

## GENERAL TEST REQUISITION

Arrows "▶" Mandatory for Processing

Patient Information	
▶ <b>DOB</b> MM - DD - YEAR	▶ <b>Last Name</b> ▶ <b>First Name</b> M Initial
▶ <b>Gender</b> <input type="checkbox"/> F <input type="checkbox"/> M	▶ <b>Street Address, City, State, Zip</b>
▶ <b>Ethnicity</b> <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: _____	▶ <b>Home Phone</b> Work/Cell
▶ <b>Specimen Collection Date:</b> _____ 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
▶ <b>Facility Name and Address</b> ID#	
Additional Results Recipient	
Medical Professional Name:	
Facility Name and Address <input type="checkbox"/> Same As Above	
Ph	Fx
▶ <b>Form Completed by</b> ▶ <b>Phone</b>	
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. 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 <b>Medical Professional Signature</b> Mandatory for New York State X _____      Date	

▶ <b>Indication for Testing</b> <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>
▶ <b>ICD-9 Codes:</b> _____	▶ <b>Sweat Chloride:</b>

Billing Information - Mandatory For Processing			
AMBRY GENETICS provides a selection of convenient billing options. Please choose an option below and supply all requested information for your billing option. 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"/> <b>Bill Facility</b> <input type="checkbox"/> same as ordering facility	<input type="checkbox"/> <b>Bill Insurance</b> Include card copy (both sides)	<input type="checkbox"/> <b>Pre-Payment</b>	
Facility Name	Name of Insured	Relation to patient? <input type="checkbox"/> Self <input type="checkbox"/> Parent <input type="checkbox"/> Spouse	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	Insurance Company Name and Address		Card Number      Exp Date
Contact Person	Insurance Phone		Cardholder Name      Amount \$
Contact Person Phone	Member ID #	Group #	Signature X _____      Date
	Authorization #	Date	▶ <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. X _____      Date

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

Thank You for Choosing Ambry Genetics

Toll Free 866 262 7943 | Ph 949 900 5500 | Fx 949 900 5501 | www.ambrygen.com | 100 Columbia #200 | Aliso Viejo, CA 92656

