Ambry Genetics®
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Comprehensive Test Requisition Form - Page 1 of 6

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

PATIENT INFORMATION

Name (Last, First, MI)	Biological Sex ☐F ☐M	Date of Birth (MM/DD/YY)	MRN

Ethnicity: ☐African American ☐Asian ☐Caucasian ☐Hispanic ☐Portuguese ☐Other: | Ashkenazi Jewish ☐Yes ☐No

Address	City	State	Zip

Preferred Method Of Contact ☐Phone ☐Text (requires mobile phone number) ☐Email	Phone	Email

SPECIMEN INFORMATION* (For phlebotomy service, select all services you are requesting)

Type(s) ☐Blood (EDTA preferred) ☐Saliva ☐Buccal Swab^ ☐DNA ☐Cord Blood** ☐Other**: | ☐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.

***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.*

^Only for Fragile X syndrome and chromosomal microarray

Phlebotomy Services Request: ☐Phlebotomy draw ☐Insurance preverification first ☐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? ☐Yes ☐No

PRENATAL SAMPLES ONLY

Sample type: ☐Direct CVS ☐Cultured CVS ☐Cultured amnio ☐POC ☐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)

Patient Relation to Policy Holder? ☐Self ☐Spouse ☐Child	Name and DOB of Policy Holder (if not self)	
Insurance Company	Policy #	HMO Auth #

Out Of Pocket:

We will start testing immediately, unless you check the box below. We will attempt to contact the patient if the estimated out-of-pocket costs are > USD $100

☐ Do not start testing until the patient approves payment terms regarding estimated out-of-pocket costs. By checking this box, I understand that there will be a delay in starting this test until Ambry is able to reach the patient to communicate OOP costs.

Special Billing Notes:

☐ INSTITUTIONAL BILLING

Facility Name ☐ *Send invoice to facility address above*

Address

Contact Name

Phone Number	E-mail/Fax

☐ PATIENT PAYMENT

☐Check (Payable to Ambry Genetics)
☐Credit Card (Call 949-900-5795)

Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. For direct insurance billing: I authorize my insurance benefits to be paid directly to Ambry Genetics Corporation (Ambry), authorize Ambry to release medical information concerning my testing to my insurer, to be my designated representative for purposes of appealing any denial of benefits as needed and to request additional medical records for this purpose. I understand that I am financially responsible for any amounts not covered by my insurer and responsible for sending Ambry money received from my health insurance company.
For patient payment by credit card: 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.

FOR NY RESIDENTS:

☐ I am a New York resident and I give Ambry 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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Patient Name: _____ DOB: _____

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PLEASE SUBMIT THE FOLLOWING WITH THE TRF:

1. Clinic Notes 2. Pedigree 3. Insurance Card and Authorization Documents

CLINICAL HISTORY

PLEASE ATTACH PEDIGREE /CLINICAL CONSULTATION NOTES, IF AVAILABLE

Birth and Neonatal History ☐ Not Applicable

Gestational age at birth: _____ Birth weight: _____

Head circumference at birth (if available): _____

☐ Congenital anomalies, explain: _____

☐ Positive newborn screen, explain: _____

Seizure History ☐ Not Applicable

Age at first unprovoked seizure (first seizure without fever or other acute metabolic or structural cause): _____

Seizure types (choose all that apply):

☐ Infantile/epileptic spasms ☐ Myoclonic ☐ Generalized tonic clonic
☐ Tonic ☐ Typical absence ☐ Focal seizures
☐ Atonic ☐ Atypical absence

Are seizures: ☐ refractory ☐ well-controlled

Has this patient been diagnosed with an epilepsy syndrome?

