

COMPLETE ENTIRE FORM AND SUBMIT PEDIGREE/CLINIC NOTES TO AVOID DELAYS

Aliso Viejo, CA 92656 USA
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PATIENT INFORMATION						
Name (Last, First, MI)		Date of Birth (MM/DD/YY)	Date of Death (if applicable)	Phone Number/Email		
Address	City	State	Zip	Biological Sex <input type="checkbox"/> F <input type="checkbox"/> M	Ethnicity: <input type="checkbox"/> African American <input type="checkbox"/> Asian <input type="checkbox"/> Caucasian <input type="checkbox"/> Hispanic <input type="checkbox"/> Jewish (Ashkenazi) <input type="checkbox"/> Portuguese <input type="checkbox"/> Other:	
SPECIMEN INFORMATION* (For phlebotomy service, select all services you are requesting)						
Type(s) <input type="checkbox"/> Blood (EDTA preferred) <input type="checkbox"/> Saliva <input type="checkbox"/> Buccal Swab* <input type="checkbox"/> DNA <input type="checkbox"/> Cord Blood** <input type="checkbox"/> Other**:				<input type="checkbox"/> Personal history of allogenic bone marrow or peripheral stem cell transplant		
Collection Date	Specimen ID			Medical Record #		
<p><i>*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.</i></p> <p><i>**If submitting Cord Blood or a fetal specimen, please see bottom of page 5 for Maternal Cell Contamination sample submission test codes.</i></p> <p><i>*Only for Fragile X syndrome and chromosomal microarray</i></p>						
Phlebotomy Services Request: <input type="checkbox"/> Phlebotomy draw <input type="checkbox"/> Insurance preverification first <input type="checkbox"/> Send kit to patient* *As the patient's clinician, I am unaware of any potential for complication or difficulty in drawing blood for the listed patient(s). I understand that the phlebotomist has full authority to refuse to draw any patient if the safety of the phlebotomist and/or patient(s) are in question.						
INDICATION(S) FOR TESTING						
ICD-10 code(s):						
Will patient management be changed depending on the test results? <input type="checkbox"/> Yes <input type="checkbox"/> No						
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 Ambyr Genetics to facilitate the provision of pre-test genetic counseling services by a third party service, Informed DNA (unless otherwise noted), as required by the patient's insurance provider (unless this box is checked <input type="checkbox"/>). Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the attached letter of medical necessity.						
Signature Required for Processing Medical Professional Signature:					Date:	
INSURANCE BILLING (Include copy of both sides of insurance card)			INSTITUTIONAL BILLING			
Patient Relation to Policy Holder? <input type="checkbox"/> Self <input type="checkbox"/> Spouse <input type="checkbox"/> Child		Name and DOB of Policy Holder (if not self)		Facility Name <input type="checkbox"/> Send invoice to facility address above		
Insurance Company	Policy #	HMO Auth #	Address			
Out Of Pocket: We will start testing immediately, unless you check the box below. We will attempt to contact you if your estimated out-of-pocket costs are > USD \$100 <input type="checkbox"/> Do not start testing until I approve payment terms regarding estimated out-of-pocket costs Patient agrees to contact regarding out-of-pocket amount by: <input type="checkbox"/> Email <input type="checkbox"/> Phone (includes texts) - confirm mobile # _____			Contact Name			
			Phone Number	E-mail/Fax		
Special Billing Notes:			<input type="checkbox"/> PATIENT PAYMENT		<input type="checkbox"/> Check (Payable to Ambyr 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 Ambyr Genetics Corporation (Ambyr), authorize Ambyr 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 Ambyr money received from my health insurance company. For patient payment by credit card: I hereby authorize Ambyr Genetics Corporation to bill my credit card as indicated above. In order to expedite consideration for eligibility for Ambyr'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 Ambyr Genetics Corporation to verify the above information for the sole purpose of assessing financial need, including the right to seek supporting documentation.						
FOR NY RESIDENTS: <input type="checkbox"/> I am a New York resident and I give Ambyr Genetics permission to store my sample for longer than 60 days. NOTE: If left blank, consent is interpreted as "NO".						
Signature Required For Insurance/Self-Pay Patients and NY Sample Storage Consent:					Date:	

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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

