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

COLLECTION DATE (REQUIRED)

If date of collection is not provided, three calendar days before specimen receipt will be used (for specimens stored longer than 30 days, the day of archive retrieval will be used as the date of service)

2. PATIENT INFORMATION

Legal Name (Last, First, MI)	Date of Birth (Month/DD/YYYY)	Sex Assigned at Birth ☐F ☐M	Gender (optional) ☐Man ☐Woman ☐Nonbinary ☐Self-described

Genetic Ancestry: ☐Ashkenazi Jewish ☐Asian ☐Black/African American ☐French Canadian/Cajun ☐Hispanic/Latino ☐Mediterranean ☐Middle Eastern ☐Native American ☐Pacific Islander ☐Portuguese ☐White ☐Unknown ☐Other:

MRN/National ID

Address	State/Province	Country

Phone	Email

SPECIMEN INFORMATION* (Please see ambrygen.com/specimen-requirements for details)

☐ Personal history of allogenic bone marrow or peripheral stem cell transplant

Specimen ID	Specimen Type

** Fetal specimens, cord blood and POC samples are not accepted from International clients.*

INDICATION(S) FOR TESTING

ICD-10 code(s):

ORDERING LICENSED PROVIDER/SENDING FACILITY (Each listed person will receive a copy of the report)

Facility Name (Facility Code)	Address	
State /Province	Country	Phone

Ordering Licensed Provider Name (Last, First)(Code)	NPI# (US only)	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 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 he/she has obtained and documented patient consent to genetic testing, consistent with all applicable privacy and health information laws in the patient's country of residence. The patient has also expressly consented to the transfer of their genetic information to the United States and the processing of their genetic information by a laboratory based the United States. 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.

Signature Required for Processing Medical Professional Signature: Date:

BILLING

☐ **INSURANCE BILLING** (Include copy of both sides of insurance card)

☐ **INSTITUTIONAL BILLING**

Facility Name ☐ *Send invoice to facility address above*

Address

Contact Name

Phone Number	Email/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.

☐ I agree to be contacted regarding future research studies for which I may be a candidate. Any future research projects will be subject to a separate informed consent process and participation is voluntary. Learn more about Ambry's privacy practices at https://www.ambrygen.com/legal/notice-of-privacy-practices.

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.

Patient Signature (I agree to terms above): Date:

Patient Consent To Testing

I acknowledge and agree that my health care provider has ordered genetic testing for processing at Ambry Genetics, a laboratory based in the United States and subject to U.S. based privacy laws, including the Health Insurance Portability and Accountability Act (HIPAA). I have provided my health care provider with the appropriate consent acknowledgment and documentation as required under the applicable laws in my country of residence for my genetic information to be sent to and processed in the United States. I have reviewed Ambry's Notice of Privacy Practices and understand and agree to the use of my health information in accordance with such privacy practices. I understand that I can exercise my rights to privacy consistent with HIPAA and Ambry's privacy notices.

Patient Signature (I agree to terms above): Date:

Ambry Genetics

Patient Name: _____ DOB: _____

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 Metastatic: ☐ Yes ☐ No Tumor is ☐ MSI-High or ☐ IHC-Abnormal

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:
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+

**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) ☐ Limited family history

Previously Detected Alteration(s): _____ Gene Name: _____ Testing Lab: _____

Patient previously tested at Ambry? ☐ Yes ☐ No Family previously tested at Ambry? ☐ Yes ☐ No

Name: _____ DOB: _____ Relation: _____

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

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

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

Relationship to Patient	Mat	Pat	Age at Dx	Family Testing and Cancer Type Details	Reason relative has not been tested
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact
	☐	☐			☐ Deceased ☐ Declines ☐ No Contact

Ambry Genetics®

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. Preverification will only be performed for ExomeNext or SNP Array testing.

For Reflex or Concurrent Testing:

Test 1: _____ ☐ Reflex to ☐ Concurrent with Test 2: _____ ☐ Reflex to ☐ Concurrent with Test 3: _____

See Reflex or Concurrent Testing section of the Supplemental Information page.

CANCER TEST ORDERS

! REQUIRED: Select a Primary Test Order

For Patients Meeting *BRCA1/2* Testing Criteria

☐ *BRCA1/2* test

For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (Lynch)

Lynch Syndrome test: ☐ *MLH1, MSH2, MSH6, PMS2, EPCAM*

For Patients Meeting Colorectal Cancer Syndrome Testing Criteria (polyposis)

Polyposis test: ☐ *APC/MUTYH*

☐ Other: _____

☐ None of the above (patient does not meet any genetic testing criteria)

Other Supplemental Test Options (Select if applicable)

☐ +RNAinsight® (Not available with BRCAplus, pancreatitis panel, or STAT orders; PAXgene® tube required for RNA)

Select an Optional Supplemental Test (Per payer policy, all tests in this section will be processed and billed separately; tests may be performed as a reflex.)

