Ambry Genetics™

15 Argonaut,
Aliso Viejo, CA 92656

Toll Free 866 262 7943
Fax 949 900 5501
ambrygen.com

General Test Requisition (All Blue Highlighted Fields Required)

ORDERING CHECKLIST

- ☐ Letter of Medical Necessity (Recommended)
- ☐ ICD-9 Codes
- ☐ Clinician & Patient Signatures
- ☐ Copy Patient Insurance Card

PATIENT INFORMATION

Last Name	First Name	Middle Initial	DOB (MM/DD/YY)	Date of Death (if applicable)

Street Address	City	State	Zip

Preferred Contact Phone Number	Gender	Ethnicity
	☐F ☐M ☐Unknown	☐African American/Black ☐Asian ☐Caucasian ☐Hispanic ☐Jewish (Ashkenazi) ☐Specify:_____

ORDERING PHYSICIAN

Authorized Ordering Physician	Facility Name	NPI#

Street Address	City	State	Zip

Phone	Fax	Ordering Physician E-mail

ADDITIONAL RESULTS RECIPIENTS

Medical Professional Name	Phone	E-mail/Fax

Medical Professional Name	Phone	E-mail/Fax

Form Completed By	Phone	Fax	E-mail

CONFIRMATION OF INFORMED CONSENT FOR GENETIC TESTING

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 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. Furthermore, additional results recipients information is true and correct to the best of my knowledge.

Does this patient give consent to the use of their sample for research? ☐ Yes ☐ No Consent is implied if a box is not marked (For patients in NY State, research consent will NOT be implied if left blank).

Medical Professional Signature _____ Date: _____

(MD/DO, Clinical Nurse Specialist, Nurse-Midwives, Nurse Practitioner, Physician Assistant) Mandatory for Medicare/Medicaid

☐ INSURANCE BILLING (INCLUDE COPY OF BOTH SIDES OF INSURANCE CARD)

A completed Advance Beneficiary Notice (ABN) of coverage is required for Medicare patients who do not meet medical criteria for testing (see website for form)

Name of Insured	Relation to Patient? ☐Self ☐Parent ☐Spouse	Insurance Company Name

Street Address	City	State	Zip

Insurance Phone	Member ID	Group #

Authorization #	Authorization Date

PAYMENT INFORMATION / PREPAYMENT

Payment Type:
☐Check (Payable to Ambry Genetics) ☐Visa ☐Mastercard ☐American Express ☐Discover

Card Number	Exp. Date	CVC #

Cardholder Name	Amount $

Signature	Date

☐ INSTITUTIONAL BILLING

Facility Name

Street Address

City	State	Zip Code

Contact Name

Phone Number

NOTES

Ambry may require credit card information for insurance samples prior to initiating testing (does not apply to Medicaid and Medicare). Your patient will be notified if this information is needed.

Complete and detailed clinical information on page 2 provides a clear indication for testing (i.e. medical necessity) and assists in determination of insurance coverage.

Ambry recommends a patient-specific letter of medical necessity (LMN), as most insurance carriers will require one for processing (a LMN is not required for Medicare patients that meet medical guidelines).

Out-of-Pocket Expense Policy: Ambry Genetics will contact the above patient if the out-of-pocket (OOP) amount for this test order is estimated to exceed $300.

For any tests priced under $400, pre-verification of insurance coverage will not be performed.

Patient Acknowledgement: I acknowledge that the information provided by me is true to the best of my knowledge. For direct insurance/3rd party billing: 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. If applicable, I authorize Ambry Genetics Corporation to be my Designated Representative for purposes of appealing any denial of benefits. I understand that I am financially responsible for any amounts not covered by my insurer for this test order.

I also fully understand that I am legally responsible for sending Ambry Genetics any money received from my health insurance company for performance of this genetic test. Failing to do so will result in my account being sent to collection.