Cancer/Tumor	Age at Dx	Pathology and Other Info
Brain		
Breast		Type: ER <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk PR <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk HER2/neu <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk
2nd primary breast		Type: ER <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk PR <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk HER2/neu <input type="checkbox"/> (+) <input type="checkbox"/> (-) <input type="checkbox"/> unk
Colorectal		Location: _____
Ovarian		<input type="checkbox"/> Fallopian tube <input type="checkbox"/> Primary peritoneal
Melanoma/skin		
Prostate		Gleason Score: _____ Metastatic: <input type="checkbox"/> Yes <input type="checkbox"/> No
Uterine		
Hematologic*		Type: <input type="checkbox"/> Allogenic bone marrow or peripheral stem cell transplant*
Other Cancer		Type: _____
GI polyps		<input type="checkbox"/> Adenomatous <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+ 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)

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

FAMILY HISTORY (Completion of this section is required for orders including parental samples)

Mother - Name: _____ DOB: _____ unaffected affected, list symptoms/dx: _____ Dx age: _____
 Father - Name: _____ DOB: _____ unaffected affected, list symptoms/dx: _____ Dx age: _____

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

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Please check the box next to the test(s) being ordered below.

All tests include gene sequence and deletion/duplication analyses unless otherwise indicated.

If this TRF is sent to Ambry without or ahead of the sample, it will be treated as a preverification. If test ordered is different than the test preverified, we will honor what is on the TRF order form with the sample.

