

COMPLETE ENTIRE FORM TO AVOID DELAYS

PATIENT INFORMATION							
Name (Last, First, MI)				Date of Birth (MM/DD/YY)		Date of Death (if applicable)	Phone Number/Email
Address		City	State/Province	Postal Code	Country	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*		
Type(s) <input type="checkbox"/> Blood (EDTA preferred) <input type="checkbox"/> Saliva <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 #

*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.
** If submitting Cord blood, please see bottom of page 5 for Maternal Cell Contamination sample submission test codes.

INDICATION(S) FOR TESTING
ICD-10 code(s):

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/Province	Postal Code	Country	Phone
Ordering Licensed Provider Name (Last, First)(Code)		NPI#	Phone	Fax/Email

ADDITIONAL RESULTS RECIPIENTS	
Genetic Counsellor or Other Medical Provider Name (Last, First) (Code)	Phone/Fax/Email
Genetic Counsellor or Other Medical Provider Name (Last, First) (Code)	Phone/Fax/Email

CONFIRMATION OF INFORMED CONSENT 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. Furthermore, all information on this TRF is true to the best of my knowledge. My signature applies to the attached letter of medical necessity (unless this box is checked).

<i>Signature Required for Processing</i> Medical Professional Signature:	Date:
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INSTITUTIONAL BILLING (to be completed by institution or health authority providing payment)		
Facility Name	Address (with country)	
Contact Name	Phone Number	E-mail/Fax

PATIENT PAYMENT <input type="checkbox"/> Check (Payable to Ambry Genetics) <input type="checkbox"/> Credit card (Call +1 949-900-5794)
Additional billing comments or other order #s (i.e. POs)

Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. **For patient payment by credit card:** I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above. 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.

<i>Signature Required for Self-Pay Patients and Research Consent Patient or Legal Guardian Signature:</i>	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: _____ # 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: _____ <input type="checkbox"/> HCM <input type="checkbox"/> DCM <input type="checkbox"/> ARVC <input type="checkbox"/> LVNC <input type="checkbox"/> RCM <input type="checkbox"/> Other cardiomyopathy types: _____ History of Arrhythmia <input type="checkbox"/> Y <input type="checkbox"/> N Age at dx: _____ <input type="checkbox"/> Long QT <input type="checkbox"/> Short QT <input type="checkbox"/> Brugada <input type="checkbox"/> CPVT <input type="checkbox"/> ARVC <input type="checkbox"/> Other arrhythmia types: _____ <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: unaffected affected, list symptoms/dx: _____ Dx age: _____
 Father: 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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ORDERING CHECKLIST	
<input type="checkbox"/>	Clinic notes (with pedigree if available)
<input type="checkbox"/>	Clinician & patient signatures

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 Testing							
<input type="checkbox"/>	BrainTumorNext	8847	27 gene brain tumor test	<input type="checkbox"/>	MelanomaNext	8849	8 gene melanoma test
<input type="checkbox"/>	BRCAPlus	8836	8 gene breast cancer test	<input type="checkbox"/>	OvaNext	8830	25 gene ovarian/breast/uterine cancer test
<input type="checkbox"/>	BreastNext	8820	17 gene breast cancer test	<input type="checkbox"/>	PancNext	8042	13 gene pancreatic cancer test
<input type="checkbox"/>	CancerNext	8824	34 gene cancer test	<input type="checkbox"/>	Pancreatitis panel	8022	4 gene pancreatitis test
<input type="checkbox"/>	CancerNext-Expanded	8874	67 gene cancer test	<input type="checkbox"/>	PGLNext	5504	12 gene PGL/PCC test
<input type="checkbox"/>	ColoNext	8822	17 gene colorectal cancer test	<input type="checkbox"/>	ProstateNext	8845	14 gene prostate cancer test
<input type="checkbox"/>	CustomNext-Cancer	9510	Up to 68 gene custom test [*]	<input type="checkbox"/>	RenalNext	5900	19 gene renal cancer test
<input type="checkbox"/>	GYNplus	8835	13 gene ovarian/uterine cancer test				

See Hereditary Cancer Testing Supplemental Information on page 6 for details, such as included genes.

