

Patient Information					
<b>▶ Last Name</b>	<b>▶ First Name</b>	<b>▶ M.I</b>	<b>▶ Date of Birth</b>	<b>▶ Gender</b> <input type="checkbox"/> M <input type="checkbox"/> F	
<b>▶ Street Address, City, State, ZIP and Phone Number</b>			<b>▶ ICD-9</b>	<b>▶ Indication for Testing</b> <input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier Screen <input type="checkbox"/> Family History <input type="checkbox"/> Other: _____	
Insurance Information					
<b>▶ Name of Insured</b>			<b>▶ Member ID#</b>		
<b>▶ Insurance Company Name, Address and Phone Number</b>				<b>▶ Policy Type</b> <input type="checkbox"/> HMO <input type="checkbox"/> Medicare <input type="checkbox"/> PPO <input type="checkbox"/> Medicaid <input type="checkbox"/> EPO <input type="checkbox"/> POS <input type="checkbox"/> Other: _____	
Family History			Patient History		
Please list any family history			Please list any patient history		
Contact and Organization Information					
Contact Preference <input type="checkbox"/> Fax <input type="checkbox"/> Email <input type="checkbox"/> Phone					
Select a test from the next page(s) or write in the test code/test name here. Test codes can be found at <a href="http://www.ambrygen.com/test-codes.html">www.ambrygen.com/test-codes.html</a>					
Test Code: _____ Test Name: _____					
Test Code: _____ Test Name: _____					
Test Code: _____ Test Name: _____					
Test(s) Requested					
<b>▶ Client / Institutional Name</b>		<b>▶ Form Completed By</b>		<b>▶ Return Fax #</b>	<b>▶ Return Phone #</b>
<b>▶ Authorized Ordering Physician</b>			<b>▶ E-Mail</b>		<b>▶ NPI#</b>
Please complete this form and fax to 949-900-5501 with a copy of the patient's insurance card. We will respond to you as soon as possible. For any assistance, please call 949-900-5500 ext# 362 or email <a href="mailto:preverification@ambrygen.com">preverification@ambrygen.com</a>					
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.					


**► Neurology / Intellectual Disability Test Directory Preverification Form**

- This section (EDTA Tube)
- 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)
  - 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)
  - 1820 Gaucher Disease (*GBA* gene sequence)
  - 1600 Glutaric Acidemia Type 1 (*GCDH* gene sequence)
  - 1880 Glycogen Storage Disease Type Ia (*G6PC* gene sequence)
  - 1900 Glycogen Storage Disease Type Ib (*SLC37A4* gene sequence)
  - 1940 Hunter Syndrome (*IDS* gene sequence)
  - 2160 Hurler Syndrome (*IDUA* gene sequence)
  - 1860 Niemann-Pick Disease Types A & B (*SMPD1* gene sequence)
  - 1760 Phenylketonuria - PKU (*PAH* gene sequence)
  - 1740 Pompe Disease (*GAA* gene sequence)
  - 2106 *PTEN* gene sequence and deletion/duplication
  - 2180 Smith-Lemli-Opitz Syndrome (*DHCR7* gene sequence)
  - 1240 Tay-Sachs (*HEXA* gene sequence)
  - 1560 Transthyretin Amyloidosis (*TTR* gene sequence)
  - 1840 Wilson Disease (*ATP7B* gene sequence)

**CHROMOSOMAL MICROARRAY ANALYSIS (aCGH)**

- 3002 Ambry CMA: 180K Oligo Array (1 EDTA + 1 Na Heparin)  
Note: This CMA has increased coverage on X chromosome

**CHROMOSOME STUDIES**

- 3660 High Resolution Chromosome Analysis/Karyotype (Na Heparin)
- 3662 High Resolution Chromosome Analysis/Karyotype, Rule Out Mosaic (Na Heparin)
- 3664 Routine Chromosome Analysis/Karyotype (Na Heparin)
- 3666 Routine Chromosome Analysis/Karyotype, Rule Out Mosaic (Na Heparin)
- 3668 Solid Tissue Chromosome Analysis/Karyotype (fresh tissue in RPM)

**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* gene sequence — Step 1
- 2300 *SOS1* gene sequence — Step 2
- 2320 *RAF1* gene sequence — Step 2
- 2340 *KRAS* gene sequence — Step 2
- 8460 LEOPARD Syndrome (*PTPN11* and partial *RAF1* gene sequence)

**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

**Additional Clinical Information**
**XLMR (See Specimen Requirements)**

- 8630 **XLMR Evaluation** Steps 1 and 2 (reflex to next step when negative)  
Step 1 Ambry CMA: 180K Oligo Array (1 EDTA + 1 Na Heparin)  
Note: This CMA has increased coverage on X chromosome  
Step 2 XLMR Next Gen SuperPanel™ (sequencing panel for 81 genes) (1 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 (1 EDTA + 1 Na Heparin)  
Step 2 Ambry CMA: 180K Oligo Array (1 EDTA + 1 Na Heparin)  
Note: This CMA has increased coverage on X chromosome  
Step 3 XLMR Next Gen SuperPanel™ (sequencing panel for 81 genes) (1 EDTA)
  - 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)
- To order any test in a different order, select that test above and write in the sequence order  
Note: Multiple tests require multiple samples from each patient.
- |   |  |
|---|--|
| <input type="checkbox"/> 3180 <i>CASK</i> -Related XLMR   | <input type="checkbox"/> 3500 <i>SYP</i> -Related XLMR     |
| <input type="checkbox"/> 3140 <i>ARX</i> -Related XLMR    | <input type="checkbox"/> 4400 <i>ATRX</i> -Related XLMR    |
| <input type="checkbox"/> 3220 <i>CUL4B</i> -Related XLMR  | <input type="checkbox"/> 4260 <i>SLC16A2</i> -Related XLMR |
| <input type="checkbox"/> 3380 <i>NLGN3</i> -Related XLMR  | <input type="checkbox"/> 3440 <i>PQBP1</i> -Related XLMR   |
| <input type="checkbox"/> 3400 <i>NLGN4</i> -Related XLMR  | <input type="checkbox"/> 3540 <i>UPF3B</i> -Related XLMR   |
| <input type="checkbox"/> 4780 <i>L1CAM</i> -Related XLMR  | <input type="checkbox"/> 3600 <i>ZNF81</i> -Related XLMR   |
| <input type="checkbox"/> 3640 <i>ZNF711</i> -Related XLMR |  |

**Additional Clinical Information**

Required for complimentary family studies, if indicated

Please attach pedigree / clinical consultation notes, if available

**Intellectual delay/mental retardation**

 ID/MR  mild  moderate  severe  profound Overall IQ: \_\_\_\_\_

 Verbal Aptitude  normal  mild deficiency  
 moderate deficiency  non-verbal

 Autism  no autistic behaviors  
 autistic behaviors (describe): \_\_\_\_\_

Dysmorphic features (describe): \_\_\_\_\_

Congenital anomalies (describe): \_\_\_\_\_

 History of Seizures  No  Yes  diagnosed epilepsy

**Previous Studies**

MRI/CT studies (findings): \_\_\_\_\_

Chromosome analysis: \_\_\_\_\_

Microarray analysis: \_\_\_\_\_

Other molecular studies: \_\_\_\_\_

**Growth indices**

Head circumference: \_\_\_\_\_ % Weight: \_\_\_\_\_ % Height: \_\_\_\_\_ %

**SPECIFIC MUTATION / GENE ANALYSIS**

- 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)