Patient Signature: _____ Date: _____

General Test Requisition

Patient Name_____

SPECIMEN INFORMATION (HIGHLIGHTED FIELDS ARE REQUIRED)

Collection Date	Specimen ID	MRN

Specimen Type

☐ Blood (EDTA Preferred) ☐ Saliva (adult) ☐ Saliva (pediatric) ☐ DNA ☐ Na Heparin (chromosomes only)

☐ Blood spots ☐ Amnio/CVS ☐ Other _____

ICD-9 CODES (CHECK ALL THAT APPLY) – (USE V CODES FOR SECONDARY DX)

☐ 277.00	Cystic fibrosis without meconium ileus	☐ 577.1	Chronic pancreatitis	☐ 783.42	Delayed milestones
☐ 299.00	Autism, current infantile or childhood	☐ 742.1	Microcephaly	☐ V18.9	Genetic disease carrier
☐ 314.01	Attention deficit hyperactivity disorder	☐ 756.0	Anomalies of skull and face bones	☐ V26.31	Female genetic disease carrier
☐ 319	Unspecified intellectual disabilities	☐ 759.9	Congenital anomaly unspecified	☐ V26.34	Male genetic disease carrier
☐ 345.9	Epilepsy unspecified	☐ 781.3	Hypotonia, hypotonicity	☐ V82.71	Scr for genetic disease carrier status
☐ 448.0	Hereditary hemorrhagic telangiect	☐ 783.41	Failure to thrive	☐ Others	_____

INDICATIONS FOR TESTING ARE NOT MUTUALLY EXCLUSIVE, PLEASE SELECT ALL THAT APPLY.

☐ Diagnostic ☐ Carrier screening ☐ Research ☐ Family history

☐ Positive or normal control ☐ Other_____

☐ ID/DD without syndromic features (intellectual disability/develop delay)

☐ Autism spectrum disorder

☐ Neurologic- including senorineural:

visions, hearing, etc. ☐ Childhood onset ☐ Adult onset

☐ Cardiopulmonary ☐ Childhood onset ☐ Adult onset

☐ Psychiatric

☐ Immunologic/infectious/CF

☐ Hematologic

☐ Musculoskeletal/malformations/birth defects

☐ Endocrine/metabolic

☐ Renal

☐ Positive newborn screen

☐ Genetic syndrome

FAMILY HISTORY

Relation to Patient	Hx of Disease	Age at Diagnosis
Maternal (mother's side)		
Paternal (father's side)		
Additional/Other (siblings/children)		

PRENATAL SAMPLES ONLY (CLINICAL OR ULTRASOUND FINDINGS)

LMP/EDC	Gestational Age (weeks/days)	NT (mm)

Ultrasound Findings

_____

_____

_____

OTHER CLINICAL FINDINGS

ex. (sweat chloride levels, positive newborn screen)

_____

_____

_____

_____

_____

NEUROLOGY/ID SAMPLES ONLY (PLEASE ATTACH PEDIGREE / CLINICAL CONSULTATION NOTES, IF AVAILABLE)

Intellectual Delay/Intellectual Disability ☐ 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 ☐ Yes ☐ No ☐ diagnosed epilepsy

Previous Studies

MRI/CT studies (findings): _____

Chromosome analysis: _____

Microarray analysis: _____

Other molecular studies:_____

Growth Indices

Head circumference: _____% Weight: _____% Height: _____ %

General Test Requisition

Patient Name_____

MATERNAL CELL CONTAMINATION (REQUIRED FOR FETAL SPECIMENS)

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

HEREDITARY CANCER PANELS

☐ *8820* **BreastNext:** next-generation sequencing panel of 14 genes associated with increased risk for breast cancer *(ATM, BARD1, BRIP1, CDH1, CHEK2, MRE11A, MUTYH, NBN, PALB2, PTEN, RAD50, RAD51C, STK11, TP53)*

☐ *8822* **ColoNext:** next-generation sequencing panel of 14 genes associated with increased risk for colon cancer *(APC, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PTEN, PMS2, SMAD4, STK11, TP53)*

