

Carrier Screening and Prenatal Menu

Why choose Ambry?

- Comprehensive test menu with flexible options and competitive pricing:
 - Specific site analysis for as low as \$200
 - Genetic testing panel options for select hereditary cancer syndromes
 - Excellent customer service for more than a decade, including:
 - Live customer representative to take your call from 8am-8pm, Mon-Fri, Eastern Time
 - Insurance pre-verification in 24 hours
 - Availability of genetic counselors for test selection, case reviews and results interpretation
 - free test kits and shipping
 - Ongoing variant re-classification and complementary family studies program
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Overview:

We are committed to providing comprehensive testing, friendly, knowledgeable client services and innovative research and development. Genetic testing can play a critical role in the medical management of individuals with a predisposition to a hereditary cancer syndrome, and it is our goal to provide thorough and accurate test results for healthcare providers to utilize in their clinical care of patients and their families. We provide clinicians with comprehensive testing offerings with excellent mutation detection rates as well as in-depth results interpretation, including extensive literature review and assessment of all available databases when interpreting a gene mutation or variant of uncertain significance. We offer complimentary family studies for variants of unknown significance to assist in interpretation of the variant of unknown significance, and provide re-classification updates when available.

Over a Decade of Service:

We are here for you. Live client services representatives are available to direct your call from 8am-8pm M-F, and make every effort to get you an immediate answer to your questions or concerns. Genetic counselors are readily available for consultation regarding test selection, case reviews and results interpretation. Free sample submission kits with auto replenish capabilities are available for returning clients. You can be confident that we are here for you with experience, knowledge and solutions.

Pre-Verification and Billing:

We understand that the medical insurance process can be challenging. Our team of experienced billing and insurance specialists are here to help. We offer complete customer service when it comes to pre-verification and billing issues.* The first step is to complete an insurance pre-verification request form and fax it to the billing department at (949) 900-5501 with a copy of the patient's insurance card. You will have a designated billing specialist assigned to your facility, who will review your test request and the patient's insurance benefits and eligibility. If additional proof of medical necessity and/or authorization is necessary, your specialist will contact you to compile the required documents for our submission. In most cases, we will get verification from the insurer within 24 hours. Our responsibility to you is to provide this information as quickly as we receive it from the insurer. We can contact you to let you know of any anticipated out of pocket expense for your patient at their request.

*pre-verification is performed on all tests over \$200.

Consider us your full service cytogenetics and molecular genetics lab.

All forms, including the test requisition and insurance pre-verification form, can be found on our website: www.ambrygen.com/forms

WE ACCEPT. | Blood | DNA | Saliva

DISEASE	GENE(S)	TEST(S)
AmbrySCREEN Carrier Screening	Test for over 300 mutations	Mutation Analysis
Ashkenazi Jewish FlexPanel	<i>ASPA, BCKDHA, BCKDHB, BLM, CFTR, CLRN1, DLD, FANCC, GBA, G6PC, HEXA, IKBKAP, MCOLN1, NEB, PCDH15, SMPD1, TMEM216</i>	Mutation Analysis
Angelman Syndrome/ Prader-Willi Syndrome	<i>SNRPN, UBE3A</i>	<i>SNRPN</i> Methylation, <i>UBE3A</i> Gene Sequence Analysis
Bloom Syndrome	<i>BLM</i>	Mutation Analysis
Beta Thalassemia	<i>HBB</i>	Gene Sequence Analysis
Canavan Disease	<i>ASPA</i>	Mutation Analysis, Gene Sequence & Deletion/Duplication Analysis
Chromosomal Abnormalities		Amniocentesis & CVS Chromosome Analysis, AF-AFP Analysis, AF-AChE Analysis, Aneuploidy FISH Screen.
Cystic Fibrosis	<i>CFTR</i>	CF Screening Panel, 508 First, Gene Sequence & Deletion/Duplication Analysis
Dihydrolipoamide Dehydrogenase Deficiency	<i>DLD</i>	Mutation Analysis
Fabry Disease	<i>GLA</i>	Gene Sequence Analysis
Familial Dysautonomia	<i>IKBKAP</i>	Mutation Analysis
Fanconi Anemia Group C	<i>FANCC</i>	Mutation Analysis
Fragile X Syndrome	<i>FMR1</i>	PCR and Methylation Analysis
Gaucher Disease	<i>GBA</i>	Mutation Analysis, Gene Sequence Analysis
Glycogen Storage Disease Type 1A	<i>G6PC</i>	Mutation Analysis
Joubert Syndrome 2	<i>TMEM216</i>	Mutation Analysis
Maple Syrup Urine Disease	<i>BCKDHA, BCKDHB, DLD</i>	Mutation Analysis
Maternal Cell Contamination		MCC for amniotic cultures + CVS
Mucopolipidosis Type IV	<i>MCOLN1</i>	Mutation Analysis
Niemann-Pick Disease Types A & B	<i>SMPD1</i>	Mutation Analysis, <i>SMPD1</i> Gene Sequence Analysis
Nemaline Myopathy 2	<i>NEB</i>	Mutation Analysis
Noonan/Leopard Syndrome	<i>PTPN11, SOS1, RAF1, KRAS</i>	Partial and Full Gene Sequence Analysis
Pompe Disease	<i>GAA</i>	Mutation Analysis
Sickle Cell/Beta Thalassemia	<i>HBB</i>	Gene Sequence Analysis
Spinal Muscular Atrophy	<i>SMN1</i>	Gene Dosage Analysis
Tay-Sachs Disease	<i>HEXA</i>	Mutation Analysis, Gene Sequence Analysis
Thrombophilia Panel	<i>Factor II, Factor V, MTHFR</i>	Mutation Analysis
Usher Syndrome Type IF	<i>PCDH15</i>	Mutation Analysis
Usher Syndrome III	<i>CLRN1</i>	Mutation Analysis
Y Chromosome Microdeletion	Chromosomal regions: <i>AZFa, AZFb, AZFc and AZFd</i>	Y Chromosome Microdeletion