

## GENERAL TEST REQUISITION

Arrows "▶" Mandatory for Processing

Patient Information	
▶ DOB MM - DD - YEAR	▶ Last Name    ▶ First Name    Middle Initial
▶ Gender <input type="checkbox"/> F <input type="checkbox"/> M	▶ Street Address, City, State, Zip
▶ Ethnicity <input type="checkbox"/> African American <input type="checkbox"/> Asian <input type="checkbox"/> Caucasian <input type="checkbox"/> Hispanic <input type="checkbox"/> Jewish (Ashkenazi) <input type="checkbox"/> Specify: _____	▶ Home Phone    Work/Cell
<b>Specimen</b> ▶ Collection Date: _____ Specimen ID: _____ MR#: _____ Specimen Type (See Requirements)  <input type="checkbox"/> Blood <input type="checkbox"/> Blood Spot <input type="checkbox"/> DNA <input type="checkbox"/> Cultured Amniocytes <input type="checkbox"/> Cultured CVS <input type="checkbox"/> CVS Tissue <input type="checkbox"/> Other: _____	<b>Previous Test History</b> Previously Detected Mutations: _____ Testing Lab: _____ Patient previously tested at Ambry? <input type="checkbox"/> Yes <input type="checkbox"/> No Family previously tested at Ambry? <input type="checkbox"/> Yes <input type="checkbox"/> No Name: _____ Relation: _____ Name: _____ Relation: _____ Name: _____ Relation: _____ Name: _____ Relation: _____

Contact and Organization Information	
▶ Authorized Ordering Physician	NPI#
▶ Ph	▶ FX
▶ Ordering Clinician Email	
▶ Facility Name and Address	ID#
Additional Results Recipient	
Medical Professional Name:	
Facility Name and Address	<input type="checkbox"/> Same As Above
Ph	Fx
▶ Form Completed by	▶ Phone
<p>By ordering testing, the medical professional or authorized person acknowledges the patient has been supplied information regarding genetic testing and the patient has given consent for genetic testing to be performed and that the signed consent form is on file. I confirm that this is medically necessary for the diagnosis or detection of a disease, illness, impairment, syndrome or disorder, and that these results will be used in the medical management and treatment decisions for this patient.</p> <p><b>Medical Professional Signature*</b>    Mandatory for Medicare/Medicaid</p> <p>X _____ Date: _____</p> <p><small>* MD/DO, Clinical Nurse Specialist, Nurse-Midwives, Nurse Practitioner, Physician Assistant</small></p> <p>Does this patient give consent to the use of their sample for research?  <input type="checkbox"/> Yes <input type="checkbox"/> No    Consent is implied if a box is not marked</p>	

▶ Indication for Testing <i>(please list clinical findings)</i> <input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier Screening <input type="checkbox"/> Research <input type="checkbox"/> Positive Newborn Screen <input type="checkbox"/> Family History <input type="checkbox"/> Other _____	<b>List Clinical Findings:</b>
ICD-9 Codes: _____	Sweat Chloride:

**Billing Information - Mandatory For Processing**     Preverification already performed, paperwork is on file, no need to fill out billing information again

AMBRY GENETICS provides a selection of convenient billing options. Please choose an option below and supply all requested information for your selection. Keep in mind that patient testing will be delayed until all of the billing requirements have been met. Choose an option below.

<input type="checkbox"/> Bill Facility <input type="checkbox"/> same as ordering facility	<input type="checkbox"/> Bill Insurance Include card copy (both sides)	<input type="checkbox"/> Pre-Payment
Facility Name	A completed Advance Beneficiary Notice of coverage (ABN) is required for Medicare patients. Ambry will pre-verify patient insurance coverage and if estimated patient out-of-pocket costs exceed \$350, patient is notified. Ambry Genetics will no longer perform Preverification for tests priced under \$200.	Payment Type <input type="checkbox"/> Check <input type="checkbox"/> Mastercard <input type="checkbox"/> Discover <input type="checkbox"/> Visa <input type="checkbox"/> American Express
Address, City, State, Zip	Name of Insured    Relation to patient? <input type="checkbox"/> Self <input type="checkbox"/> Parent <input type="checkbox"/> Spouse	Card Number    Exp Date
Contact Person	Insurance Company Name and Address	Cardholder Name    Amount \$
Contact Person Phone	Insurance Phone	Signature    Date X _____
	Member ID #    Group #	<b>Patient Acknowledgement</b> I hereby authorize my insurance benefits to be paid directly to Ambry Genetics Corporation and authorize them to release medical information concerning my testing to my insurer. I hereby acknowledge I am financially responsible for any amounts not paid by insurer.
	Authorization #    Date	
		X    Date