☐	BrainTumorNext®	8847	29 gene brain tumor test
☐	BRCANext®	8855	19 gene breast cancer test
☐	BRCANext-*Expanded*®	8860	21 gene breast cancer test
☐	BRCAPlus®	8836	13 gene breast cancer test
☐	CancerNext®	8824	34 gene cancer test
☐	CancerNext-*Expanded*®	8874	71 gene cancer test
☐	ColoNext®	8822	20 gene colorectal cancer test
☐	CustomNext-*Cancer*® Notes: _____ _____ _____	9510	up to 91 gene custom test^^
☐	MelanomaNext®	8849	9 gene melanoma test
☐	PancNext®	8042	13 gene pancreatic cancer test
☐	Pancreatitis panel	8022	6 gene pancreatitis test
☐	PGLNext®	5504	14 gene PGL/PCC test
☐	ProstateNext®	8845	14 gene prostate cancer test
☐	RenalNext®	5900	20 gene renal cancer test

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	
☐	*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

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: _____

Order	Test Name	Test Code	Description
CARDIOLOGY			
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
CLINICAL GENOMICS			
☐	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
☐	ExomeNext-*Duo* plus mtDNA	9992	Duo exome sequencing plus mtDNA sequencing
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*
GASTROENTEROLOGY			
☐	*CFTR* gene sequence and deletion/duplication analysis	1007	☐ Report poly T/TG status
☐	Hirschsprung disease (*RET*-related)	2680	*RET* gene sequence
HEMATOLOGY/ONCOLOGY			
☐	DBANext	8550	11 genes for Diamond-Blackfan anemia
☐	DCNext	8161	7 genes for dyskeratosis congenita
MULTIPLE CONGENITAL ANOMALIES			
☐	NoonanNext™	8402	18 genes for Noonan syndrome and RASopathies

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 *SLC01B1* 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*
☐	ExomeNext-*Trio*	9995	Trio exome sequencing
☐	ExomeNext-*Trio* plus mtDNA	9996	Trio exome sequencing plus mtDNA sequencing
☐	ExomeNext-*Rapid*®	9999R	Rapid Trio exome sequencing plus mtDNA sequencing (Institutional billing or patient payment only)

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

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	14 gene PGL/PCC test
☐	von-Hippel Lindau disease	2606	*VHL*
☐	Juvenile polyposis syndrome	8604	*BMPR1A, SMAD4*
☐	Pancreatitis	8022	*CASR, CFTR, CPA1, CTRC, PRSS1, SPINK1*
☐	Peutz-Jeghers syndrome	2766	*STK11*
☐	Shwachman-Diamond syndrome	1440	*SBDS*

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Patient Name: _____ DOB: _____

NEUROLOGY

☐ **Opt-in to Reporting of Variants of Unknown Significance (VUS)**
For patients undergoing an epilepsy, neurodevelopmental disorder, or familial hemiplegic migraine panel, checking this box indicates that VUS identified on the test(s) ordered below will be reported for this patient. If you do not check this box, VUS will NOT be reported.

☐ **Parental samples provided for cosegregation**
Cosegregation testing of family members is available for the following panels: EpiRapid, EpilepsyNext, EpilepsyNext-Expanded, AutismNext, NeurodevelopmentNext, Familial hemiplegic migraine

Order	Test Name	Test Code	Description
Epilepsy			
☐	EpiRapid®	6862	22 epilepsy genes with treatment associations
☐	EpilepsyNext®	6864	124 genes for epilepsy
☐	EpilepsyNext-*Expanded*™	6865	>890 genes associated with seizures, primarily with neonatal to childhood onset
Hereditary Neuropathy			
☐	Familial transthyretin amyloidosis	1560	*TTR*
Migraine			
☐	Familial hemiplegic migraine	6866	*ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1*

Note: Gene lists for EpilepsyNext-*Expanded* are updated annually due to proactive review of current literature using an internal, peer-reviewed clinical validity scheme (Smith ED, Radtke K, Rossi M, *et al.* 2017 Human mutation 38(5):600-608). The patient's test report will include a list of genes evaluated. For up-to-date gene lists, visit ambrygen.com

Order	Test Name	Test Code	Description
Neurodevelopmental Disorders			
☐	AutismNext®	6863	72 genes for non-syndromic autism spectrum disorders and/or intellectual disability
☐	Autism, macrocephaly	2106	*PTEN*
☐	Fragile X syndrome	4544	*FMR1* repeat expansion analysis and methylation studies
☐	NeurodevelopmentNext™	6861	202 genes known to cause developmental delay, intellectual disability and/or autism spectrum disorders
Neurocutaneous/Neuro-Oncology Disorders			
☐	Ataxia-telangiectasia	9014	*ATM*
☐	BrainTumorNext®	8847	29 genes for brain tumors
☐	HHTNext®	8672	*ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*
☐	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*

For Reflex or Concurrent Testing:

Test 1: _____ ☐ Reflex to ☐ Concurrent with Test 2: _____ ☐ Reflex to ☐ Concurrent with Test 3: _____

See Reflex or Concurrent Testing section of the Supplemental Information page.