**► Test Directory**
**Gene Sequence Analysis (unless otherwise indicated)**

CANCER	DYSKERATOSIS CONGENITA (DC)
<p><b>Lynch Syndrome (HNPCC)</b></p> <input type="checkbox"/> 8500 Ambry SEQUENCE™: HNPCC/Lynch Syndrome (Complete pathway, steps 1 and 2) Step 1 <i>MLH1</i> & <i>MSH2</i> sequence, Step 2 <i>MLH1MSH2/MSH6</i> del/dup, <i>MSH6</i> sequence <input type="checkbox"/> 8508 HNPCC/Lynch Syndrome, <i>MLH1</i> -Related (sequence and deletion/duplication) <input type="checkbox"/> 8510 HNPCC/Lynch Syndrome, <i>MSH2</i> -Related (sequence and deletion/duplication) <input type="checkbox"/> 8512 HNPCC/Lynch Syndrome, <i>MSH6</i> -Related (sequence and deletion/duplication) <input type="checkbox"/> 4640 HNPCC/Lynch Syndrome, <i>PMS2</i> -Related (Gene Sequence Analysis) <input type="checkbox"/> 2240 HNPCC/Lynch Syndrome, <i>TACSTD1/EPCAM</i> related (deletion/duplication) Call HNPCC/Lynch Syndrome (single gene deletion/duplication) GENE _____	<input type="checkbox"/> 8160 Ambry SEQUENCE™: DC (All Genes, steps 1-3) <input type="checkbox"/> 1960 <i>DKC1</i> - Related DC <input type="checkbox"/> 1980 <i>TINF2</i> - Related DC <input type="checkbox"/> 2120 <i>TERC</i> - Related DC Step 1 <input type="checkbox"/> 2060 <i>NHP2</i> - Related DC <input type="checkbox"/> 2080 <i>NOPT10</i> - Related DC <input type="checkbox"/> 2140 <i>TERT</i> - Related DC Step 2 Step 3
<p><b>Juvenile Polyposis Syndrome (JPS)</b></p> <input type="checkbox"/> 8602 JPS AMPLIFIED ( <i>BMPR1A</i> , <i>SMAD4</i> , Del/Dup) <input type="checkbox"/> 8600 JPS Deletion/Duplication ( <i>BMPR1A</i> , <i>SMAD4</i> ) <input type="checkbox"/> 2820 <i>BMPR1A</i> Gene Sequence Analysis <input type="checkbox"/> 1685 <i>SMAD4</i> Gene Sequence Analysis Call JPS (single gene deletion/duplication) GENE _____	<p style="text-align: center;"><b>FAMILIAL HYPERCHOLESTEROLEMIA</b></p> <input type="checkbox"/> 8680 Familial Hypercholesterolemia Comprehensive Evaluation ( <i>LDLR</i> , <i>PCSK9</i> , and partial <i>APOB</i> Sequence + <i>LDLR</i> Del/Dup) <input type="checkbox"/> 8582 Familial Hypercholesterolemia AMPLIFIED™ ( <i>LDL</i> and partial <i>APOB</i> Sequence reflex to <i>LDLR</i> Del/Dup) <input type="checkbox"/> 2780 <i>LDLR</i> Related FH Gene Sequence Analysis <input type="checkbox"/> 2784 <i>LDLR</i> Related FH Deletion/Duplication <input type="checkbox"/> 2800 <i>APOB</i> Related FH Partial Gene Analysis <input type="checkbox"/> 2804 <i>PCSK9</i> Related FH Gene Sequence Analysis
<p><b>Other Genes and Syndromes</b></p> <input type="checkbox"/> 3040 APC Amplified (FAP)(APC sequence and deletion/duplication) <input type="checkbox"/> 2866 <i>TP53</i> AMPLIFIED™ (sequence and deletion/duplication)(Li-Fraumeni Syndrome) <input type="checkbox"/> 2864 <i>TP53</i> deletion/duplication only <input type="checkbox"/> 2640 Multiple Endocrine Neoplasia Type 1 ( <i>MEN1</i> ) <input type="checkbox"/> 2680 Ambry SEQUENCE™: Multiple Endocrine Neoplasia Type 2 (MEN2)( <i>RET</i> ) <input type="checkbox"/> 2684 Step 1only: exons 10,11,13-16 <input type="checkbox"/> 2360 <i>PALB2</i> -Related Pancreatic Cancer <input type="checkbox"/> 2100 <i>PTEN</i> -Related Disorders Gene Sequence Analysis <input type="checkbox"/> 2104 <i>PTEN</i> Deletion/Duplication <input type="checkbox"/> 2766 Peutz-Jeghers AMPLIFIED ( <i>STK11</i> sequence and deletion/duplication) <input type="checkbox"/> 2600 Von Hippel-Lindau Disease ( <i>VHL</i> )	<p style="text-align: center;"><b>GASTROENTEROLOGY</b></p> <input type="checkbox"/> 8022 Pancreatitis Panel Plus ( <i>CFTR</i> , <i>PRSS1</i> , <i>SPINK1</i> , <i>CTRC</i> ) <input type="checkbox"/> 8020 Pancreatitis Panel ( <i>CFTR</i> , <i>PRSS1</i> , <i>SPINK1</i> ) <input type="checkbox"/> 8040 Pancreatitis Amplified Panel ( <i>CFTR</i> , <i>PRSS1</i> , <i>SPINK1</i> , <i>CFTR</i> del/dup) <input type="checkbox"/> 1100 <i>PRSS1</i> <input type="checkbox"/> 1120 <i>SPINK1</i> <input type="checkbox"/> 1660 <i>CTRC</i> —Chymotrypsin C-Related Pancreatitis <input type="checkbox"/> 1840 Wilson Disease ( <i>ATP7B</i> ) <input type="checkbox"/> 1440 Shwachman-Diamond Syndrome ( <i>SBDS</i> )