**MARK A TEST ON SUBSEQUENT PAGES FOR PROCESSING**

Thank You for Choosing Ambry Genetics



► Test Directory

Requisition Form

**DYSKERATOSIS CONGENITA (DC) - (EDTA Tube)**

- 8161 DC - *DKC1, TINF2, TERC, NHP2, NOP10, TERT* gene sequence (concurrent)
- 8160 DC - Steps 1 through 3
- 1960 *DKC1* gene sequence  Step 1
- 1980 *TINF2* exon 6 sequence only  Step 1
- 2120 *TERC* gene sequence  Step 1
- 2060 *NHP2* exon 4 sequence only  Step 2
- 2080 *NOP10* exon 2 sequence only  Step 2
- 2140 *TERT* gene sequence  Step 3
  
- 5160 *WRAP53* gene sequence

**EXOME SEQUENCING FOR CLINICAL DIAGNOSIS - (EDTA Tube)**

TEST NOT ORDERABLE HERE. PLEASE OBTAIN THE EXOME TEST REQUISITION.

Additional Information

**FAMILIAL HYPERCHOLESTEROLEMIA - (EDTA Tube)**

- 8680 Familial Hypercholesterolemia Comprehensive Evaluation (*LDLR* and *PCSK9* gene sequence and *APOB* partial gene sequence with *LDLR* deletion/duplication)
- 8582 Familial Hypercholesterolemia (*LDLR* and *APOB* partial gene sequence reflex to *LDLR* deletion/duplication)
- 2780 *LDLR* gene sequence
- 2784 *LDLR* deletion/duplication
- 2800 *APOB* partial gene sequence
- 2804 *PCSK9* gene sequence

**GASTROENTEROLOGY - (EDTA Tube)**

- 8022 Pancreatitis Plus (*CFTR, PRSS1, SPINK1, CTSC* gene sequence)
- 8020 Pancreatitis (*CFTR, PRSS1, SPINK1* gene sequence)
- 8040 Pancreatitis Amplified (*CFTR, PRSS1, SPINK1* with *CFTR* del/dup)
- 1100 *PRSS1* gene sequence
- 1120 *SPINK1* gene sequence
- 1660 *CTSC* gene sequence
- 1840 Wilson Disease (*ATP7B* gene sequence)
- 1440 Shwachman-Diamond Syndrome (*SBDS* gene sequence)