☐ *8824* **CancerNext:** next-generation sequencing panel of 22 genes associated with increased risk for hereditary cancers *(APC, ATM, BARD1, BRIP1, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, SMAD4, STK11, TP53)*

☐ *8830* **OvaNext:** next-generation sequencing panel of 19 genes associated with increased risk for breast, ovarian, and/or uterine cancers *(ATM, BARD1, BRIP1, CDH1, CHEK2, EPCAM, MRE11A, MLH1, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, STK11, TP53)*

CANCER TEST MENU

Lynch Syndrome (HNPCC)

☐ 8518 HNPCC/Lynch Syndrome - *MLH1, MSH2 & MSH6* gene sequence with *MLH1, MSH2, MSH6 and EPCAM* deletion/duplication (all concurrent)
☐ 8500 HNPCC/Lynch Syndrome - Steps 1 and 2 (reflex option)
Step 1 *MLH1 & MSH2* gene sequence
Step 2 *MSH6* gene sequence and *MLH1/MSH2/MSH6/EPCAM* deletion/duplication
☐ 4646 *PMS2* gene sequence and deletion/duplication
☐ Run reflex to 8518 or 8500
☐ 8508 *MLH1* gene sequence and deletion/duplication
☐ 8510 *MSH2* gene sequence and deletion/duplication + *EPCAM* deletion/duplication
☐ 8512 *MSH6* gene sequence and deletion/duplication
☐ 2240 *EPCAM* deletion/duplication
☐ 2246 *MSH6* deletion/duplication
☐ 8512 *MSH6* gene sequence and deletion/duplication
☐ 8519 *EPCAM* deletion/duplication
☐ 8519 *MLH1* deletion/duplication
☐ 8519 *MSH2* deletion/duplication

Familial Adenomatous Polyposis Syndrome (FAP)
☐ 3040 *APC* gene sequence and deletion/duplication
☐ 4660 *MUTYH* gene sequence
☐ 8726 FAP - APC and *MUTYH* gene sequence and *APC* deletion/duplication (concurrent)

Juvenile Polyposis Syndrome (JPS)
☐ 1685 *SMAD4* gene sequence
☐ 1694 *SMAD4* deletion/duplication
☐ 1694 *BMPR1A* deletion/duplication
☐ 2820 *BMPR1A* gene sequence
☐ 8604 *BMPR1A and SMAD4* gene sequence and deletion/duplication (concurrent)
☐ 8602 JPS - Steps 1 and 2
Step 1 *BMPR1A & SMAD4* gene sequence
Step 2 *BMPR1A & SMAD4* deletion/duplication
☐ 8600 *BMPR1A and SMAD4* deletion/duplication

Other Genes and Syndromes
☐ 2106 *PTEN* gene sequence and deletion/duplication
☐ 2360 *PALB2* gene sequence (Pancreatic Cancer)
☐ 2606 *VHL* gene sequence and deletion/duplication (Von Hippel-Lindau Disease)
☐ 2680 *RET* gene sequence (Multiple Endocrine Neoplasia Type 2)
☐ 2766 *STK11* gene sequence and deletion/duplication (Peutz-Jeghers syndrome)

CANCER TEST MENU CONTINUED

☐ 2866 *TP53* gene sequence and deletion/duplication (Li-Fraumeni Syndrome)
☐ 4982 *CHEK2* specific mutation analysis for 1100delC - Founder Mutation (Related Cancer)
☐ 5260 *DICER1* gene sequence (Pleuropulmonary blastoma & related cancers)
☐ 5684 *PTCH1* gene sequence and deletion/duplication (Gorlin syndrome/ holoprosencephaly)
☐ 5620 *RAD51D* gene sequence
☐ 5426 *RB1* gene sequence and deletion/duplication
☐ 4726 *CDH1* gene sequence and deletion/duplication
☐ 4700 *CDKN2A*($p16^{INK4a}$)/*ARF*($p14^{ARF}$) Gene Sequence Analysis (Malignant Melanoma)
☐ 4980 *CHEK2* gene sequence Analysis
☐ 4984 *CHEK2* gene sequence with exon 9-10 deletion (Related Cancer)
☐ 2646 *MEN1* gene sequence and deletion/duplication (Multiple Endocrine Neoplasia Type 1)
☐ 5602 *ATM* gene sequence (Related Cancer)

