

Patient Information			
▶ Last Name	▶ First Name	▶ M.I	▶ Date of Birth
			▶ Gender <input type="checkbox"/> M <input type="checkbox"/> F
▶ Street Address, City, State, ZIP and Phone Number		▶ ICD-9	▶ Indication for Testing <input type="checkbox"/> Diagnostic <input type="checkbox"/> Carrier Screen <input type="checkbox"/> Family History <input type="checkbox"/> Other: _____
Insurance Information			
▶ Name of Insured		▶ Member ID#	
▶ Insurance Company Name, Address and Phone Number			▶ Policy Type <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 www.ambrygen.com/test-codes.html			
Test Code: _____		Test Name: _____	
Test Code: _____		Test Name: _____	
Test Code: _____		Test Name: _____	
Test(s) Requested			
▶ Client / Institutional Name	▶ Form Completed By	▶ Return Fax #	▶ Return Phone #
▶ Authorized Ordering Physician		▶ E-Mail	▶ NPI#
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 preverification@ambrygen.com			
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.			

► Test Directory

Preverification 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 (*SERPING1* 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

Preverification 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* —
- 2320 *RAF1* — Step 2
- 2340 *KRAS* —
- 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.