☐ yes ☐ no ☐ unknown If yes, please specify: _____

Pulmonology History ☐ Not Applicable

☐ Positive newborn screen ☐ CBAVD ☐ Meconium ileus

☐ Infections: _____

☐ Sweat chloride: _____mmol/L ☐ Sweat chloride: ☐<40 ☐ 40-60 ☐ >60

☐ Pancreatic insufficiency IRT level: _____

☐ Respiratory distress, explain: _____

☐ Respiratory assistance devices: _____

☐ Ultrasound findings: _____

Developmental History ☐ Not Applicable

Developmental delay: ☐ yes ☐ no ☐ unknown

Delay prior to seizure onset: ☐ yes ☐ no ☐ unknown ☐ N/A

Type of delay (choose all that apply): ☐ motor ☐ language ☐ global

Intellectual disability: ☐ yes ☐ no ☐ unknown

Regression or plateau: ☐ yes ☐ no ☐ unknown

Does patient meet DSM-V diagnostic criteria for an autism spectrum disorder?:
☐ yes ☐ no ☐ unknown

Cardiac History ☐ Not Applicable

Sudden cardiac arrest ☐ Y ☐ N (if yes): # Episodes: _____

Age first incident: _____

Syncope ☐ Y ☐ N If yes, # Episodes: _____ Age first incident: _____

History of cardiomyopathy ☐ Y ☐ N Age at dx: _____

Cardiomyopathy type: _____

History of Arrhythmia ☐ Y ☐ N Age at dx: _____

Arrhythmia type: _____

☐ Congenital heart defect _____

Other History ☐ Not Applicable

☐ Hearing problems: _____ ☐ Vision problems: _____

☐ Migraine: _____ ☐ Psychiatric: _____

☐ Hematological: _____

☐ Suspected genetic condition: _____

☐ Other clinical findings: _____

Cancer History ☐ Not Applicable

Cancer/Tumor	Age at Dx	Pathology and Other Info
Brain		
Breast		Type: ER☐(+) ☐(-) ☐unk PR☐(+) ☐(-) ☐unk HER2/neu☐(+) ☐(-) ☐unk
2nd primary breast		Type: ER☐(+) ☐(-) ☐unk PR☐(+) ☐(-) ☐unk HER2/neu☐(+) ☐(-) ☐unk
Colorectal		Location:
Ovarian		☐ Fallopian tube ☐ Primary peritoneal
Melanoma/skin		
Prostate		Gleason Score: Metastatic: ☐ Yes ☐ No
Uterine		
Hematologic*		Type: ☐ Allogenic bone marrow or peripheral stem cell transplant*
Other Cancer		Type:
GI polyps		☐ Adenomatous Polyp #: ☐1 ☐2-5 ☐6-9 ☐10-19 ☐20-99 ☐100+ ☐ Other type: Polyp #: ☐1 ☐2-5 ☐6-9 ☐10-19 ☐20-99 ☐100+

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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
	☐	☐		
	☐	☐		
	☐	☐		
	☐	☐		

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Patient Name: _____ DOB: _____

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:

☐ Hereditary polyposis[1]

☐ Lynch syndrome/HNPCC[2]

☐ Hereditary breast and ovarian cancer[3]

☐ Testing is clinically indicated for other gene(s):

☐ None of the above

To complete your multi-gene order, please select a test option to the right. (See supplemental information on page 6 for details).

[1] APC/MUTYH [2] MLH1, MSH2, MSH6, PMS2, EPCAM [3] BRCA1/2
** Required: completed CustomNext-Cancer supplemental form.* ambrygen.com/forms

☐	BrainTumorNext®	8847	27 gene brain tumor test
☐	BRCAplus®	8836	8 gene breast cancer test
☐	BreastNext®	8820	17 gene breast cancer test
☐	CancerNext®	8824	34 gene cancer test
☐	CancerNext-*Expanded*®	8874	67 gene cancer test
☐	ColoNext®	8822	17 gene colorectal cancer test
☐	CustomNext-*Cancer*®	9510	up to 81 gene custom test*
☐	GYNplus®	8835	13 gene ovarian/uterine cancer test
☐	MelanomaNext®	8849	8 gene melanoma test
☐	OvaNext®	8830	25 gene ovarian/breast/uterine cancer test
☐	PancNext®	8042	13 gene pancreatic cancer test
☐	Pancreatitis panel	8022	6 gene pancreatitis test
☐	PGLNext®	5504	12 gene PGL/PCC test
☐	ProstateNext®	8845	14 gene prostate cancer test
☐	RenalNext®	5900	19 gene renal cancer test