Paraganglioma-Pheochromocytoma Syndrome (PGL/PCC)
☐ 2606 *VHL* gene sequence and deletion/duplication (Von Hippel-Lindau Disease)
☐ 2680 *RET* gene sequence (Multiple Endocrine Neoplasia Type 2)
☐ 5380 *SDHB* gene sequence
☐ 5386 *SDHC* gene sequence
☐ 5392 *SDHD* gene sequence
☐ 5398 *SDHAF2* gene sequence
☐ 5410 *TMEM127* gene sequence
☐ 5416 PGL/PCC - *SDHB, SDHC, SDHD, SDHAF2* deletion/duplication
☐ 5419 PGL/PCC - SDHB, SDHC, SDHD, SDHAF2 gene sequence and deletion/duplication and *MAX, TMEM127* gene sequence (concurrent)
☐ 5500 *SDHA* gene sequence
☐ 5520 *MAX* gene sequence

CHROMOSOMAL MICROARRAY ANALYSIS

☐ 3002 Ambry CMA: 180K Oligo Array
☐ 5480 SNP + CGH Array
Note: These microarrays have increased coverage on X chromosome

BLOOD 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)
☐ 5220 Y Chromosome Microdeletion Analysis

CEREBRAL CAVERNOUS MALFORMATIONS (CCM)

☐ 5368 CCM - Steps 1 and 2 (reflex option)
Step 1 *KRIT1* gene sequence
Step 2 *CCM2* and *PDCD10* gene sequence with *CCM2, KRIT1* and *PDCD10* del/dup
☐ 5370 CCM - All genes listed below for gene sequence and del/dup (concurrent)
☐ 5320 *CCM2* gene sequence ☐ 5324 *CCM2* deletion/duplication
☐ 5340 *KRIT1* gene sequence ☐ 5344 *KRIT1* deletion/duplication
☐ 5360 *PDCD10* gene sequence ☐ 5364 *PDCD10* deletion/duplication
☐ 5366 CCM - All genes for del/dup

DIAMOND-BLACKFAN ANEMIA (DBA)

☐ 8548 DBA—*RPS19, RPL5, RPL11, RPL35A, RPS26, RPS10, RPS24, RPS17, RPS7* gene sequence (concurrent)
☐ 8540 DBA - Steps 1 through 3 (reflex option)
Step 1 *RPS19* gene sequence
Step 2 *RPL5, RPL11, RPL35A, RPS26* gene sequence
Step 3 *RPS10, RPS24, RPS17, RPS7* gene sequence
☐ 2560 *RPS19* gene sequence ☐ 2460 *RPL5* gene sequence
☐ 2480 *RPL11* gene sequence ☐ 2500 *RPL35A* gene sequence
☐ 2588 *RPS26* gene sequence ☐ 2584 *RPS10* gene sequence
☐ 2580 *RPS24* gene sequence ☐ 2540 *RPS17* gene sequence
☐ 2520 *RPS7* gene sequence ☐ 5080 *RPL19* gene sequence
☐ 5100 *RPL26* gene sequence

General Test Requisition

Patient Name_____

DYSKERATOSIS CONGENITA (DC)