<p style="text-align: center;"><b>NOTES OR CLINICAL FINDINGS</b></p>	<p style="text-align: center;"><b>GENETICS</b></p> <input type="checkbox"/> 1640 Alagille AMPLIFIED™ ( <i>JAG1</i> ) <input type="checkbox"/> 1641 Alagille Deletion/Duplication ( <i>JAG1</i> ) <input type="checkbox"/> 8620 Ambry SEQUENCE™: ALS ( <i>SOD1</i> , reflex to <i>ANG</i> , <i>FIG4</i> , <i>FUS</i> , <i>TARDBP</i> ) <input type="checkbox"/> 8622 ALS ( <i>SOD1</i> only) <input type="checkbox"/> 1320 Aminoglycoside-Related Hearing Loss ( <i>MT-RNR1</i> ) <input type="checkbox"/> 8520 Ambry SEQUENCE™: Angelman Syndrome ( <i>SNRPN</i> , <i>UBE3A</i> ) <input type="checkbox"/> 2400 Angelman Syndrome, <i>UBE3A</i> -Related <input type="checkbox"/> 2420 Angelman-like Syndrome ( <i>SLC9A6</i> ) <input type="checkbox"/> 2440 Angelman/Prader-Willi Syndrome ( <i>SNRPN</i> ) <input type="checkbox"/> 1800 Ashkenazi Jewish FlexPanel™ with all 11 conditions <input type="checkbox"/> 1804 Ashkenazi Jewish FlexPanel™ as marked below <input type="checkbox"/> Bloom <input type="checkbox"/> GSD1a <input type="checkbox"/> Canavan <input type="checkbox"/> MSUD <input type="checkbox"/> CF <input type="checkbox"/> MLIV <input type="checkbox"/> Fam. Dysaut. <input type="checkbox"/> Niemann-Pick A <input type="checkbox"/> Fanconi An.-C <input type="checkbox"/> Tay-Sachs <input type="checkbox"/> Gaucher <input type="checkbox"/> 1040 Beta Thalassemia Plus ( <i>HBB</i> ) <input type="checkbox"/> 1220 Canavan AMPLIFIED™ ( <i>ASPA</i> ) <input type="checkbox"/> 1344 Congenital Hyperinsulinism related <i>GCK</i> <input type="checkbox"/> 1370 Congenital Hyperinsulinism <i>GLUD1</i> (Hyperinsulinism-Hyperammonemia) <input type="checkbox"/> 1364 Congenital Hyperinsulinism <i>KCNJ11</i> <input type="checkbox"/> 2380 CHARGE Syndrome ( <i>CHD7</i> ) <input type="checkbox"/> 1720 Fabry Disease ( <i>GLA</i> ) <input type="checkbox"/> 1820 Gaucher Disease ( <i>GBA</i> ) <input type="checkbox"/> 1600 Glutaric Acidemia Type 1 ( <i>GCDH</i> ) <input type="checkbox"/> 1880 Glycogen Storage Disease Type Ia ( <i>G6PC</i> ) <input type="checkbox"/> 1900 Glycogen Storage Disease Type Ib ( <i>SLC37A4</i> ) <input type="checkbox"/> 2746 HAE AMPLIFIED™ ( <i>SERPING1</i> Sequence and Deletion/Duplication Analysis) <input type="checkbox"/> 2700 <i>RET</i> -Related Hirschsprung Disease (Steps 1 and 2) Step 1: exons 2,3,5,6,9,10,12,13,17, Step 2: rest of gene <input type="checkbox"/> 1940 Hunter Syndrome ( <i>IDS</i> ) <input type="checkbox"/> 2160 Hurler Syndrome ( <i>IDUA</i> ) <input type="checkbox"/> 2040 Infantile Spasms ( <i>CDKL5</i> ) <input type="checkbox"/> 1360 Neonatal Diabetes Related <i>KCNJ11</i> <input type="checkbox"/> 1620 Neonatal Diabetes Related <i>INS</i> <input type="checkbox"/> 1348 Neonatal Diabetes Related <i>GCK</i> <input type="checkbox"/> 1860 Niemann-Pick Disease Types A & B ( <i>SMPD1</i> )
<p style="text-align: center;"><b>CHROMOSOMAL MICROARRAY ANALYSIS (aCGH)</b></p> <input type="checkbox"/> 3002 Ambry CMA: 180K Oligo Array	
<p style="text-align: center;"><b>CHROMOSOME STUDIES</b></p> <input type="checkbox"/> 3660 High Resolution Chromosomes <input type="checkbox"/> 3662 High Resolution Chromosomes, Rule Out Mosaic <input type="checkbox"/> 3664 Routine Blood Chromosomes <input type="checkbox"/> 3666 Routine Blood Chromosomes, Rule Out Mosaic <input type="checkbox"/> 3668 Solid Tissue Chromosomes	
<p style="text-align: center;"><b>CYSTIC FIBROSIS (CFTR) &amp; PULMONOLOGY</b></p> <input type="checkbox"/> 1002 508 FIRST™ (deltaF508 screen, reflex to CF AMPLIFIED™) <input type="checkbox"/> 1012 508 ONLY™ (deltaF508 mutation only) <input type="checkbox"/> 1006 CF AMPLIFIED™ <input type="checkbox"/> Report PolyT / TG Repeat Status <input type="checkbox"/> 1000 CF Gene Sequence Analysis <input type="checkbox"/> 1004 CF Deletion/Duplication <input type="checkbox"/> 1010 CF TG Repeat Analysis (Poly T Variant & TG Repeat) <input type="checkbox"/> 1140 Alpha-1 Antitrypsin Deficiency ( <i>SERPINA1</i> ) <input type="checkbox"/> 1580 Congenital Central Hypoventilation Syndrome ( <i>PHOX2B</i> ) <input type="checkbox"/> 8140 IPF Telomerase ( <i>TERT</i> , <i>TERC</i> ) <input type="checkbox"/> 8120 PCD 61 ( <i>DNAH5</i> & <i>DNAI1</i> Mutation Panel) <input type="checkbox"/> 1540 PAH AMPLIFIED™ – <i>BMPR2</i> related Pulmonary Arterial Hypertension) <input type="checkbox"/> 1541 PAH Deletion/Duplication ( <i>BMPR2</i> ) <input type="checkbox"/> 1300 <i>ABCA3</i> Related Surfactant Deficiency <input type="checkbox"/> 1160 Surfactant Protein B ( <i>SFTPB</i> ) <input type="checkbox"/> 1180 Surfactant Protein C ( <i>SFTPC</i> )	
<p style="text-align: center;"><b>DIAMOND-BLACKFAN ANEMIA (DBA)</b></p> <input type="checkbox"/> 8540 Ambry SEQUENCE™: DBA (All Genes, steps 1-3) <input type="checkbox"/> 2560 <i>RPS19</i> -Related DBA — Step 1 <input type="checkbox"/> 2460 <i>RPL5</i> -Related DBA — Step 2 <input type="checkbox"/> 2480 <i>RPL11</i> -Related DBA — Step 2 <input type="checkbox"/> 2500 <i>RPL35A</i> -Related DBA — Step 2 <input type="checkbox"/> 2588 <i>RPS26</i> -Related DBA — Step 2 <input type="checkbox"/> 2584 <i>RPS10</i> -Related DBA — Step 3 <input type="checkbox"/> 2580 <i>RPS24</i> -Related DBA — Step 3 <input type="checkbox"/> 2540 <i>RPS17</i> -Related DBA — Step 3 <input type="checkbox"/> 2520 <i>RPS7</i> -Related DBA — Step 3	



