Ambry Genetics
Aliso Viejo, CA 92656 USA
Toll Free: +1.866.262.7943 Direct: +1.949.900.5500 | Fax: +1.949.900.5501 | ambrygen.com

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 ☐F ☐M	Ethnicity: ☐African American ☐Asian ☐Caucasian ☐Hispanic ☐Jewish (Ashkenazi) ☐Portuguese ☐Other:

SPECIMEN INFORMATION*

Type(s) ☐Blood (EDTA preferred) ☐Saliva ☐DNA ☐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.*

INDICATION(S) FOR TESTING

ICD-10 code(s):

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 /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 ☐).

Signature Required for Processing Medical Professional Signature: Date:

☐ 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 ☐ Check (Payable to Ambry Genetics) ☐ 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.

Signature Required for Self-Pay Patients and Research Consent Patient or Legal Guardian Signature: *Date:*

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Patient Name: _____ DOB: _____

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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: _____ # Episodes:_____ Age first incident:_____

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

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

☐ HCM ☐ DCM ☐ ARVD ☐ LVNC ☐ RCM

☐ Other cardiomyopathy types:_____

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

☐ Long QT ☐ Short QT ☐ Brugada ☐ CPVT ☐ ARVD

☐ Other arrhythmia types: _____

☐ 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: ☐ unaffected ☐ affected, list symptoms/dx:_____ Dx age: _____

Father: ☐ unaffected ☐ affected, list symptoms/dx:_____ Dx age: _____

Relation to patient	Maternal	Paternal	Disease	Dx age
	☐	☐		
	☐	☐		
	☐	☐		
	☐	☐		

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ORDERING CHECKLIST

- ☐ Clinic notes (with pedigree if available)
- ☐ 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).

ALLERGY AND IMMUNOLOGY

☐	Angioedema, hereditary	2746	*SERPING1*

CANCER

Multi-Gene Testing

☐	BrainTumorNext	8847	27 gene brain tumor test
☐	BRCAplus	8836	6 gene breast cancer test
☐	BRCAplus-*Expanded*	8837	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 67 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	4 gene pancreatitis test
☐	PGLNext	5504	12 gene PGL/PCC test
☐	ProstateNext	8845	14 gene prostate cancer test
☐	RenalNext	5900	19 gene renal cancer test

See Hereditary Cancer Testing Supplemental Information on page 7 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
Hereditary Breast and/or Ovarian Cancer			
☐	*ATM*	9014	Ataxia-telangiectasia
☐	*BRCA1/2*	8838	Hereditary breast and ovarian cancer
☐	*BRCA1/2* del/dup only	5890	
☐	*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)
☐	*SMARCA4*	9028	Rhabdoid tumor predisposition syndrome type 2
☐	*TP53*	2866	Li-Fraumeni syndrome
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
☐	*GREM1* duplication	8878	Mixed polyposis 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*
☐	*MLH1*	8508	Lynch syndrome
☐	*MSH2 + EPCAM* del/dup	8510	Includes *MSH2* inversion
☐	*MSH2* inversion	2226	Lynch syndrome

Check to order	Test Name	Test Code	Description
Gastrointestinal Cancer (Cont.)			
☐	*MSH6*	8512	Lynch syndrome
☐	*MUTYH*	4661	*MUTYH*-associated polyposis
☐	*PMS2*	4646	Lynch syndrome
☐	*POLD1* and *POLE* gene sequence	8877	Polymerase proofreading-associated polyposis
☐	*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
Endocrine Tumors			
☐	*MAX*	9062	Hereditary PGL/PCC
☐	*MEN1*	2646	Multiple endocrine neoplasia type 1
☐	*RET* gene sequence	2680	Multiple endocrine neoplasia type 2
☐	*SDHA*	9052	Hereditary PGL/PCC
☐	*SDHAF2*	9060	Hereditary PGL/PCC
☐	*SDHB*	9054	Hereditary PGL/PCC
☐	*SDHC*	9056	Hereditary PGL/PCC
☐	*SDHD*	9058	Hereditary PGL/PCC
☐	*TMEM127*	9066	Hereditary PGL/PCC
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
☐	*RB1*	5426	Hereditary retinoblastoma
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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CARDIOLOGY