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

CARDIOMYOPATHIES AND CARDIO CHANNELOPATHIES

☐ 8840 Pan Cardio Panel: next-generation sequencing panel of 79 genes associated with cardiomyopathies, channelopathies, and structural heart defects
(ABCC9, ACTC1, ACTN2, AKAP9, ANK2, ANKRD1, BAG3, CACNA1C, CACNA2D1, CACNB2, CALR3, CASQ2, CAV3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EMD, EYA4, FXN, GATA4, GLA, GPD1L, ILK, JAG1, JPH2, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ8, KCNQ1, LAMP2, LDB3/ZASP, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2.5, PDLIM3, PKP2, PLN, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SGCD, SNTA1, TAZ, TBX1, TBX5, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL)
☐ 8842 Brugada Syndrome Panel: next-generation sequencing panel of 9 genes
(CACNA1C, CACNA2D1, CACNB2, GPD1L, KCNE3, KCNJ8, SCN1B, SCN3B, SCN5A)
☐ 8844 DCM Panel: next-generation sequencing panel of 37 genes
(ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DMD, EMD, EYA4, ILK, LAMP2, LDB3/ZASP, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEBL, NEXN, PDLIM3, PLN, RBM20, SCN5A, SGCD, TAZ, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL)
☐ 8846 HCM Panel: next-generation sequencing panel of 31 genes
(ACTC1, ACTN2, ANKRD1, CALR3, CAV3, CSRP3, DES, FXN, GLA, JPH2, LAMP2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOM1, MYOZ2, NEXN, PLN, PRKAG2, PTPN11, RAF1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL)
☐ 8848 Long QT Syndrome Panel: next-generation sequencing panel of 12 genes
(AKAP9, ANK2, CACNA1C, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN4B, SCN5A, SNTA1)
☐ 8850 Arrhythmia Panel: next-generation sequencing panel of 29 genes associated with Long QT, Brugada, CPVT and/or ARVD/C
(AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CASQ2, CAV3, DES, DSC2, DSG2, DSP, GPD1L, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ8, KCNQ1, LMNA, PKP2, RYR2, SCN1B, SCN3B, SCN4B, SCN5A, SNTA1, TMEM43)
☐ 8852 Cardiomyopathy Panel: next-generation sequencing panel of 56 genes associated with cardiomyopathies, including DCM, HCM, and ARVD/C
(ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CALR3, CAV3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, EMD, EYA4, FXN, GLA, ILK, JPH2, JUP, LAMP2, LDB3/ZASP, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOM1, MYOZ2, MYPN, NEBL, NEXN, PDLIM3, PKP2, PLN, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, SGCD, TAZ, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, TXNRD2, VCL)

FAMILIAL HYPERCHOLESTEROLEMIA

☐ 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

☐ 1100 *PRSS1* gene sequence
☐ 1120 *SPINK1* gene sequence
☐ 1440 Shwachman-Diamond syndrome (*SBDS* gene sequence)
☐ 1660 *CTRC* gene sequence
☐ 1840 Wilson Disease (*ATP7B* gene sequence)
☐ 8020 *CFTR, PRSS1, SPINK1* gene sequence (Pancreatitis)
☐ 8022 *CFTR, PRSS1, SPINK1* and *CTRC* gene sequence (Pancreatitis)
☐ 8040 *CFTR, PRSS1, SPINK1* gene sequence & *CFTR* deletion/duplication (Pancreatitis)

GENERAL GENETICS

☐ 1640 Alagille (*JAG1* gene sequence and deletion/duplication)
☐ 1641 Alagille (*JAG1* deletion/duplication)
☐ 5280 Andermann Syndrome (*SLC12A6* gene sequence)
☐ 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)