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

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"/>	BMPR1A 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				Cardiomyopathy Panels (Cont.)			
<input type="checkbox"/>	CardioNext with <i>TTN</i>	8911	85 genes for hereditary cardiomyopathies and arrhythmias	<input type="checkbox"/>	CMNext with <i>TTN</i>	8887	55 genes for hereditary cardiomyopathy
<input type="checkbox"/>	CustomNext- <i>Cardio</i>	9520	Up to 106 gene custom hereditary cardiomyopathies and arrhythmias test. Required: completed CustomNext-<i>Cardio</i> supplemental form. ambrygen.com/forms	<input type="checkbox"/>	ARVCNext	8904	9 genes for arrhythmogenic right ventricular cardiomyopathy
Arrhythmia, Long QT, and Brugada Panels				<input type="checkbox"/>	LVNCNext	8906	8 genes for left ventricular non-compaction
<input type="checkbox"/>	RhythmFirst	8888	12 genes for long QT syndrome	Familial Hypercholesterolemia			
<input type="checkbox"/>	RhythmNext	8900	36 genes for inherited arrhythmias	<input type="checkbox"/>	FHNNext	8680	4 genes (<i>APOB</i> , <i>LDLR</i> , <i>LDLRAP1</i> , <i>PCSK9</i>)
<input type="checkbox"/>	RhythmNext Reflex	8901	RhythmFirst reflex to RhythmNext	<input type="checkbox"/> Check this box if you would like to have the <i>SLC01B1</i> c.521T>C polymorphism reported, which has been associated in medical literature with statin-induced myopathies			
<input type="checkbox"/>	CPVTNext	8902	6 genes for catecholaminergic polymorphic ventricular tachycardia	Thoracic Aortic Aneurysms and Related Disorders			
Cardiomyopathy Panels				<input type="checkbox"/>	TAADNext	8789	22 genes for thoracic aortic aneurysms/dissections
<input type="checkbox"/>	HCMFirst	8935	<i>MYBPC3</i> , <i>MYH7</i>	<input type="checkbox"/>	Marfan syndrome	8781	<i>FBN1</i>
<input type="checkbox"/>	HCMNext	8936	27 genes for hypertrophic cardiomyopathy	<input type="checkbox"/>	Marfan reflex to TAADNext	8783	<i>FBN1</i> reflex to TAADNext
<input type="checkbox"/>	HCMNext Reflex	8883	HCMFirst reflex to HCMNext	<input type="checkbox"/>	Ehlers-Danlos vascular type (EDS IV)	8790	<i>COL3A1</i>
<input type="checkbox"/>	DCMNext	8884	36 genes for dilated cardiomyopathy	<input type="checkbox"/>	Ehlers-Danlos reflex to TAADNext	8791	<i>COL3A1</i> reflex to TAADNext
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"/>	Parental 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	*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>HNFA1</i> , <i>HNFA4</i> , <i>HNFB1</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"/>	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>PRSS1</i> , <i>SPINK1</i> , <i>CFTR</i> , <i>CTRC</i>
				<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"/>	DCNNext	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
Dystrophinopathies				<input type="checkbox"/>	Autism, macrocephaly	2106	<i>PTEN</i>
<input type="checkbox"/>	Duchenne/Becker muscular dystrophy	6314	DMD deletion/duplication analysis via MLPA	<input type="checkbox"/>	Fragile X syndrome	4544	<i>FMR1</i> repeat expansion analysis and methylation studies
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"/>	EpiRapid **blood only, no saliva**	7033	16 epilepsy genes with treatment associations	<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"/>	EpiRapid reflex to EpilepsyNext **blood only, no saliva**	7034	16 epilepsy genes with treatment associations, reflex to 100 genes for epilepsy	<input type="checkbox"/>	Rett syndrome	2026	<i>MECP2</i>