Check to order	Test Name	Test Code	Description
Thoracic Aortic Aneurysms and Related Disorders			
☐	TAADNext	8789	22 genes for thoracic aortic aneurysms/dissections
☐	Marfan syndrome	8781	*FBN1*
☐	Marfan reflex to TAADNext	8783	*FBN1* reflex to TAADNext
☐	Ehlers-Danlos vascular type (EDS IV)	8790	*COL3A1*
☐	Ehlers-Danlos reflex to TAADNext	8791	*COL3A1* reflex to TAADNext
Arrhythmia, Long QT, and Brugada Panels			
☐	RhythmFirst	8888	12 genes for long QT syndrome
☐	RhythmNext	8900	36 genes for inherited arrhythmias
☐	RhythmNext Reflex	8901	RhythmFirst reflex to RhythmNext
☐	CPVTNext	8902	6 genes for catecholaminergic polymorphic ventricular tachycardia
Cardiomyopathy Panels			
☐	HCMFirst	8935	*MYBPC3, MYH7*
☐	HCMNext	8936	27 genes for hypertrophic cardiomyopathy
☐	HCMNext Reflex	8883	HCMFirst reflex to HCMNext

Check to order	Test Name	Test Code	Description
Cardiomyopathy Panels			
☐	DCMNext	8884	36 genes for dilated cardiomyopathy
☐	CMNext without *TTN*	8886	54 genes for hereditary cardiomyopathy
☐	CMNext with *TTN*	8887	55 genes for hereditary cardiomyopathy
☐	ARVDNext	8904	9 genes for arrhythmogenic right ventricular dysplasia
☐	LVNCNext	8906	8 genes for left ventricular non-compaction
Comprehensive Cardiovascular Panels			
☐	CardioNext without *TTN*	8910	84 genes for hereditary cardiomyopathies and arrhythmias
☐	CardioNext with *TTN*	8911	85 genes for hereditary cardiomyopathies and arrhythmias
☐	CustomNext-*Cardio*^^	9520	Up to 106 gene custom hereditary cardiomyopathies and arrhythmias test
Familial Hypercholesterolemia			
☐	FHNext	8680	4 genes (*APOB, LDLR, LDLRAP1, PCSK9*)

☐ Check this box if you would like to have the *SLC01B1* c.521T>C polymorphism reported, which has been associated in medical literature with statin-induced myopathies

CLINICAL GENOMICS

☐	Karyotype	3660	Chromosome analysis (requires green-top sodium-heparin tube)
☐	Karyotype, rule out mosaic	3662	Chromosome analysis (requires green-top sodium-heparin tube)
☐	SNP Array	5490	Chromosomal microarray (>2.6 million copy number probes and 750,000 SNP probes)
☐	Follow-up parental FISH studies – **ONLY available following SNP Array (5490) completed at Ambry**	3750	Sodium heparin tube, submit proband sample for positive control. Name of proband tested at Ambry: _____
☐	ExomeNext	9999	☐ Opt-out of analysis and reporting of Novel Genetic Etiologies
☐	ExomeNext-*Rapid*	9999R	☐ Opt-out of analysis and reporting of Novel Genetic Etiologies

Must be ordered through AP2*	ExomeNext-*Select*	9500	Up to 500 gene custom exome sequencing test

**AP2 is AmbryPort 2.0, our online portal ambrygen.com/ap2*

If ordering ExomeNext/ExomeNext-Rapid, please complete:

Secondary Findings Report: Check below to order the ACMG Recommended List of secondary findings. If neither box is checked secondary findings will not be reported. Secondary findings results are issued in a separate report. (For expanded secondary findings options and pricing please complete the "ExomeNext Expanded Secondary Findings Request Form" and submit with sample).