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

CANCER							
Multi-Gene Orders							
Select the indication for testing:				<input type="checkbox"/>	BrainTumorNext	8847	27 gene brain tumor test
<input type="checkbox"/> Hereditary polyposis ¹				<input type="checkbox"/>	BRCAPlus	8836	8 gene breast cancer test
<input type="checkbox"/> Lynch syndrome/HNPCC ²				<input type="checkbox"/>	BreastNext	8820	17 gene breast cancer test
<input type="checkbox"/> Hereditary breast and ovarian cancer ³				<input type="checkbox"/>	CancerNext	8824	34 gene cancer test
<input type="checkbox"/> Testing is clinically indicated for other gene(s):				<input type="checkbox"/>	CancerNext-Expanded	8874	67 gene cancer test
<input type="checkbox"/> None of the above				<input type="checkbox"/>	ColoNext	8822	17 gene colorectal cancer test
To complete your multi-gene order, please select a test option to the right. (See supplemental information on page 6 for details).				<input type="checkbox"/>	CustomNext-Cancer	9510	up to 81 gene custom test*
¹ APC/MUTYH ² MLH1, MSH2, MSH6, PMS2, EPCAM ³ BRCA1/2				<input type="checkbox"/>	GYNplus	8835	13 gene ovarian/uterine cancer test
* Required: completed CustomNext-Cancer supplemental form. ambrygen.com/forms				<input type="checkbox"/>	MelanomaNext	8849	8 gene melanoma test
				<input type="checkbox"/>	OvaNext	8830	25 gene ovarian/breast/uterine cancer test
				<input type="checkbox"/>	PancNext	8042	13 gene pancreatic cancer test
				<input type="checkbox"/>	Pancreatitis panel	8022	6 gene pancreatitis test
				<input type="checkbox"/>	PGLNext	5504	12 gene PGL/PCC test
				<input type="checkbox"/>	ProstateNext	8845	14 gene prostate cancer test
				<input type="checkbox"/>	RenalNext	5900	19 gene renal cancer test
Single Syndrome Orders							
Check to order	Test Name	Test Code	Description	Check to order	Test Name	Test Code	Description
Breast and/or Ovarian Cancer				Gastrointestinal Cancer (Cont.)			
<input type="checkbox"/>	ATM	9014	Ataxia-telangiectasia	<input type="checkbox"/>	MLH1	8508	Lynch syndrome
<input type="checkbox"/>	BRCA1/2	8838	Hereditary breast and ovarian cancer	<input type="checkbox"/>	MSH2 + EPCAM del/dup	8510	Includes MSH2 inversion
<input type="checkbox"/>	BRCA1/2 Ashkenazi Jewish 3-site mutation panel	5892		<input type="checkbox"/>	MSH2 inversion	2226	Lynch syndrome
<input type="checkbox"/>	BRCA1/2 Ashkenazi Jewish 3-site mutation panel with reflex to BRCA1/2 analysis if negative	5894		<input type="checkbox"/>	MSH6	8512	Lynch syndrome
<input type="checkbox"/>	CHEK2	9016		<input type="checkbox"/>	MUTYH	4661	MUTYH-associated polyposis
<input type="checkbox"/>	DICER1	5260		<input type="checkbox"/>	PMS2	4646	Lynch syndrome
<input type="checkbox"/>	PALB2	2366		<input type="checkbox"/>	STK11	2766	Peutz-Jeghers syndrome
<input type="checkbox"/>	PTEN	2106	PTEN-related disorders (including Cowden syndrome)	Genitourinary Cancer			
<input type="checkbox"/>	TP53	2866	Li-Fraumeni syndrome	<input type="checkbox"/>	BAP1	9044	
Endocrine Tumors				<input type="checkbox"/>	FH	6301	Hereditary leiomyomatosis and renal cell cancer
<input type="checkbox"/>	MEN1	2646	Multiple endocrine neoplasia type 1	<input type="checkbox"/>	FLCN	5921	Birt-Hogg-Dubé syndrome
<input type="checkbox"/>	RET gene sequence	2680	Multiple endocrine neoplasia type 2	<input type="checkbox"/>	VHL	2606	Von-Hippel Lindau disease
Gastrointestinal Cancer				<input type="checkbox"/>	TSC1 and TSC2	5904	Tuberous sclerosis complex
<input type="checkbox"/>	APC	3040	Familial adenomatous polyposis	Skin Cancer/Melanoma			
<input type="checkbox"/>	APC and MUTYH concurrent	8726	Adenomatous polyposis	<input type="checkbox"/>	CDKN2A and CDK4 concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
<input type="checkbox"/>	BMPRIA and SMAD4 concurrent	8604	Juvenile polyposis syndrome	<input type="checkbox"/>	PTCH1	5684	Gorlin syndrome
<input type="checkbox"/>	CDH1	4726	Hereditary diffuse gastric cancer	Other Hereditary Cancer Testing			
<input type="checkbox"/>	EPCAM del/dup	8519	Lynch syndrome	<input type="checkbox"/>	NF1	5704	Neurofibromatosis type 1
<input type="checkbox"/>	Lynch syndrome (concurrent)	8517	MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup	<input type="checkbox"/>	NF2	9024	Neurofibromatosis type 2
<input type="checkbox"/>	Lynch syndrome (sequential)	8515	Step 1: MLH1, MSH2, and MSH6 + EPCAM del/dup; Step 2: PMS2	<input type="checkbox"/>	RB1	5426	Hereditary retinoblastoma
				<input type="checkbox"/>	SMARCB1	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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CARDIOLOGY							
Check to order	Test Name	Test Code	Description	Check to order	Test Name	Test Code	Description
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</i> , <i>LDLR</i> , <i>LDLRAP1</i> , <i>PCSK9</i>)
<input type="checkbox"/>	CustomNext- <i>Cardio</i>	9520	Up to 167 genes related to hereditary cardiomyopathies, arrhythmias, TAAD, HHT, Noonan, and lipidemias. Required: completed CustomNext- <i>Cardio</i> 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			