<input type="checkbox"/>	EpiFirst-Fever	7011	13 genes for febrile seizures <input type="checkbox"/> Check if parental samples are included	Neurocutaneous/Neuro-Oncology Disorders			
<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"/>	Ataxia-telangiectasia	9014	<i>ATM</i>
<input type="checkbox"/>	EpiFirst-IS	7013	17 genes for infantile spasms <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	BrainTumorNext	8847	27 genes for brain tumors
<input type="checkbox"/>	EpilepsyNext	7019	100 genes for epilepsy <input type="checkbox"/> Check if parental samples are included	<input type="checkbox"/>	HHTNext	8672	<i>ACVRL1, ENG, SMAD4, GDF2, RASA1</i>
Hereditary Neuropathy				<input type="checkbox"/>	Legius syndrome	5724	<i>SPRED1</i>
<input type="checkbox"/>	Familial transthyretin amyloidosis	1560	<i>TTR</i>	<input type="checkbox"/>	Li-Fraumeni syndrome	2866	<i>TP53</i>
Migraine				<input type="checkbox"/>	Neurofibromatosis 1	5704	<i>NF1</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"/>	Neurofibromatosis 2	9024	<i>NF2</i>
PULMONOLOGY				<input type="checkbox"/>	Nevoid basal cell carcinoma syndrome/Gorlin syndrome	5684	<i>PTCH1</i>
Congenital Central Hypoventilation Syndrome				<input type="checkbox"/>	Tuberous sclerosis complex	5904	<i>TSC1, TSC2</i>
<input type="checkbox"/>	Congenital central hypoventilation syndrome	1580	<i>PHOX2B</i> gene sequence	<input type="checkbox"/>	von Hippel-Lindau disease	2606	<i>VHL</i>
Cystic Fibrosis				Primary Ciliary Dyskinesia			
<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"/>	PCDNext	8122	21 genes for primary ciliary dyskinesia <input type="checkbox"/> Report poly T/TG status
<input type="checkbox"/>	<i>CFTR</i> gene sequence and deletion/duplication analysis	1007	<input type="checkbox"/> Report poly T/TG status	Pulmonary Fibrosis			
RHEUMATOLOGY				<input type="checkbox"/>	Telomere-related pulmonary fibrosis	8140	<i>TERT, TERC</i>
<input type="checkbox"/>	Familial Mediterranean fever	5000	<i>MEFV</i>	Respiratory Distress Syndrome			
VASCULAR				<input type="checkbox"/>	Surfactant dysfunction (respiratory distress syndrome)	8100	<i>ABCA3, SFTPB, SFTPC</i> gene sequence
<input type="checkbox"/>	EDS IV reflex to TAADNext	8791	<i>COL3A1</i> reflex to TAADNext	SINGLE SITE ANALYSIS (Please include a copy of relative's report)			
<input type="checkbox"/>	Ehlers-Danlos type IV, vascular type	8790	<i>COL3A1</i>	Gene(s): _____ Mutation(s): _____ Relative Name: _____			
<input type="checkbox"/>	HHTNext	8672	<i>ACVRL1, ENG, SMAD4, GDF2, RASA1</i>	Relationship to Relative: _____ Accession # (if tested at Ambry): _____			
FOR PRENATAL SPECIMENS ONLY: MATERNAL CELL CONTAMINATION (Both test codes required for fetal specimens)				Positive control sample: <input type="checkbox"/> will be provided <input type="checkbox"/> already at Ambry <input type="checkbox"/> not available			
<input type="checkbox"/> 1260 MCC for amniotic fluid culture or CVS (run concurrently with test)				<input type="checkbox"/> 1262 MCC Reference for maternal blood sample (No Charge)			

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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, PRKARIA, 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, PRKARIA, 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 68 genes) Required: complete CustomNext-Cancer supplemental form. ambrygen.com/forms	9510	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, NTHL1, PALB2, PHOX2B, POT1, PMS2, POLD1, POLE, PRKARIA, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, 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 (4 genes)	8022	CFTR, PRSS1, SPINK1, CTRC
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