

# INSURANCE PREVERIFICATION REQUEST FORM

Arrows "▶" Mandatory for Processing

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.					

**► Test Directory**
**Preverification Form**
**CANCER**

- Lynch Syndrome (HNPCC) - (EDTA Tube)**
- 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
- Call HNPCC/Lynch Syndrome (single gene deletion/duplication) GENE \_\_\_\_\_
- Juvenile Polyposis Syndrome (JPS) - (EDTA Tube)**
- 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
  - 2820 *BMPR1A* gene sequence
  - 1685 *SMAD4* gene sequence
- Call JPS (single gene deletion/duplication) GENE \_\_\_\_\_
- Familial Adenomatous Polyposis Syndrome (FAP) - (EDTA Tube)**
- 8726 FAP - *APC* and *MUTYH* gene sequence and *APC* deletion/duplication (concurrent)
  - 8722 FAP PLUS [*APC* gene sequence and deletion/duplication + MAP (see below)]
  - 8720 *MUTYH* associated Polyposis (MAP)  
Step 1 *MUTYH* specific mutation analysis of Y179C and G396D  
Step 2 If only one mutation detected, reflex to *MUTYH* gene sequence analysis
  - 4660 *MUTYH* gene sequence
  - 3040 *APC* gene sequence and deletion/duplication
- Hereditary Diffuse Gastric Cancer - (EDTA Tube)**
- 4726 *CDH1* gene sequence and deletion/duplication
- Other Genes and Syndromes - (EDTA Tube)**
- 2866 *TP53* gene sequence and deletion/duplication (Li-Fraumeni Syndrome)
  - 2864 *TP53* deletion/duplication
  - 5260 *DICER1* gene sequence (Pleuropulmonary blastoma & related cancers)
  - 4980 *CHEK2* gene sequence with exon 9\_10 deletion (Related Cancer)
  - 4984 *CHEK2* exon 9-10 deletion (Related Cancer)
  - 4982 *CHEK2* specific mutation analysis for 1100delC - Founder Mutation (Related Cancer)
  - 2640 *MEN1* gene sequence (Multiple Endocrine Neoplasia Type 1)
  - 2680 *RET* gene sequence (Multiple Endocrine Neoplasia Type 2)
  - 4700 *CDKN2A/p16* gene sequence (Malignant Melanoma)
  - 2360 *PALB2* gene sequence (Pancreatic Cancer)
  - 2106 *PTEN* gene sequence and deletion/duplication
  - 5426 *RB1* gene sequence and deletion/duplication
  - 2766 *STK11* gene sequence and deletion/duplication (Peutz-Jeghers)
  - 2606 *VHL* gene sequence and deletion/duplication (Von Hippel-Lindau Disease)
- Testing of Tumor Characteristics (Screening Tests for HNPCC) - (Na Heparin Tube)**
- 8700(4) MMR Profile by IHC (*MLH1*, *MSH2*, *MSH6*, *PMS2*)
  - 8702 MSI by PCR
  - 7978 *MLH1* Hypermethylation Analysis
  - 7980 *BRAF* SMA V600E Analysis
  - 7982 *MLH1* Hypermethylation with *BRAF* (Analysis of V600E only) Analysis

**CEREBRAL CAVERNOUS MALFORMATIONS (CCM) - (EDTA Tube)**

- 5370 CCM - All genes listed below for gene sequence and del/dup (concurrent)
- 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
- 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

**CYSTIC FIBROSIS (CFTR) & PULMONOLOGY - (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)
- 1140 Alpha-1 Antitrypsin Deficiency (*SERPINA1* gene sequence)
- 1580 Congenital Central Hypoventilation Syndrome (*PHOX2B* gene sequence)
- 8140 IPF Telomerase (*TERT* and *TERC* gene sequence)
- 8120 Primary Ciliary Dyskinesia 61 (*DNAH5* & *DNAI1* mutation panel)
- 8122 Primary Ciliary Dyskinesia NextGen Sequencing Panel
- 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)

**DIAMOND BLACKFAN ANEMIA - (EDTA Tube)**

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

## Additional Information

**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 RPMI)

► 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)**

- 8800 Clinical Diagnostic Exome (CDE)  
Required For Testing To Proceed:  
- CDE Consent Form  
- Detailed Family & Clinical History  
- Indicate Family Members Undergoing Exome Sequencing

Name	Relationship to Proband	Affected? (Y/N)

More details can be found at [www.ambrygen.com/ClinicalDiagnosticExome.html](http://www.ambrygen.com/ClinicalDiagnosticExome.html)

**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*)
  - Maple Syrup Urine Disease (*BCKDHA/B*)
  - Maple Syrup Urine Disease Type 3 (*DLD*)
  - Mucopolidiosis 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 Mucopolidiosis 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)

Patient Name

**► Test Directory**

**Requisition Form**

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

- 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
    - 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 \_\_\_\_\_

**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

**Additional Information**

**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)

**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)

**PARAGANGLIOMA-PHEOCHROMOCYTOMA SYNDROME (PGL/PCC) - (EDTA Tube)**

- 5418 PGL/PCC - *SDHB*, *SDHC*, *SDHD*, *SDHAF2* gene sequence and deletion/duplication with *TMEM127* gene sequence (concurrent)
  - 5416 PGL/PCC - *SDHB*, *SDHC*, *SDHD*, *SDHAF2* deletion/duplication
  - 5380 *SDHB* gene sequence
  - 5386 *SDHC* gene sequence
  - 5392 *SDHD* gene sequence
  - 5398 *SDHAF2* gene sequence
  - 5410 *TMEM127* gene sequence
- Call Any single gene deletion/duplication GENE \_\_\_\_\_
- 2680 *RET* gene sequence (Multiple Endocrine Neoplasia Type 2)
  - 2606 *VHL* gene sequence and deletion/duplication (Von Hippel-Lindau Disease)

**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 NOTES OR CLINICAL FINDINGS**

**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

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.

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