☐ **Add +RNAinsight™ to selected panel^**
^Not available with BRCAplus (8836), STAT orders, or pancreatitis panel (8022); PAXgene® tube required for RNA

Add AmbryScore: ☐ Breast (Supplemental Ordering Form REQUIRED) ☐ Prostate

Single Syndrome Orders

Check to order	Test Name	Test Code	Description
Breast and/or Ovarian Cancer			
☐	*ATM*	9014	Ataxia-telangiectasia
☐	*BRCA1/2*	8838	Hereditary breast and ovarian cancer
☐	*BRCA1/2* Ashkenazi Jewish 3-site mutation panel	5892	
☐	*BRCA1/2* Ashkenazi Jewish 3-site mutation panel with reflex to *BRCA1/2* analysis if negative	5894	
☐	*CHEK2*	9016	
☐	*DICER1*	5260	
☐	*PALB2*	2366	
☐	*PTEN*	2106	*PTEN*-related disorders (including Cowden syndrome)
☐	*TP53*	2866	Li-Fraumeni syndrome
Endocrine Tumors			
☐	*MEN1*	2646	Multiple endocrine neoplasia type 1
☐	*RET* gene sequence	2680	Multiple endocrine neoplasia type 2
Gastrointestinal Cancer			
☐	*APC*	3040	Familial adenomatous polyposis
☐	*APC* and *MUTYH* concurrent	8726	Adenomatous polyposis
☐	*BMPR1A* and *SMAD4* concurrent	8604	Juvenile polyposis syndrome
☐	*CDH1*	4726	Hereditary diffuse gastric cancer
☐	*EPCAM* del/dup	8519	Lynch syndrome
☐	Lynch syndrome (concurrent)	8517	*MLH1, MSH2, MSH6, PMS2 + EPCAM* del/dup
☐	Lynch syndrome (sequential)	8515	Step 1: *MLH1, MSH2,* and *MSH6 + EPCAM* del/dup; Step 2: *PMS2*

Check to order	Test Name	Test Code	Description
Gastrointestinal Cancer (Cont.)			
☐	*MLH1*	8508	Lynch syndrome
☐	*MSH2 + EPCAM* del/dup	8510	Includes *MSH2* inversion
☐	*MSH2* inversion	2226	Lynch syndrome
☐	*MSH6*	8512	Lynch syndrome
☐	*MUTYH*	4661	*MUTYH*-associated polyposis
☐	*PMS2*	4646	Lynch syndrome
☐	*STK11*	2766	Peutz-Jeghers syndrome
Genitourinary Cancer			
☐	*BAP1*	9044	
☐	*FH*	6301	Hereditary leiomyomatosis and renal cell cancer
☐	*FLCN*	5921	Birt-Hogg-Dubé syndrome
☐	*VHL*	2606	Von-Hippel Lindau disease
☐	*TSC1* and *TSC2*	5904	Tuberous sclerosis complex
Skin Cancer/Melanoma			
☐	*CDKN2A* and *CDK4* concurrent	4708	Familial atypical multiple mole melanoma (FAMMM)
☐	*PTCH1*	5684	Gorlin syndrome
Other Hereditary Cancer Testing			
☐	*NF1*	5704	Neurofibromatosis type 1
☐	*NF2*	9024	Neurofibromatosis type 2
☐	*RB1*	5426	Hereditary retinoblastoma
☐	*SMARCB1*	7180	Schwannomatosis
Other Single Syndrome Orders			
☐	Please visit ambrygen.com/hereditary-cancer-single-gene-tests for details. Test Code(s): _____ Gene/Test Name(s): _____		