<input type="checkbox"/>				<input type="checkbox"/>	FCSNext (Familial Chylomicronemia Syndrome)	8920	<i>APOA5</i> , <i>APOC2</i> , <i>GPIHBP1</i> , <i>LMF1</i> , <i>LPL</i>
<input type="checkbox"/>				<input type="checkbox"/>	Sitosterolemia	8930	<i>ABCG5</i> , <i>ABCG8</i>
Arrhythmia Panels				Aneurysms and Related Disorders			
<input type="checkbox"/>	LongQTNext	8890	17 genes for long QT, Brugada and short QT syndromes	<input type="checkbox"/>	TAADNext	8789	35 genes for thoracic aortic aneurysms/dissections, Marfan syndrome, Ehlers-Danlos and related disorders
<input type="checkbox"/>	RhythmNext	8900	42 genes for long QT syndrome, Brugada and short QT syndromes, CPVT and ARVC	<input type="checkbox"/>	Marfan reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext
<input type="checkbox"/>	CPVNext	8902	4 genes for catecholaminergic polymorphic ventricular tachycardia	Hereditary Hemorrhagic Telangiectasia (HHT)			
Cardiomyopathy Panels				<input type="checkbox"/>	HHTNext	8672	<i>ACVRL1</i> , <i>ENG</i> , <i>EPHB4</i> , <i>GDF2</i> , <i>RASA1</i> , <i>SMAD4</i>
<input type="checkbox"/>	HCMNext	8936	30 genes for hypertrophic cardiomyopathy	Noonan Syndrome			
<input type="checkbox"/>	HCMNext Reflex	8883	<i>MYBPC3</i> , <i>MYH7</i> reflex to HCMNext	<input type="checkbox"/>	NoonanNext	8402	18 genes for RASopathies
<input type="checkbox"/>	DCMNext	8884	37 genes for dilated cardiomyopathy	Other			
<input type="checkbox"/>	CMNext	8887	56 genes for hereditary cardiomyopathy	<input type="checkbox"/>	Transthyretin amyloidosis	1560	<i>TTR</i>
<input type="checkbox"/>	ARVCNext	8904	11 genes for arrhythmogenic right ventricular cardiomyopathy	<input type="checkbox"/>	SNP Array*	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)
CLINICAL GENOMICS							
<input type="checkbox"/>	Karyotype	3660	Chromosome analysis (requires green-top sodium-heparin tube)	<input type="checkbox"/>	ExomeNext- <i>Trio</i>	9995	Trio exome sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	Karyotype, rule out mosaic	3662	Chromosome analysis (requires green-top sodium-heparin tube)	<input type="checkbox"/>	ExomeNext- <i>Trio</i> plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing <input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)	<input type="checkbox"/>	ExomeNext- <i>Rapid</i>	9999R	<input type="checkbox"/> Opt-out of Candidate (novel) Genetic Etiologies
<input type="checkbox"/>	Familial targeted microarray	5495	Paid option. Only available following SNP Array (5490) completed at Ambry. Incidental findings unrelated to the variant(s) detected in the proband, will NOT be reported. Name of proband tested at Ambry: _____	<input type="checkbox"/>	ExomeNext- <i>Select</i>	9500	Up to 500 gene custom exome sequencing test
<input type="checkbox"/>	ExomeNext- <i>Proband</i>	9993	Proband only exome sequencing	Must be ordered through AP* *AP is AmbryPort, our online portal ambrygen.com/ap			
<input type="checkbox"/>	ExomeNext- <i>Proband</i> plus mtDNA	9994	Proband only 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. Secondary findings are not available for ExomeNext- <i>Select</i> orders. <input type="checkbox"/> Opt-out: I choose to decline the ACMG Recommended List of secondary findings			
ENDOCRINOLOGY							
<input type="checkbox"/>	Hereditary leiomyomatosis renal cell carcinoma	6301	<i>FH</i>	<input type="checkbox"/>	Multiple endocrine neoplasia type 2 and familial medullary thyroid cancer (FMTC)	2680	<i>RET</i> gene sequence
<input type="checkbox"/>	Maturity-onset diabetes of the young	8310	<i>HNF1A</i> , <i>HNF4A</i> , <i>HNF1B</i> , <i>GCK</i> , <i>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	12 gene PGL/PCC test
<input type="checkbox"/>				<input type="checkbox"/>	von-Hippel Lindau disease	2606	<i>VHL</i>
GASTROENTEROLOGY							
<input type="checkbox"/>	<i>CFTR</i> gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	<input type="checkbox"/>	Juvenile polyposis syndrome	8604	<i>BMPR1A</i> , <i>SMAD4</i>
<input type="checkbox"/>	Hirschsprung disease (<i>RET</i> -related)	2680	<i>RET</i> gene sequence	<input type="checkbox"/>	Pancreatitis	8022	<i>CASR</i> , <i>CFTR</i> , <i>CPA1</i> , <i>CTRC</i> , <i>PRSS1</i> , <i>SPINK1</i>
<input type="checkbox"/>				<input type="checkbox"/>	Peutz-Jeghers syndrome	2766	<i>STK11</i>
HEMATOLOGY/ONCOLOGY							
<input type="checkbox"/>	DBANext	8550	11 genes for Diamond-Blackfan anemia	<input type="checkbox"/>	Shwachman-Diamond syndrome	1440	<i>SBDS</i>
<input type="checkbox"/>	DCNext	8161	7 genes for dyskeratosis congenita				
MULTIPLE CONGENITAL ANOMALIES							
<input type="checkbox"/>	CHARGE syndrome	2380	<i>CHD7</i>	<input type="checkbox"/>	Noonan syndrome	8402	<i>PTPN11</i> , <i>SOS1</i> , <i>KRAS</i> , <i>RAF1</i>
<input type="checkbox"/>	CdLSNext - Cornelia de Lange syndrome	7040	<i>NIPBL</i> , <i>SMC1A</i> , <i>HDAC8</i> , <i>RAD21</i> , <i>SMC3</i>				