## Patient Consent Molecular Genetic Testing

**Test Purpose:** The purpose of this molecular genetic test is to ascertain if I am, **my child** is, or **my unborn child** is [please circle appropriate] carrying mutation(s) predisposing to or causing the specific disease or condition: \_\_\_\_\_.  
A supplemental disease description sheet is available from Ambry Genetics.

**Test Method:** The blood, body fluid, or tissue specimen submitted is required for isolation and purification of DNA for molecular genetic testing. The test will cover all disorders requested on the Ambry Genetics requisition form.

**Test Results:** I understand that due to the complexity of DNA based testing and the important implications of the test results, these results will be reported only through the patient's designated physician(s) or genetic counselor (where allowed) and that I must contact my provider to obtain the results of the test. The test results, in addition, could be released to all who, by law, may have access to such data.

I understand that if results of the molecular genetics tests are **positive**, I may be a carrier of, predisposed to, or have the specific disease or condition tested for and I may want to consider further independent testing, consult with my physician, or pursue genetic counseling. I understand that if results of the molecular genetics tests are **negative**, I may not be a carrier of, predisposed to, or have the specific disease or condition tested for and I may want to consider further independent testing, consult with my physician, or pursue genetic counseling. I understand the limitations of these results: the test results could be based upon probabilities, and may not provide a 100% definitive conclusion to either genetic disease predisposition or manifestations. I understand that the molecular genetic test may not generate results and that an additional blood, body fluid, or tissue sample may be needed to obtain accurate results. I understand that the molecular genetic test may not generate accurate results for the following reasons: sample mix-up, samples unavailable from critical family members, maternal contamination of prenatal samples, inaccurate reporting of family relationships, or technical problems, but not limited to these.

**Ambry's Rights:** Ambry reserves the right to: 1) suggest additional molecular testing if it would help in resolving the patient's clinical genotyping, 2) report additional testing results (other than requested) if they are clinically relevant to the patients and their families, and 3) refuse testing if one of the conditions in the Patient Consent form is not met.

**Use of Specimens:** After testing is completed, I understand that my blood, body fluid or tissue specimens may be disposed of or retained indefinitely for research, test validation, and/or education by Ambry Genetics, as long as my privacy is maintained. I understand that no compensation will be given nor will funds be forthcoming due to any invention(s) resulting from research and development using the specimens submitted. I understand that I may refuse to submit my specimen for use in this way and may withdraw my consent at anytime by contacting the medical director. I understand that my refusal to consent to medical research will not affect my results. Indicate consent or denial below. If a box is not marked consent is implied.

I consent to the use of my sample for research.     YES     NO

**Financial Responsibility:** I understand that if test cancellations are received prior to test set-up, processing will be honored at no charge. I understand that when requests for test cancellation are received after set-up, a cancellation report will be generated and a set-up fee will be charged. Once testing is initiated cancellation is not possible. I understand that I am responsible for all charges for testing and will be contacted for payment in the event my health plan does not reimburse for the test or Ambry Genetics does not receive a response from my health plan in a reasonable length of time.

I have read or have had read to me all of the above statements and understand the information regarding molecular genetics testing and have had the opportunity to ask questions I might have about the testing, the procedure, the risks, and the alternatives prior to my informed consent. I agree to have the molecular genetic testing.

**Patient Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

**Patient Name (please print):** \_\_\_\_\_