADNext®	8789	35 genes for thoracic aortic aneurysms
<input type="checkbox"/>	Marfan syndrome reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext				

**SPECIFIC SITE ANALYSIS (Please include a copy of relative's report)**

Gene(s): \_\_\_\_\_ Mutation(s): \_\_\_\_\_ Relative Name: \_\_\_\_\_  
 Relationship to Relative: \_\_\_\_\_ Accession # (if tested at Ambry): \_\_\_\_\_  
 Positive control sample:  will be provided  already at Ambry  not available

**FOR PRENATAL SPECIMENS, POC OR CORD BLOOD: MATERNAL CELL CONTAMINATION ANALYSIS REQUIRED**  
 Both maternal and fetal specimens are required.

1260 MCC for fetal specimen or cord blood  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 <sup>®</sup> (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 <sup>™</sup> (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 <sup>™</sup> (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>
BRCAplus <sup>®</sup> (8 genes)	8836	<i>ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, TP53</i>
CancerNext <sup>®</sup> (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 <sup>®</sup> (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, MITF, 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 <sup>®</sup> (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 <sup>®</sup> (up to 91 genes) Required: complete CustomNext-Cancer supplemental form. <a href="http://ambrygen.com/forms">ambrygen.com/forms</a>	9510	<i>AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, BMPR1A, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CFTR, CHEK2, CPA1, CTNNA1, CTSC, DICER1, EGFR, EGLN1, EPCAM, FAM175A(ABRAXAS1)<sup>†</sup>, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MLH3<sup>†</sup>, MRE11A<sup>†</sup>, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PALLD<sup>†</sup>, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50<sup>†</sup>, RAD51C, RAD51D, RB1, RECQL, RET, RINT1<sup>†</sup>, RPS20<sup>†</sup>, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, STK11, SUFU, TERT<sup>†</sup>, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2</i>  For Medicare Patients: At a minimum, the following core genes must be included in the panel to ensure Medicare coverage: <i>APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, TP53</i> .
HBOC	8838	<i>BRCA1, BRCA2</i>
Lynch syndrome/HNPCC	8517	<i>MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup</i>
MelanomaNext <sup>®</sup> (9 genes)	8849	<i>BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, TP53</i>
PancNext <sup>®</sup> (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, CTSC</i>
PGLNext <sup>®</sup> (14 genes)	5504	<i>EGLN1, FH, KIF1B, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL</i>
ProstateNext <sup>®</sup> (14 genes)	8845	<i>ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, RAD51D, TP53</i>
RenalNext <sup>®</sup> (20 genes)	5900	<i>BAP1, CHEK2, EPCAM, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL</i>

† Limited evidence gene

**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.

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 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.