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NEUROLOGY							
Check to order	Test Name	Test Code	Description	Check to order	Test Name	Test Code	Description
Comprehensive Neuro Testing				Neurodevelopmental Disorders			
<input type="checkbox"/>	CustomNext-Neuro	9540	Up to 196 gene custom neurology test. <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	AutismNext	7024	48 genes for syndromic and non-syndromic autism spectrum disorders <input type="checkbox"/> Check if parental samples are included
Epilepsy				<input type="checkbox"/>	Autism, macrocephaly	2106	<i>PTEN</i>
<input type="checkbox"/>	EpiRapid	7033	16 epilepsy genes with treatment associations	<input type="checkbox"/>	Fragile X syndrome	4544	<i>FMR1</i> repeat expansion analysis and methylation studies
<input type="checkbox"/>	EpiRapid reflex to EpilepsyNext	7034	16 epilepsy genes with treatment associations, reflex to 100 genes for epilepsy	<input type="checkbox"/>	IDNext	7027	140 genes for syndromic and non-syndromic intellectual disability <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	EpiFirst-Fever	7011	13 genes for febrile seizures <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Neurodevelopment-Expanded	7028	196 genes for intellectual disability, autism spectrum disorders, and epilepsy <input type="checkbox"/> Check if parental samples are included
<input type="checkbox"/>	EpiFirst-Focal	7017	11 gene for non-lesional focal epilepsy <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Rett syndrome	2026	<i>MECP2</i>
<input type="checkbox"/>	EpiFirst-IS	7013	17 genes for infantile spasms <input type="checkbox"/> Check if parental samples are included	Neurocutaneous/Neuro-Oncology Disorders			
<input type="checkbox"/>	EpilepsyNext	7019	100 genes for epilepsy <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Ataxia-telangiectasia	9014	<i>ATM</i>
Hereditary Neuropathy				<input type="checkbox"/>	BrainTumorNext	8847	27 genes for brain tumors
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	<i>TTR</i>	<input type="checkbox"/>	HHTNext	8672	<i>ACVRL1, ENG, SMAD4, GDF2, RASA1</i>
Migraine				<input type="checkbox"/>	Legius syndrome	5724	<i>SPRED1</i>
<input type="checkbox"/>	Familial hemiplegic migraine	7035	<i>ATP1A2, CACNA1A, PRRT2, SCN1A</i> <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	Li-Fraumeni syndrome	2866	<i>TP53</i>
				<input type="checkbox"/>	Neurofibromatosis 1	5704	<i>NF1</i>
				<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							
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"/>	Surfactant dysfunction (respiratory distress syndrome)	8100	<i>ABCA3, SFTPB, SFTPC</i> gene sequence