☐ Yes: I choose to receive the ACMG Recommended List of secondary findings

☐ No: I choose to decline the ACMG Recommended List of secondary findings

ENDOCRINOLOGY

☐	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*
☐	Multiple endocrine neoplasia type 2 and familial medullary thyroid cancer (FMTC)	2680	*RET* gene sequence

☐	Neurofibromatosis type 1	5704	*NF1*
☐	PGLFirst	5419	7 gene non-syndromic PGL/PCC test
☐	PGLNext	5504	12 gene PGL/PCC test
☐	von-Hippel Lindau disease	2606	*VHL*

GASTROENTEROLOGY

☐	Alagille syndrome	1640	*JAG1*
☐	Alpha-1 antitrypsin deficiency	1140	*SERPINA1*
☐	Hirschsprung disease (*RET*-related)	2680	*RET* gene sequence
☐	Juvenile polyposis syndrome	8604	*BMPR1A, SMAD4*
☐	Pancreatitis	8022	*PRSS1, SPINK1, CFTR, CTRC*
☐	Peutz-Jeghers syndrome	2766	*STK11*

☐	Wilson disease	1840	*ATP7B*
☐	*CFTR* gene sequence	1000	
☐	*CFTR* gene sequence with reflex to deletion/duplication analysis	1006	☐ Report poly T/TG status
☐	*CFTR* gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status

HEMATOLOGY/ONCOLOGY

☐	DBANext	8550	11 genes for Diamond-Blackfan anemia
☐	DCNext	8161	7 genes for dyskeratosis congenita

☐	*HBB*-related disorders	1048	*HBB*
☐	Shwachman-Diamond syndrome	1440	*SBDS*

^*Required: completed CustomNext-TAAD Supplemental Ordering Form. ambrygen.com/forms*

^^*Required: completed CustomNext-Cardio Supplemental Ordering Form. ambrygen.com/forms*

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Patient Name: _____ DOB: _____

INHERITED METABOLIC DISORDERS

Check to order	Test Name	Test Code	Description
Lysosomal Storage Disorders			
☐	NCLNext	7025	13 genes for neuronal ceroid lipofuscinosis/Batten disease ☐ Check if parental samples are included
☐	CLN1– Batten	7050	*PPT1*
☐	CLN2– Batten	7051	*TPP1*
☐	CLN3– Batten	7054	*CLN3*
☐	CLN10– Batten	7052	*CTSD*
☐	Gaucher disease	1820	*GBA*
☐	Hunter syndrome	1940	*IDS*
☐	Niemann-Pick disease, type C	8740	*NPC1, NPC2*
☐	Tay-Sachs disease	1240	*HEXA*
Leukodystrophies			
☐	Canavan disease	1226	*ASPA*
☐	Pelizaeus-Merzbacher disease	4180	*PLP1*
☐	X-linked adrenoleukodystrophy	3760	*ABCD1*
Other			
☐	Lesch-Nyhan syndrome	3940	*HPRT1*
☐	Menkes and related syndromes	3800	*ATP7A*
☐	Ornithine transcarbamylase (OTC) deficiency	4100	*OTC*
☐	Wilson disease	1840	*ATP7B*

MULTIPLE CONGENITAL ANOMALIES

☐	Alagille syndrome	1640	*JAG1*
☐	CHARGE syndrome	2380	*CHD7*
☐	Coffin-Lowry syndrome	4240	*RPS6KA3*
☐	CdLSNext - Cornelia de Lange syndrome	7040	*NIPBL, SMC1A, HDAC8, RAD21, SMC3*
☐	Kleefstra syndrome	7095	*EHMT1*
☐	*KRAS* - Noonan	2340	*KRAS*
☐	Lowe syndrome	4600	*OCRL*
☐	Noonan syndrome	8402	*PTPN11, SOS1, KRAS, RAF1*
☐	Oral-facial-digital syndrome	4080	*OFD1*
☐	*PTPN11*- Noonan	2280	*PTPN11*
☐	Prader-Willi syndrome	2440	Chromosome 15q11.2-q13 methylation studies
☐	*RAF1* - Noonan	2320	*RAF1*
☐	Rubinstein-Taybi syndrome	7083	*CREBBP*
☐	*SOS1* - Noonan	2300	*SOS1*
☐	Simpson-Golabi-Behmel syndrome	4420	*GPC3*
☐	Smith-Lemli-Opitz syndrome	2180	*DHCR7*
☐	Smith-Magenis syndrome	7163	*RAI1*
☐	Sotos syndrome	7143	*NSD1*