GENERAL GENETICS CONTINUED

☐ 1040 Beta Thalassemia Plus (*HBB* gene sequence with 619del check)
☐ 1220 Canavan (*ASPA* gene sequence reflex deletion/duplication)
☐ 1226 Canavan (*ASPA* gene sequence and deletion/duplication) (concurrent)
☐ 1240 Tay-Sachs Plus (*HEXA* gene sequence)
☐ 1360 Neonatal Diabetes (*KCNJ11* gene sequence)
☐ 1364 Congenital Hyperinsulinism (*KCNJ11* gene sequence)
☐ 1370 Congenital Hyperinsulinism-Hyperammonemia (*GLUD1* gene sequence)
☐ 1560 Transthyretin Amyloidosis (*TTR* gene sequence)
☐ 1600 Glutaric Acidemia Type 1 (*GCDH* gene sequence)
☐ 1620 Neonatal Diabetes (*INS* gene sequence)
☐ 1720 Fabry Disease (*GLA* gene sequence)
☐ 1740 Pompe Disease (*GAA* gene sequence)
☐ 1760 Phenylketonuria - PKU (*PAH* gene sequence)
☐ 1820 Gaucher Disease (*GBA* gene sequence)
☐ 1860 Niemann-Pick Disease Types A & B (*SMPD1* 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)
☐ 2180 Smith-Lemli-Opitz Syndrome (*DHCR7* gene sequence)
☐ 2380 CHARGE Syndrome (*CHD7* gene sequence)
☐ 2400 Angelman Syndrome (*UBE3A* gene sequence)
☐ 2420 Angelman-like Syndrome (*SLC9A6* gene sequence)
☐ 2440 Angelman/Prader-Willi Syndrome (*SNRPN* methylation)
☐ 2700 Hirschsprung Disease Steps 1 and 2 (*RET*)
Step 1 only: exons 2,3,5,6,9,10,12,13,17 gene sequence
Step 2 only: rest of gene sequence
☐ 2708 Hirschsprung Disease (*RET* gene sequence) (concurrent)
☐ 2746 Hereditary Angioedema (*SERPING1* gene sequence and deletion/duplication)
☐ 3200 Infantile Spasms (*CDKL5* gene sequence)
☐ 3753 Spinal Muscular Atrophy (SMA) Carrier Test (Deletion Analysis)
1 Extra EDTA tube Required (3-5cc)
☐ 4680 Niemann-Pick Disease Types C1 (*NPC1* gene sequence)
☐ 4684 Niemann-Pick Disease Types C2 (*NPC2* gene sequence)
☐ 4840 Rhizomelic Chondrodysplasia Punctata Type 1 (*PEX7* gene sequence)
☐ 4860 Lysosomal Free Sialic Acid-Storage (Salla) Diseases (*SLC17A5* gene sequence)
☐ 4880 Glutathione Synthetase Deficiency (*GSS* gene sequence)
☐ 4900 MCAD - Medium-chain acyl-CoA dehydrogenase (*ACADM* gene sequence)
☐ 4920 VLCAD - Very long-chain acyl-CoA dehydrogenase (*ACADVL* gene sequence)
☐ 4940 Aspartylglucosaminuria (*AGA* gene sequence)
☐ 4960 Dihydropyrimidine Deyhyrogenase Deficiency (*DPYD* gene sequence)
☐ 5000 Familial Mediterranean Fever (*MEFV* gene sequence)
☐ 5020 Hyperoxaluria Type 2 (*GRHPR* gene sequence)
☐ 5180 Mucolipidosis Type IV (*MCOLN1* gene sequence)
☐ 5240 Tay-Sachs Enzyme Assay (*HEXA* Leukocytes) (1 ACD tube required)
☐ 5602 Atexia-Telangiectasia (*ATM gene sequence*)
☐ 5700 *NF1* Full gene sequence (Neurofibromatosis type 1)
☐ 5702 *NF1* deletion/duplication (Neurofibromatosis type 1)
☐ 5704 *NF1* gene sequence reflex to deletion/duplication (Neurofibromatosis type 1)
☐ 5720 *SPRED1* gene sequence (Legius syndrome/Neurofibromatosis type 1-like syndrome)
☐ 5722 *SPRED1* deletion/duplication (Legius syndrome/Neurofibromatosis type 1-like syndrome)
☐ 5724 *SPRED1* gene sequence reflex to deletion/duplication (Legius syndrome/Neurofibromatosis type 1-like syndrome)
☐ 5728 *NF1* and *SPRED1* gene sequence and deletion/duplication (concurrent)
☐ 5730 *NF1* and *SPRED1* gene sequence and deletion/duplication (reflex)
☐ 8122 Primary Ciliary Dyskinesia NextGen Sequencing Panel
(DNAH5, DNAI1, DNAI2, DNAH11, TXNDC3, RSPH4A, RSPH9, DNAAF1, DNAAF2, RPGR, OFD1, CFTR)
☐ 8520 Angelman Syndrome (*SNRPN* methylation reflex to *UBE3A* gene sequence)