Additional Information

**GENETICS - (EDTA Tube)**

- 8640 AmbrySCREEN™
- 1640 Alagille (*JAG1* gene sequence and deletion/duplication)
- 1641 Alagille (*JAG1* deletion/duplication)
- 8642 Amyotrophic Lateral Sclerosis (*SOD1, ANG, FIG4, FUS* and *TARDBP* gene sequence) (concurrent)
- 8620 Amyotrophic Lateral Sclerosis (*SOD1* reflex to *ANG, FIG4, FUS, TARDBP* gene sequence)
- 8622 Amyotrophic Lateral Sclerosis (*SOD1* gene sequence)
- 1320 Aminoglycoside-Related Hearing Loss (*MT-RNR1* gene sequence)
- 5280 Andermann Syndrome (*SLC12A6* gene sequence)
- 8520 Angelman Syndrome (*SNRPN* methylation reflex to *UBE3A* gene sequence)
- 2400 Angelman Syndrome (*UBE3A* gene sequence)
- 2420 Angelman-like Syndrome (*SLC9A6* gene sequence)
- 2440 Angelman/Prader-Willi Syndrome (*SNRPN* methylation)
- 1808 Ashkenazi Jewish Panel™ with all 16 conditions
- 1804 Ashkenazi Jewish FlexPanel™ as marked below
  - Bloom (*BLM*)
  - Cystic Fibrosis (*CFTR*)
  - Glycogen Storage Disease 1a (*GSD1a*)
  - Maple Syrup Urine Disease (*BCKDHA/B*)
  - Maple Syrup Urine Disease Type 3 (*DLD*)
  - Mucopolipidosis Type IV (*MLDV*)
  - Canavan (*ASPA*)
  - Joubert Syndrome (*TMEM216*)
  - Familial Dysautonomia (*IKBKAP*)
  - Niemann-Pick A (*SMPD1*)
  - Gaucher (*GBA*)
  - Fanconi Anemia Type C (*FANCC*)
  - Tay-Sachs (*HEXA*)
  - Nemaline Myopathy (*NEB*)
  - Usher Syndrome Type 1F (*PCDH15*)
  - Usher Syndrome Type III (*CLRN1*)
- 4940 Aspartylglucosaminuria (*AGA* gene sequence)
- 1040 Beta Thalassemia Plus (*HBB* gene sequence with 619del check)
- 1226 Canavan (*ASPA* gene sequence and deletion/duplication) (concurrent)
- 1220 Canavan (*ASPA* gene sequence reflex deletion/duplication)
- 1370 Congenital Hyperinsulinism-Hyperammonemia (*GLUD1* gene sequence)
- 1364 Congenital Hyperinsulinism (*KCNJ11* gene sequence)
- 2380 CHARGE Syndrome (*CHD7* gene sequence)
- 4960 Dihydropyrimidine Deyhydrogenase Deficiency (*DPYD* gene sequence)
- 1720 Fabry Disease (*GLA* gene sequence)
- 5000 Familial Mediterranean Fever (*MEFV* gene sequence)
- 1820 Gaucher Disease (*GBA* gene sequence)
- 1600 Glutaric Acidemia Type 1 (*GCDH* gene sequence)
- 4880 Glutathione Synthetase Deficiency (*GSS* gene sequence)
- 1880 Glycogen Storage Disease Type Ia (*G6PC* gene sequence)
- 1900 Glycogen Storage Disease Type Ib (*SLC37A4* gene sequence)
- 2746 Hereditary Angioedema (*SERPINC1* gene sequence and deletion/duplication)
- 2708 Hirschsprung Disease (*RET* gene sequence) (concurrent)
- 2700 Hirschsprung Disease Steps 1 and 2 (*RET*)
  - 2704 Step 1 only: exons 2,3,5,6,9,10,12,13,17 gene sequence
  - 2706 Step 2 only: rest of gene sequence
- 1940 Hunter Syndrome (*IDS* gene sequence)
- 2160 Hurler Syndrome (*IDUA* gene sequence)
- 5020 Hyperoxaluria Type 2 (*GRHPR* gene sequence)
- 3200 Infantile Spasms (*CDKL5* gene sequence)
- 4860 Lysosomal Free Sialic Acid-Storage (Salla) Diseases (*SLC17A5* gene sequence)
- 8780 Marfan Syndrome NextGen Sequencing Panel
- 8782 Marfan Syndrome NextGen Sequencing Panel Steps 1 and 2
  - Step 1 *FBN1* gene sequence
  - Step 2 *ACTA2, CBS, FBN2, MYH11, COL3A1, SLC2A10, SMAD3, TGFBFR1, TGFBFR2* gene sequence
- 4900 MCAD - Medium-chain acyl-CoA dehydrogenase (*ACADM* gene sequence)
- 5180 Mucopolipidosis Type IV (*MCOLN1* gene sequence)
- 1360 Neonatal Diabetes (*KCNJ11* gene sequence)
- 1620 Neonatal Diabetes (*INS* gene sequence)
- 1860 Niemann-Pick Disease Types A & B (*SMPD1* gene sequence)
- 8122 Primary Ciliary Dyskinesia NextGen Sequencing Panel
- 4840 Rhizomelic Chondrodysplasia Punctata Type 1 (*PEX7* gene sequence)
- 1760 Phenylketonuria - PKU (*PAH* gene sequence)
- 1740 Pompe Disease (*GAA* gene sequence)
- 2180 Smith-Lemli-Opitz Syndrome (*DHCR7* gene sequence)
- 1240 Tay-Sachs Plus (*HEXA* gene sequence)
- 5240 Tay-Sachs Enzyme Assay (*HEXA* Leukocytes)
- 1560 Transthyretin Amyloidosis (*TTR* gene sequence)
- 4920 VLCAD - Very long-chain acyl-CoA dehydrogenase (*ACADVL* gene sequence)
- 1700 Warfarin Sensitivity (*CYP2C9* & *VKORC1* SNP analysis)
- 5220 Y Chromosome Microdeletion Analysis
- Thrombophilia (5140) (1 EDTA Lavender Top)**
  - 5141 Factor II (Prothrombin G20210A)
  - 5143 Factor V (Leiden)
  - 5145 *MTHFR* (C677T and A1298C)