NEUROLOGY

Ataxia			
☐	Ataxia-telangiectasia	9014	*ATM*
Epilepsy			
☐	EpiRapid ***blood only, no saliva****	7033	16 epilepsy genes with treatment associations
☐	EpiRapid reflex to EpilepsyNext ***blood only, no saliva****	7034	16 epilepsy genes with treatment associations, reflex to 100 genes for epilepsy

Please complete the following for any EpiRapid test order:

Phone number for verbal results: _____

Name of provider to receive results: _____

Secure email: _____

If there is no answer at the above number, we will send an email to the address provided above. Please send an email prior to shipment including FedEx tracking number to receiving@ambrygen.com.

☐	CustomNext-*Epilepsy*^	9530	Up to 100 gene custom epilepsy test ☐ Check if parental samples are included
☐	EpiFirst-*Fever*	7011	13 genes for febrile seizures ☐ Check if parental samples are included
☐	EpiFirst-*Fever* reflex to EpilepsyNext	7012	13 genes for febrile seizures, reflex to 100 genes for epilepsy ☐ Check if parental samples are included
☐	EpiFirst-*Focal*	7017	11 gene for non-lesional focal epilepsy ☐ Check if parental samples are included
☐	EpiFirst-*Focal* reflex to EpilepsyNext	7018	11 gene for non-lesional focal epilepsy, reflex to 100 genes for epilepsy ☐ Check if parental samples are included
☐	EpiFirst-*IS*	7013	17 genes for infantile spasms ☐ Check if parental samples are included
☐	EpiFirst-*IS* reflex to EpilepsyNext	7014	17 genes for infantile spasms, reflex to 100 genes for epilepsy ☐ Check if parental samples are included
Epilepsy (Cont.)			
☐	Tuberous sclerosis (infantile spasms)	5904	*TSC1, TSC2*
☐	EpiFirst-*Neonate*	7015	10 genes for neonatal seizures ☐ Check if parental samples are included
☐	EpiFirst-*Neonate* reflex to EpilepsyNext	7016	10 genes for neonatal seizures, reflex to 100 genes for epilepsy ☐ Check if parental samples are included
☐	EpilepsyNext	7019	100 genes for epilepsy ☐ Check if parental samples are included
☐	PMEFirst	7020	*CSTB, EPM2A, NHLRC1* (includes *CSTB* repeat expansion) ☐ Check if parental samples are included
☐	PMENext	7022	21 genes for progressive myoclonus epilepsy (includes *CSTB* repeat expansion) ☐ Check if parental samples are included
☐	PMEFirst reflex to PMENext	7021	*CSTB, EPM2A, NHLRC1*, reflex to 21 genes for progressive myoclonus epilepsy (includes *CSTB* repeat expansion) ☐ Check if parental samples are included
☐	*CSTB* repeat expansion	7084	*CSTB* repeat expansion analysis
☐	Neurodevelopment-*Expanded*	7028	196 genes for epilepsy, intellectual disability, autism spectrum disorders ☐ 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

^*Required: completed CustomNext-Epilepsy Supplemental Ordering Form.* ambrygen.com/forms

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NEUROLOGY (CONT.)