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Patient Name: _____ DOB: _____

CARDIOLOGY

Check to order	Test Name	Test Code	Description
Comprehensive Cardiovascular Panels			
☐	CardioNext®	8911	92 genes for hereditary cardiomyopathies and arrhythmias
☐	CustomNext-*Cardio*®	9520	Up to 167 genes related to hereditary cardiomyopathies, arrhythmias, TAAD, HHT, Noonan, and lipidemias. Required: completed CustomNext-*Cardio* supplemental form. ambrygen.com/forms
Arrhythmia Panels			
☐	LongQTNext™	8890	17 genes for long QT, Brugada and short QT syndromes
☐	RhythmNext®	8900	42 genes for long QT syndrome, Brugada and short QT syndromes, CPVT and ARVC
☐	CPVTNext®	8902	4 genes for catecholaminergic polymorphic ventricular tachycardia
Cardiomyopathy Panels			
☐	HCMNext®	8936	30 genes for hypertrophic cardiomyopathy
☐	HCMNext Reflex	8883	*MYBPC3, MYH7* reflex to HCMNext
☐	DCMNext®	8884	37 genes for dilated cardiomyopathy
☐	CMNext®	8887	56 genes for hereditary cardiomyopathy
☐	ARVCNext®	8904	11 genes for arrhythmogenic right ventricular cardiomyopathy

Check to order	Test Name	Test Code	Description
Familial Hypercholesterolemia			
☐	FHNext®	8680	4 genes (*APOB, LDLR, LDLRAP1, PCSK9*)
☐ Check this box if you would like to have the *SLCO1B1* c.521T>C polymorphism reported with FHNext, which has been associated in medical literature with statin-induced myopathies			
☐	FCSNext (Familial Chylomicronemia Syndrome)	8920	*APOA5, APOC2, GPIHBP1, LMF1, LPL*
☐	Sitosterolemia	8930	*ABCG5, ABCG8*
Aneurysms and Related Disorders			
☐	TAADNext®	8789	35 genes for thoracic aortic aneurysms/dissections, Marfan syndrome, Ehlers-Danlos and related disorders
☐	Marfan reflex to TAADNext	8783	*FBN1* reflex to TAADNext
Hereditary Hemorrhagic Telangiectasia (HHT)			
☐	HHTNext®	8672	*ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*
Noonan Syndrome			
☐	NoonanNext™	8402	18 genes for RASopathies
Other			
☐	Transthyretin amyloidosis	1560	*TTR*

CLINICAL GENOMICS

Check to order	Test Name	Test Code	Description
☐	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)
☐	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: _____
☐	ExomeNext-*Proband*	9993	Proband only exome sequencing
☐	ExomeNext-*Proband* plus mtDNA	9994	Proband only exome sequencing plus mtDNA sequencing
☐	ExomeNext-*Duo*	9991	Duo exome sequencing ☐ Opt-out of Candidate (novel) Genetic Etiologies
☐	ExomeNext-*Duo* plus mtDNA	9992	Duo exome sequencing ☐ Opt-out of Candidate (novel) Genetic Etiologies

Check to order	Test Name	Test Code	Description
☐	ExomeNext-*Trio*	9995	Trio exome sequencing ☐ Opt-out of Candidate (novel) Genetic Etiologies
☐	ExomeNext-*Trio* plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing ☐ Opt-out of Candidate (novel) Genetic Etiologies
☐	ExomeNext-*Rapid*®	9999R	☐ Opt-out of Candidate (novel) Genetic Etiologies
Must be ordered through AP*	ExomeNext-*Select*	9500	Up to 500 gene custom exome sequencing test