RHEUMATOLOGY							
<input type="checkbox"/>	Familial Mediterranean fever	5000	<i>MEFV</i>				

VASCULAR							
<input type="checkbox"/>	EDS IV reflex to TAADNext	8791	<i>COL3A1</i> reflex to TAADNext	<input type="checkbox"/>	Marfan syndrome (MFS)	8781	<i>FBN1</i>
<input type="checkbox"/>	Ehlers-Danlos type IV, vascular type	8790	<i>COL3A1</i>	<input type="checkbox"/>	MFS reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext
<input type="checkbox"/>	HHTNext	8672	<i>ACVRL1, ENG, SMAD4, GDF2, RASA1</i>	<input type="checkbox"/>	TAADNext	8789	22 genes for thoracic aortic aneurysms

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 OR CORD BLOOD: MATERNAL CELL CONTAMINATION (Both test codes required for fetal specimens)

1260 MCC for fetal specimen or cord blood (run concurrently with test) 1262 MCC Reference for maternal blood sample (No Charge)

Comprehensive TRF - Hereditary Cancer Testing Supplemental Information - Page 6 of 6

Hereditary Cancer Multi-Gene Tests		
Test Name	Test Code	Genes
Adenomatous polyposis	8726	APC, MUTYH
BrainTumorNext (27 genes)	8847	AIP, ALK, APC, CDKN1B, CDKN2A, DICER1, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL
BRCAPlus (8 genes)	8836	ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, TP53
BreastNext (17 genes)	8820	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MRE11A, MUTYH, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, TP53
CancerNext (34 genes)	8824	APC, ATM, BARD1, BRCA1, BRCA2, BRIP1, BMPR1A, CDH1, CDK4, CDKN2A, CHEK2, DICER1, EPCAM, GREM1, HOXB13, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51C, RAD51D, SMAD4, SMARCA4, STK11, TP53
CancerNext-Expanded (67 genes)	8874	AIP, ALK, APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, BMPR1A, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, DICER1, EPCAM, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, MAX, MEN1, MET, MIF, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, PALB2, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2
ColoNext (17 genes)	8822	APC, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
CustomNext-Cancer (up to 81 genes) Required: complete CustomNext-Cancer supplemental form. ambrygen.com/forms	9510	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, EPCAM, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIT, MAX, MEN1, MET, MIF, MLH1, MRE11A, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2
GYNplus (13 genes)	8835	BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53
HBOC	8838	BRCA1, BRCA2
Lynch syndrome/HNPCC	8517	MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup
MelanomaNext (8 genes)	8849	BAP1, BRCA2, CDK4, CDKN2A, MIF, PTEN, RB1, TP53
OvaNext (25 genes)	8830	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53
PancNext (13 genes)	8042	APC, ATM, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, STK11, TP53
Pancreatitis panel (6 genes)	8022	CASR, CFTR, CPA1, PRSS1, SPINK1, CTSC
PGLNext (12 genes)	5504	FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL
ProstateNext (14 genes)	8845	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, RAD51D, TP53
RenalNext (19 genes)	5900	BAP1, EPCAM, FH, FLCN, MET, MIF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL

Updated Ordering Process (as of June 8, 2016)

We have improved the ordering and reporting process for our hereditary cancer panels. This helps confirm that testing for one or more of the following genes is clinically indicated: APC, BRCA1, BRCA2, EPCAM, MLH1, MSH2, MSH6, MUTYH, and PMS2.

If you are ordering a multi-gene test, please first select a clinically indicated condition and complete your order by selecting a multi-gene order.

Please indicate if your patient meets clinical and/or insurance testing criteria, or if the testing is otherwise clinically indicated for one or more of the following conditions:

- Adenomatous polyposis (APC/MUTYH)
- Hereditary breast and ovarian cancer (BRCA1/2)
- Lynch syndrome/HNPCC (MLH1, MSH2, MSH6, PMS2, EPCAM)

If testing is not clinically indicated for your patient for any of the listed options, please either fill in the other gene(s) option or select "none of the above".

To complete your multi-gene order, please select one of the appropriate test options and/or select "other" and enter an appropriate test code(s)/test name(s).

For single gene orders, please select the appropriate test option or enter the gene(s) and/or test name(s), as well as the relevant test code in the single gene orders section.

For additional details about our single gene testing options, please visit ambrygen.com/hereditary-cancer-single-gene-tests.

Example: For a ColoNext multi-gene order, when Lynch testing is clinically indicated for the patient

MULTI-GENE ORDERS				
Select the indication for testing:	<input type="checkbox"/>	BrainTumorNext	8847	27 gene brain tumor test
<input type="checkbox"/> Hereditary polyposis ¹	<input type="checkbox"/>	BRCAPlus	8836	8 gene breast cancer test
<input checked="" type="checkbox"/> Lynch syndrome/HNPCC ²	<input type="checkbox"/>	BreastNext	8820	17 gene breast cancer test
<input type="checkbox"/> Hereditary breast and ovarian cancer ³	<input type="checkbox"/>	CancerNext	8824	34 gene cancer test
<input type="checkbox"/> Testing is clinically indicated for other gene(s):	<input checked="" type="checkbox"/>	CancerNext-Expanded	8874	67 gene cancer test
<input type="checkbox"/> None of the above	<input type="checkbox"/>	ColoNext	8822	17 gene colorectal cancer test
To complete your multi-gene order, please select a test option to the right. (See supplemental information on page 4 for details).	<input type="checkbox"/>	CustomNext-Cancer	9510	up to 81 gene custom test*
	<input type="checkbox"/>	GYNplus	8835	13 gene ovarian/uterine cancer test
	<input type="checkbox"/>	MelanomaNext	8849	8 gene melanoma test
	<input type="checkbox"/>	OvaNext	8830	25 gene ovarian/breast/uterine cancer test
	<input type="checkbox"/>	PancNext	8042	13 gene pancreatic cancer test
	<input type="checkbox"/>	Pancreatitis panel	8022	6 gene pancreatitis test
	<input type="checkbox"/>	PGLNext	5504	12 gene PGL/PCC test
	<input type="checkbox"/>	ProstateNext	8845	14 gene prostate cancer test
	<input type="checkbox"/>	RenalNext	5900	19 gene renal cancer test

¹APC/MUTYH ²MLH1, MSH2, MSH6, PMS2, EPCAM ³BRCA1/2

* Required: completed CustomNext-Cancer supplemental form. ambrygen.com/forms