Neurodevelopmental Disorders

☐	AutismFirst	7023	16 genes for syndromic autism spectrum disorders ☐ Check if parental samples are included
☐	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
☐	Angelman syndrome	7029	Methylation studies of 15q11-13, reflex to *UBE3A*
☐	Rett/AngelmanNext	7026	22 genes for Rett, Angelman, and related syndromes ☐ 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
☐	HHTFirst	8673	*ACVRL1, ENG, SMAD4*
☐	HHTNext	8672	*ACVRL1, ENG, SMAD4, GDF2, RASA1*
☐	HHTFirst reflex to HHTNext	8671	*ACVRL1, ENG, SMAD4* reflex to *GDF2, RASA1*
☐	Legius syndrome	5724	*SPRED1*
☐	Li-Fraumeni syndrome	2866	*TP53*
☐	Neurofibromatosis 1	5704	*NF1*
☐	*NF1* reflex to Legius	5730	*NF1* reflex to *SPRED1*
☐	Nevoid basal cell carcinoma syndrome/ Gorlin syndrome	5684 9050	*PTCH1* *SUFU*
☐	Tuberous sclerosis complex	5904	*TSC1, TSC2*
☐	von Hippel-Lindau disease	2606	*VHL*

PULMONOLOGY

Cystic Fibrosis

Check to order	Test Name	Test Code	Description
☐	508 ONLY	1008	*CFTR* deltaF508 mutation analysis
☐	508 FIRST	1002	*CFTR* deltaF508 mutation analysis with reflex to *CFTR* gene sequence and deletion/duplication
☐	CF 102	1018	102 *CFTR* disease-causing mutation panel
☐	*CFTR* gene sequence with reflex to deletion/ duplication analysis	1006	☐ Report poly T/TG status
☐	*CFTR* gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status
☐	*CFTR* gene sequence	1000	
☐	*CFTR* deletion/ duplication analysis	1004	
☐	Poly T/TG repeat analysis	1010	Poly T repeat analysis with reflex to TG repeat analysis
☐	*CFTR* SSA	1008	*CFTR* Single Site Analysis Mutation: _____

Alpha-1 Antitrypsin Deficiency

Check to order	Test Name	Test Code	Description
☐	Alpha-1 antitrypsin deficiency	1140	*SERPINA1*

Congenital Central Hypoventilation Syndrome

☐	Congenital central hypoventilation syndrome	1580	*PHOX2B* gene sequence

Primary Ciliary Dyskinesia

☐	PCDNext	8122	21 genes for primary ciliary dyskinesia

Pulmonary Fibrosis

☐	Telomere-related pulmonary fibrosis	8140	*TERT, TERC*

Respiratory Distress Syndrome

☐	Surfactant dysfunction (respiratory distress syndrome)	8100	*ABCA3, SFTPB, SFTPC* gene sequence
☐	*ABCA3*-related surfactant dysfunction	1300	*ABCA3* gene sequence
☐	Surfactant protein B deficiency	1160	*SFTPB* gene sequence
☐	Surfactant protein C deficiency	1180	*SFTPC* gene sequence

RHEUMATOLOGY

☐	Familial Mediterranean fever	5000	*MEFV*

VASCULAR

☐	HHTFirst - Hereditary hemorrhagic telangiectasia	8673	*ACVRL1, ENG, SMAD4*
☐	HHTNext	8672	*ACVRL1, ENG, SMAD4, GDF2, RASA1*
☐	HHTReflex to HHTNext	8671	*ACVRL1, ENG, SMAD4* reflex to *GDF2, RASA1*
☐	*GDF2* and *RASA1*	8674	
☐	Ehlers-Danlos type IV, vascular type	8790	*COL3A1*
☐	EDS IV reflex to TAADNext	8791	*COL3A1* reflex to TAADNext
☐	TAADNext	8789	22 genes for thoracic aortic aneurysms

SINGLE 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 ONLY: MATERNAL CELL CONTAMINATION (Required for fetal specimens)

☐ 1260 MCC for amniotic fluid culture or CVS (run concurrently with test) ☐ 1262 MCC Reference for maternal blood sample (No Charge)

Aliso Viejo, CA 92656 USA | Toll Free: +1.866.262.7943 Direct: +1.949.900.5500 | Fax: +1.949.900.5501 | ambrygen.com

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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, PRKAR1A, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL*
BRCAplus (6 genes)	8836	*BRCA1, BRCA2, CDH1, PALB2, PTEN, TP53*
BRCAplus-*Expanded* (8 genes)	8837	*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 67 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, 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*
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 (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, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*