**AP is AmbryPort, our online portal ambrygen.com/ap*

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-Select orders.
☐ Opt-out: I choose to decline the ACMG Recommended List of secondary findings

ENDOCRINOLOGY

Check to order	Test Name	Test Code	Description
☐	Hereditary leiomyomatosis renal cell carcinoma	6301	*FH*
☐	Maturity-onset diabetes of the young	8310	*HNF1A, HNF4A, HNF1B, GCK, PDX1*
☐	Multiple endocrine neoplasia type I	2646	*MEN1*

Check to order	Test Name	Test Code	Description
☐	Multiple endocrine neoplasia type 2 and familial medullary thyroid cancer (FMTC)	2680	*RET* gene sequence
☐	Neurofibromatosis type 1	5704	*NF1*
☐	PGLNext®	5504	12 gene PGL/PCC test
☐	von-Hippel Lindau disease	2606	*VHL*

GASTROENTEROLOGY

Check to order	Test Name	Test Code	Description
☐	*CFTR* gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status
☐	Hirschsprung disease (*RET*-related)	2680	*RET* gene sequence

Check to order	Test Name	Test Code	Description
☐	Juvenile polyposis syndrome	8604	*BMPR1A, SMAD4*
☐	Pancreatitis	8022	*CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*
☐	Peutz-Jeghers syndrome	2766	*STK11*

HEMATOLOGY/ONCOLOGY

Check to order	Test Name	Test Code	Description
☐	DBANext	8550	11 genes for Diamond-Blackfan anemia
☐	DCNext	8161	7 genes for dyskeratosis congenita

Check to order	Test Name	Test Code	Description
☐	Shwachman-Diamond syndrome	1440	*SBDS*

MULTIPLE CONGENITAL ANOMALIES

Check to order	Test Name	Test Code	Description
☐	CHARGE syndrome	2380	*CHD7*
☐	CdLSNext - Cornelia de Lange syndrome	7040	*NIPBL, SMC1A, HDAC8, RAD21, SMC3*

Check to order	Test Name	Test Code	Description
☐	Noonan syndrome	8402	*PTPN11, SOS1, KRAS, RAF1*

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Patient Name: _____ DOB: _____

NEUROLOGY

Check to order	Test Name	Test Code	Description
Comprehensive Neuro Testing			
☐	CustomNext-*Neuro*	9540	Up to 196 gene custom neurology test. ☐ Check if parental samples are included
Epilepsy			
☐	EpiRapid®	7033	16 epilepsy genes with treatment associations
☐	EpiRapid reflex to EpilepsyNext	7034	16 epilepsy genes with treatment associations, reflex to 100 genes for epilepsy
☐	EpiFirst-*Fever*®	7011	13 genes for febrile seizures ☐ Check if parental samples are included
☐	EpiFirst-*Focal*®	7017	11 gene for non-lesional focal epilepsy ☐ Check if parental samples are included
☐	EpiFirst-*IS*®	7013	17 genes for infantile spasms ☐ Check if parental samples are included
☐	EpilepsyNext®	7019	100 genes for epilepsy ☐ Check if parental samples are included
Hereditary Neuropathy			
☐	Familial transthyretin amyloidosis	1560	*TTR*
Migraine			
☐	Familial hemiplegic migraine	7035	*ATP1A2, CACNA1A, PRRT2, SCN1A* ☐ Check if parental samples are included