General Test Requisition

Patient Name_____

THROMBOPHILIA (1 EDTA LAVENDER TOP)

☐ 5140 Thrombophilia Flex Panel (Factor II, Factor V, *MTHFR*)
☐ 5141 Factor II (Prothrombin G20210A)
☐ 5143 Factor V (Leiden)
☐ 5145 *MTHFR* (C677T and A1298C)

HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)

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

MARFAN, ANEURSYM AND RELATED DISORDERS

☐ 5660 *FBN1* gene sequence
☐ 8780 Marfan, Aneurysm and Related Disorders Panel (concurrent) *ACTA2, CBS, FBN1, FBN2, MYH11, COL3A1, SLC2A10, SMAD3, TGFBR1, TGFBR2* gene sequence
☐ 8782 Marfan, Aneurysm and Related Disorders Panel Steps 1 and 2
Step 1 *FBN1* gene sequence
Step 2 *ACTA2, CBS, FBN2, MYH11, COL3A1, SLC2A10, SMAD3, TGFBR1, TGFBR2* gene sequence
☐ 8788 *FBN1, TGFBR1* and *TGFBR2* gene sequence

NEUROLOGY / INTELLECTUAL DISABILITY

☐ 8626 XLID Next Gen Panel™
ABCD1, ACSL4, AGTR2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CUL4B, DCX, DKC1, DLG3, FANCB, FGD1, FLNA, FMR1, FTSJ1, GDI1, GJB1, GK, GPC3, GRIA3, HCCS, HPRT1, HSD17B10, HUWE1, IDS, IL1RAPL1, KDM5C, KIAA2022, L1CAM, LAMP2, MAOA, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, OCRL, OFD1, OPHN1, OTC, PAK3, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TIMM8A, TSPAN7, UBE2A, UPF3B, ZDHHC9, ZNF41, NF674, ZNF711, ZNF81
☐ 8628 XLID 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 XLID Next-Gen Panel™ (sequencing panel for 81 genes)
☐ 8630 XLID 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 XLID Next-Gen Panel™ (sequencing panel for 81 genes)

Single Gene Sequence Analysis is also available for all 81 genes on XLID Next-Gen panels. To order single gene testing check box below and list gene name.

☐ Single Gene Sequence Analysis Gene: _____

☐ 3002 180K Oligo Array
☐ 3020 *FMR2* Fraxe DNA Analysis
☐ 3664 Routine Chromosome Analysis/Karyotype (1 Na Heparin)
☐ 4544 Fragile X DNA Analysis
☐ 5480 SNP+CGH Array
Note: These microarrays have increased coverage on X chromosome

NOONAN/LEOPARD SYNDROME

☐ 2280 *PTPN11* ☐ 2300 *SOS1*
☐ 2320 *RAF1* ☐ 2340 *KRAS*
☐ 8400 Noonan Syndrome - Steps 1 and 2 (reflex option)
Step 1 *PTPN11* gene sequence
Step 2 *SOS1, RAF1* (partial), KRAS gene sequence
☐ 8402 Noonan Syndrome - *PTPN11, SOS1, KRAS* gene sequence and *RAF1* partial (concurrent)
☐ 8460 LEOPARD Syndrome (*PTPN11* and partial *RAF1* gene sequence)