► Test Directory

Requisition Form

**HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT) - (EDTA Tube)**

- 8662 HHT *ACVRL1, ENG and SMAD4* gene sequence with *ACRL1* and *ENG* deletion/duplication (concurrent)
  - 1680 HHT *ACVRL1 & ENG* gene sequence and deletion/duplication
  - 8660 HHT Steps 1 through 3
    - 1683 Step 1 *ACVRL1 & ENG* gene sequence
    - 1681 Step 2 *ACVRL1 & ENG* deletion/duplication
    - 1684 Step 3 *SMAD4* gene sequence
- Call HHT Single Gene Deletion/Duplication GENE \_\_\_\_\_

**PULMONOLOGY - (EDTA Tube)**

- Cystic Fibrosis - (EDTA Tube)**
- 1002 508 FIRST™ (deltaF508 reflex to *CFTR* Amplified)
  - 1012 508 ONLY™ (deltaF508 mutation only)
  - 1007 *CFTR* Amplified (*CFTR* gene sequence and deletion/duplication) (concurrent)
    - Report PolyT / TG Status
  - 1006 *CFTR* Amplified (*CFTR* gene sequence reflex deletion/duplication)
    - Report PolyT / TG Status
  - 1000 *CFTR* gene sequence
  - 1004 *CFTR* deletion/duplication
  - 1010 *CFTR* TG repeat analysis (Poly T Variant & TG Repeat)

**NEUROLOGY / INTELLECTUAL DISABILITY**

- 8630 **XLMR Evaluation** Steps 1 and 2 (reflex to next step when negative)
  - Step 1 Ambry CMA: 180K Oligo Array (EDTA + Na Heparin)
  - Note: This CMA has increased coverage on X chromosome
  - Step 2 XLMR Next Gen SuperPanel™ (sequencing panel for 81 genes) (EDTA)
- 8628 **XLMR Comprehensive Evaluation** Steps 1-3 (reflex to next step when negative)
  - Step 1 Routine Chromosome Analysis/Karyotype and Fragile X DNA Analysis (EDTA + Na Heparin)
  - Step 2 Ambry CMA: 180K Oligo Array (EDTA + Na Heparin)
  - Note: This CMA has increased coverage on X chromosome
  - Step 3 XLMR Next Gen SuperPanel™ (sequencing panel for 81 genes) (EDTA)

To order any test in a different order, select that test above and write in the sequence order  
 Note: Multiple tests require multiple samples

Individual Test Options Below

- 8626 XLMR Next Gen SuperPanel™ (1 EDTA)
- 3664 Routine Chromosome Analysis/Karyotype (1 Na Heparin)
- 4544 Fragile X DNA Analysis (1 EDTA)
- 3020 FRAXE (*FMR2*) DNA Analysis (1 EDTA)
- 3140 *ARX*-Related X-Linked Mental Retardation
- 4400 *ATRX*-Related X-Linked Mental Retardation
- 3180 *CASK*-Related X-Linked Mental Retardation
- 3220 *CUL4B*-Related X-Linked Mental Retardation
- 4780 *L1CAM*-Related X-Linked Mental Retardation
- 3380 *NLGN3*-Related X-Linked Mental Retardation
- 3400 *NLGN4*-Related X-Linked Mental Retardation
- 3440 *PQBP1*-Related X-Linked Mental Retardation
- 4260 *SLC16A2*-Related X-Linked Mental Retardation
- 3500 *SYP*-Related X-Linked Mental Retardation
- 3540 *UPF3B*-Related X-Linked Mental Retardation
- 3640 *ZNF711*-Related X-Linked Mental Retardation
- 3600 *ZNF81*-Related X-Linked Mental Retardation