PULMONOLOGY

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

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

VASCULAR

☐	HHTNext®	8672	*ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*
☐	Marfan syndrome reflex to TAADNext	8783	*FBN1* reflex to TAADNext

☐	TAADNext®	8789	35 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

Hereditary Cancer Multi-Gene Tests

Test Name	Test Code	Genes
Adenomatous polyposis	8726	*APC, MUTYH*
BrainTumorNext® (29 genes)	8847	*AIP, ALK, APC, CDKN1B, CDKN2A, DICER1, EPCAM, LZTR1, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PHOX2B, PMS2, POT1, PRKAR1A, PTCH1, PTEN, SMARCA4, SMARCB1, SMARCE1, SUFU, TP53, TSC1, TSC2, VHL*
BRCANext™ (19 genes)	8855	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
BRCANext-*Expanded*™ (21 genes)	8860	*ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53*
BRCAPlus® (13 genes)	8836	*ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53*
CancerNext® (34 genes)	8824	*APC, ATM, AXIN2, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, SMARCA4, STK11, TP53*
CancerNext-*Expanded*® (71 genes)	8874	*AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EGLN1, EPCAM, FH, FLCN, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL*
ColoNext® (20 genes)	8822	*APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
CustomNext-*Cancer*® (up to 91 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, EGLN1, EPCAM, FAM175A(ABRAXAS1)†, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MLH3†, MRE11A†, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PALLD†, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50†, RAD51C, RAD51D, RB1, RECQL, RET, RINT1†, RPS20†, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, STK11, SUFU, TERT†, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2* For Medicare Patients: At a minimum, the following core genes must be included in the panel to ensure Medicare coverage: *APC, ATM, BRCA1, BRCA2, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, TP53.*
HBOC	8838	*BRCA1, BRCA2*
Lynch syndrome/HNPCC	8517	*MLH1, MSH2, MSH6, PMS2 + EPCAM del/dup*
MelanomaNext® (9 genes)	8849	*BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN, RB1, 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® (14 genes)	5504	*EGLN1, FH, KIF1B, 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® (20 genes)	5900	*BAP1, CHEK2, EPCAM, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL*

† Limited evidence gene

Specimen Requirements

Blood/saliva from patients with a history of allogenic bone marrow or stem cell transplant cannot be used for genetic testing. Blood/saliva from patients with active hematological disease is not recommended. An alternative specimen may be needed. Please see ambrygen.com/specimen-requirements for details.

Buccal swab sample available for chromosomal microarray (SNP array, familial targeted microarray), CustomNext-*Neuro*®, epilepsy, ExomeNext, fragile X syndrome, hereditary neuropathy (familial transthyretin amyloidosis), HHTNext, migraine (familial hemiplegic migraine), and neurodevelopmental disorder tests. Buccal swab samples from patients from patients with a history of allogenic bone marrow or stem cell transplant should not be used for genetic testing. For these patients, an alternative specimen (e.g. cultured fibroblasts) is required. Testing on buccal swab samples from patients with active hematological disease is not recommended. An alternative specimen (e.g. cultured fibroblasts) is recommended. Please see ambrygen.com/specimen-requirements for details.

Please note that Ambry cannot guarantee the viability of your specimen for testing at our laboratory, given the logistics of international specimen transfer. Testing may not be completed on specimens of inadequate quality due to specimen transfer issues not under Ambry's control, including, but not limited to, delays at customs, or other transfer-related delays. Ambry or your health care provider will reach out to you in such a case to rearrange for a specimen collection and transfer for completion of the ordered test.

Reflex or Concurrent Testing

Concurrent testing is when multiple tests are initiated at the same time. When multiple tests are ordered on the same test requisition form, testing will be run concurrently unless otherwise specified.

Reflex testing is when a subsequent test is initiated pending the outcome of the initial test. Reflex testing may result in delayed reporting of results.

For reflex test orders:

- Any diagnostic finding at any step will result in cancellation of any subsequent reflex tests.
- Non-diagnostic findings (including VUS or Uncertain results) will automatically reflex to the subsequent test.
- Secondary findings results do not impact whether a subsequent test is initiated or canceled.

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