PULMONOLOGY

Cystic Fibrosis
☐ 1000 *CFTR* gene sequence
☐ 1002 508 FIRST™ (deltaF508 reflex to *CFTR* Amplified)
☐ 1004 *CFTR* deletion/duplication
☐ 1006 *CFTR* Amplified (*CFTR* gene sequence reflex deletion/duplication)
☐ Report PolyT / TG Status
☐ 1007 *CFTR* Amplified (*CFTR* gene sequence and deletion/duplication) (concurrent)
☐ Report PolyT / TG Status
☐ 1010 *CFTR* Poly T, reflex analysis to TG repeat
☐ 1012 508 ONLY™ (deltaF508 mutation only)
☐ 1018 CFTR Screening Panel (CF102)
☐ 2000 CFTR Screening Panel (CF33)

Primary Ciliary Dyskinesia
☐ 8122 Primary Ciliary Dyskinesia NextGen Sequencing Panel with CFTR sequencing *(DNAH5, DNAI1, DNAI2, DNAH11, TXNDC3, RSPH4A, RSPH9, DNAAF1, DNAAF2, RPGR, OFD1, CFTR)*

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

RETT SYNDROME

☐ 2020 *MECP2* gene sequence
☐ 2022 *MECP2* deletion/duplication
☐ 2026 *MECP2* gene sequence reflex deletion/duplication
☐ 2028 Rett Syndrome - *CDKL5* and *MECP2* gene sequence with *MECP2* deletion/duplication (concurrent)
☐ 2040 *CDKL5* gene sequence
☐ 5440 *FOXG1* gene sequence
☐ 8200 Rett Syndrome - Steps 1-3 (reflex option)
Step 1 *MECP2* gene sequence
Step 2 *MECP2* deletion/duplication
Step 3 *CDKL5* gene sequence
☐ 8202 Rett/Atypical Rett deletion/duplication (*FOXG1, MECP2, MEF2C, CDKL5)*

General Test Requisition

Patient Name_____

PREVIOUS TEST HISTORY

Previously Detected Mutations: _____ Gene Name: _____

Testing Lab: _____

Please include copy of test results if performed at another laboratory

Patient previously tested at Ambry? ☐ Yes ☐ No

Family previously tested at Ambry? ☐ Yes ☐ No

Name:_____ DOB _____ Relation: _____

Name:_____ DOB _____ Relation: _____

Name:_____ DOB _____ Relation: _____

Name:_____ DOB _____ Relation: _____

SPECIFIC MUTATION / GENE ANALYSIS / DEL/DUP ANALYSIS

☐ Full Gene Sequencing _____
☐ Specific Site Analysis
☐ Specific Gene Del/Dup Analysis

Gene Name: _____ Mutation(s): _____

Gene Name: _____ Mutation(s): _____

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

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

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

ADDITIONAL INFORMATION

_____
_____
_____
_____
_____
_____
_____
_____
_____

CONTACT US

Insurance Verification or Benefit Questions: call (949) 900-5794 or email preverification@ambrygen.com

Billing Questions: call (949) 900-5795 or email billing@ambrygen.com

General Questions: call (949) 900-5500 or toll free number (866) 262-7943 or email info@ambrygen.com

General Fax Line: call (949) 900-5501

INSURANCE BILLING

Ambry recommends submitting a patient-specific letter of medical necessity (LMN), as most insurance carriers will require one for processing (a LMN is not required for Medicare patients that meet medical guidelines). Please visit our website for a full library of customizable forms.

PRENATAL TESTING

Prenatal testing is available on a case-by-case basis for most of the conditions listed on the general test menu.

Please contact a genetic counselor at (949) 900-5500 to discuss the case prior to sending a fetal sample.

PREVERIFICATIONS

To process a preverification, please complete this requisition form (TRF), make a copy of the insurance cards (front and back) and fax to the insurance verification department at 949-900-5501