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

## PATIENT INFORMATION

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

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

| Address | City | State | Zip |
|---|---|---|---|
| | | | |

| Phone | Email |
|---|---|
| | |

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

Type(s) ☐ Blood (EDTA preferred) ☐ Saliva ☐ Buccal Swabˆ ☐ DNA ☐ Cord Blood** ☐ Other**: ☐ Personal history of allogenic bone marrow or peripheral stem cell transplant*

| Collection Date | Specimen ID | Medical Record # |
|---|---|---|
| | | |

** 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*

*** 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.*

*ˆ 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.*

Phlebotomy Services Request: ☐ Phlebotomy draw ☐ Send blood kit to patientˆˆ ☐ Send saliva kit to patient | ☐ Insurance preverification first (available for ExomeNext and SNP array only)

*† 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 the medical management change depending on the results of the test? ☐ Yes ☐ No

**Was genetic counseling completed?** ☐ Yes ☐ No ☐ Unknown Date Genetic Counseling was Performed: _____

## 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:**
In many cases, we will start testing immediately (may vary by test or for patients with government insurance). We will attempt to contact the patient if the estimated out-of-pocket costs are > USD $100.

Special Billing Notes:

## ☐ 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. **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:

Ambry Genetics

Patient Name: _____ DOB: _____

Comprehensive Test Requisition Form - Page 2 of 6

**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+<br>☐ 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 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: _____

| 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 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 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 | 18 gene breast cancer test |
| ☐ | BRCANext-*Expanded*™ | 8860 | 23 gene breast cancer test |
| ☐ | BRCAplus® | 8836 | 8 gene breast cancer test |
| ☐ | CancerNext® | 8824 | 36 gene cancer test |
| ☐ | CancerNext-*Expanded*® | 8874 | 77 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): _____ | | |

Ambry Genetics®

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 |
| **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 | |
| 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 | | | |
| ☐ | 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* |
| **NEPHROLOGY** | | | |
| ☐ | NephrolithiasisNext | 9580 | 30 genes for nephrolithiasis and nephrocalcinosis |

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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.*

| Order | Test Name | Test Code | Description |
|---|---|---|---|
| **Comprehensive Neuro Testing** | | | |
| Must order through Ambry-Port | CustomNext-*Neuro*™ | 9545 | Customizable test of up to 500 genes from neuro menu<br>☐ Check if parental samples are included |
| **Epilepsy** | | | |
| ☐ | EpiRapid® | 6862 | 22 epilepsy genes with treatment associations<br>☐ Check if parental samples are included |
| ☐ | EpilepsyNext® | 6864 | 124 genes for epilepsy<br>☐ Check if parental samples are included |
| ☐ | EpilepsyNext-*Expanded*™ | 6865 | >890 genes associated with seizures, primarily with neonatal to childhood onset<br>☐ Check if parental samples are included |
| **Hereditary Neuropathy** | | | |
| ☐ | Familial transthyretin amyloidosis | 1560 | *TTR* |
| **Migraine** | | | |
| ☐ | Familial hemiplegic migraine | 6866 | *ATP1A2, CACNA1A, PRRT2, SCN1A*<br>☐ Check if parental samples are included |

**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

**For Reflex or Concurrent Testing:**

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

| Order | Test Name | Test Code | Description |
|---|---|---|---|
| **Neurodevelopmental Disorders** | | | |
| ☐ | AutismNext® | 6863 | 72 genes for non-syndromic autism spectrum disorders and/or intellectual disability<br>☐ Check if parental samples are included |
| ☐ | 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<br>☐ Check if parental samples are included |
| **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* |

## PULMONOLOGY

| 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<br>☐ 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<br>☐ 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

## 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® (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™ (18 genes) | 8855 | *ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53* |
| BRCANext-*Expanded*™ (23 genes) | 8860 | *ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, SMARCA4, STK11, TP53* |
| BRCAplus® (8 genes) | 8836 | *ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, TP53* |
| CancerNext® (36 genes) | 8824 | *APC, ATM, AXIN2, BARD1, BRCA1, BRCA2, BRIP1, BMPR1A, CDH1, CDK4, CDKN2A, CHEK2, DICER1, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RECQL, SMAD4, SMARCA4, STK11, TP53* |
| CancerNext-*Expanded*® (77 genes) | 8874 | *AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, BMPR1A, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, CTNNA1, DICER1, EGFR, EGLN1, EPCAM, FANCC, FH, FLCN, GALNT12, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, POT1, PMS2, POLD1, POLE, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RECQL, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2* |
| 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)<br>Required: complete CustomNext-*Cancer* supplemental form.<br>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*<br>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



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