**Primary Ciliary Dyskinesia +/- CFTR - (EDTA Tube)**

- 8122 Primary Ciliary Dyskinesia NextGen Sequencing Panel (*DNAH5, DNAI1, DNAI2, DNAH11, TXNDC3, RSPH4A, RSPH9, DNAAF1/LRRC50, DNAAF2/c14orf104, RPGR, OFD1, CFTR*)
- 8120 Primary Ciliary Dyskinesia 61 (*DNAH5 & DNAI1* mutation panel)

**Other Genes and Syndromes - (EDTA Tube)**

- 1140 Alpha-1 Antitrypsin Deficiency (*SERPINA1* gene sequence)
- 1580 Congenital Central Hypoventilation Syndrome (*PHOX2B* gene sequence)
- 8140 IPF Telomerase (*TERT and TERC* gene sequence)
- 1540 Pulmonary Arterial Hypertension (*BMPR2* gene sequence and deletion/duplication)
- 1541 Pulmonary Arterial Hypertension (*BMPR2* deletion/duplication)
- 8100 Surfactant Panel (*ABCA3, SFTPB* and *SFTPC* gene sequence) (concurrent)
- 1300 Surfactant Deficiency (*ABCA3* gene sequence)
- 1160 Surfactant Protein B (*SFTPB* gene sequence)
- 1180 Surfactant Protein C (*SFTPC* gene sequence)

Additional Comments

**Maternal Cell Contamination - (EDTA Tube)**

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

**RETT SYNDROME - (EDTA Tube)**

- 2028 Rett Syndrome - *CDKL5* and *MECP2* gene sequence with *MECP2* del/dup (concurrent)
- 8200 Rett Syndrome - Steps 1-3 (reflex to next step when negative)
- 2020 Step 1 *MECP2* gene sequence
- 2022 Step 2 *MECP2* deletion/duplication
- 2040 Step 3 *CDKL5* gene sequence
- 2026 *MECP2* gene sequence reflex deletion/duplication

**NOONAN/LEOPARD SYNDROME - (EDTA Tube)**

- 8402 Noonan Syndrome - *PTP11, SOS1, KRAS* gene sequence and *RAF1* partial (concurrent)
- 8400 Noonan Syndrome - Steps 1 and 2 (reflex to next step when negative)
- 2280 *PTPN11* — Step 1
- 2300 *SOS1* — Step 2
- 2320 *RAF1* — Step 2
- 2340 *KRAS* — Step 2
- 8460 LEOPARD Syndrome (*PTPN11* and partial *RAF1* gene sequence)

**SPECIFIC MUTATION / GENE ANALYSIS / DEL/DUP ANALYSIS - (EDTA Tube)**

- Gene Sequence Analysis (GSA)
- Single Site-Mutation Analysis (SMA)
- Single Site-Del/Dup Analysis

Gene Name: \_\_\_\_\_ Mutation(s): \_\_\_\_\_

Gene Name: \_\_\_\_\_ Mutation(s): \_\_\_\_\_

- Positive Control Not Available
- Positive Control Sent / To Be Sent

Reporting Options  Report Amino Acid changing polymorphisms (silent polymorphisms available on request)

The following will be requested when ordering known mutation analysis for a mutation identified in an outside laboratory: 1) Proband report (mandatory) and 2) Positive Control (recommended).

ACMG guidelines, CAP, and CLIA regulatory provisions recommend use of a positive control to provide evidence of amplification when interrogating a specific sequence alteration. It is recommended that individuals for a known genotype for the locus tested be included as a positive control to ensure assay performance.