Check to order	Test Name	Test Code	Description
Neurodevelopmental Disorders			
☐	AutismNext®	7024	48 genes for syndromic and non-syndromic autism spectrum disorders ☐ Check if parental samples are included
☐	Autism, macrocephaly	2106	*PTEN*
☐	Fragile X syndrome	4544	*FMR1* repeat expansion analysis and methylation studies
☐	IDNext®	7027	140 genes for syndromic and non-syndromic intellectual disability ☐ Check if parental samples are included
☐	Neurodevelopment-*Expanded*®	7028	196 genes for intellectual disability, autism spectrum disorders, and epilepsy ☐ Check if parental samples are included
☐	Rett syndrome	2026	*MECP2*
Neurocutaneous/Neuro-Oncology Disorders			
☐	Ataxia-telangiectasia	9014	*ATM*
☐	BrainTumorNext®	8847	27 genes for brain tumors
☐	HHTNext®	8672	*ACVRL1, ENG, SMAD4, GDF2, RASA1*
☐	Legius syndrome	5724	*SPRED1*
☐	Li-Fraumeni syndrome	2866	*TP53*
☐	Neurofibromatosis 1	5704	*NF1*
☐	Neurofibromatosis 2	9024	*NF2*
☐	Nevoid basal cell carcinoma syndrome/ Gorlin syndrome	5684	*PTCH1*
☐	Tuberous sclerosis complex	5904	*TSC1, TSC2*
☐	von Hippel-Lindau disease	2606	*VHL*

PULMONOLOGY

Check to order	Test Name	Test Code	Description
Congenital Central Hypoventilation Syndrome			
☐	Congenital central hypoventilation syndrome	1580	*PHOX2B* gene sequence
Cystic Fibrosis			
☐	508 FIRST®	1002	*CFTR* deltaF508 mutation analysis with reflex to *CFTR* gene sequence and deletion/duplication ☐ Report poly T/TG status
☐	*CFTR* gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status

Check to order	Test Name	Test Code	Description
Primary Ciliary Dyskinesia			
☐	PCDNext®	8122	21 genes for primary ciliary dyskinesia ☐ Report poly T/TG status
Pulmonary Fibrosis			
☐	Telomere-related pulmonary fibrosis	8140	*TERT, TERC*
Respiratory Distress Syndrome			
☐	Surfactant dysfunction (respiratory distress syndrome)	8100	*ABCA3, SFTPB, SFTPC* gene sequence

RHEUMATOLOGY

☐	Familial Mediterranean fever	5000	*MEFV*

VASCULAR

Check to order	Test Name	Test Code	Description
☐	EDS IV reflex to TAADNext	8791	*COL3A1* reflex to TAADNext
☐	Ehlers-Danlos type IV, vascular type	8790	*COL3A1*
☐	HHTNext®	8672	*ACVRL1, ENG, SMAD4, GDF2, RASA1*

Check to order	Test Name	Test Code	Description
☐	Marfan syndrome (MFS)	8781	*FBN1*
☐	MFS reflex to TAADNext	8783	*FBN1* reflex to TAADNext
☐	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, 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	*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, MITF, 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, CTRC, DICER1, EGFR, EPCAM, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIT, MAX, MEN1, MET, MITF, 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, MITF, 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, 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, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*

Genes Eligible for **+RNAinsight™**: *APC, ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MSH2, MSH6, MUTYH, NF1, PALB2, PMS2 EX1-10, PTEN, RAD51C, RAD51D, TP53*

* AmbryScore for Breast is available as an add on to this panel if all of the following eligibility criteria are met:
- Female biological sex
- 18-84 years old
- Non-Ashkenazi Jewish, N. European ancestry
- No personal history of cancer (excluding non-melanoma skin cancer)
- No personal history of atypical hyperplasia or lobular carcinoma in situ (LCIS)
- No personal or family history of a mutation in a breast cancer susceptibility gene[1]
  - [1] *ATM, BARD1, BLM* (if tested), *BRCA1, BRCA2, BRIP1, CDH1, CHEK2, FANCC* (if tested), *MRE11A, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53*

Note: AmbryScore supplemental ordering form is required for processing

^ AmbryScore for Prostate is available as an add on to this panel if all of the following eligibility criteria are met:
- Male biological sex
- 18-84 years old
- N. European ancestry
- No personal or family history of a mutation in a prostate cancer susceptibility gene[2]
  - [2] *ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, RAD51D, TP53*

Note: No additional ordering forms are required for processing