Ambry Genetics®
A Konica Minolta Company

Hereditary Gastrointestinal Cancer Testing

REFERENCE GUIDE

February 2019

There is no room for doubt when it comes to making important life impacting healthcare decisions. By providing advanced confirmation genetic testing for hereditary gastrointestinal cancer, we can help you make more informed and reliable healthcare decisions with your patients.

The more accurate the results, the more insight you have to better treat your patients.

American College of Gastroenterology

A family history of cancer and premalignant GI conditions that provides sufficient information to develop a preliminary determination of the risk of a familial predisposition to cancer should be obtained for all patients being evaluated in outpatient gastroenterology and endoscopy practices. Genetic testing should be conducted in the context of pre- and post-test genetic counseling to ensure the patient's informed decision making.

Adapted from Am J Gastroenterol, 2015.

Why Is Genetic Testing Important?

KEY BENEFITS

Identifying patients with a genetic predisposition to cancer can allow informed recommendations and personalized medical management that significantly decrease cancer risks and improve overall survival rates.

- Option to modify frequency and initial age of colonoscopy and other screening
- Consideration of prophylactic colectomy or other risk-reducing measures, as appropriate
- Option to tailor chemotherapy strategies and/or determine eligibility for clinical trials
- Identify at-risk family members

Identify Patients Who May Need Genetic Testing

If your patient or their family members have any of the following signs* for hereditary cancer, consider genetic testing:

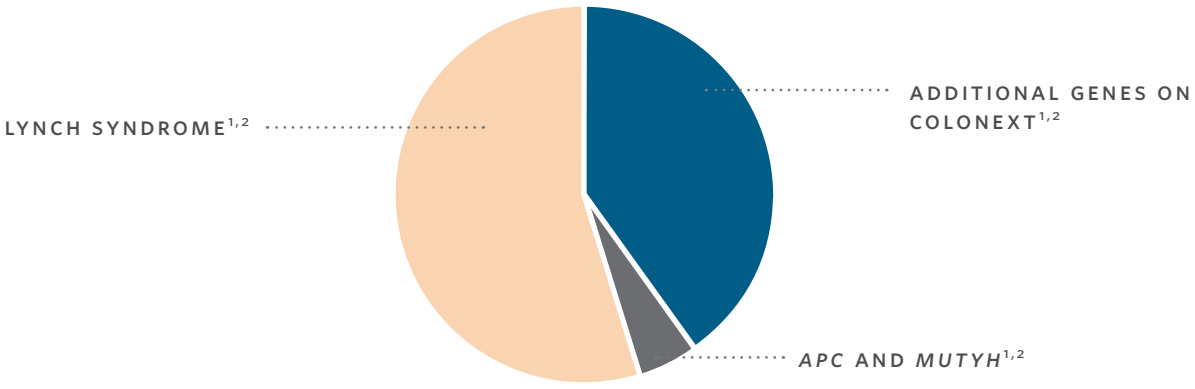
Multiple	**2 OR MORE** primary cancers in the same person	**3 OR MORE** colorectal or other cancers on the same side of the family	**10 OR MORE** colorectal polyps in a person's lifetime
Young	**COLORECTAL CANCER DIAGNOSED <50Y**		
Rare	**PANCREATIC CANCER DIAGNOSED AT ANY AGE**		
MSI/IHC	**ABNORMAL TUMOR SCREENING** Tumors with microsatellite instability (MSI) Tumors with loss of immunohistochemical (IHC) staining for MLH1, MSH2, MSH6, and/or PMS2		

** Adapted from published genetic testing guidelines*

Known Causes of Hereditary Colorectal Cancer

ORDERING THE RIGHT TEST CAN PROVIDE THE MOST ACCURATE AND COMPREHENSIVE ANSWERS

Over 135,000 cases of colorectal cancer are diagnosed each year, and about 5-10% of those are hereditary. Lynch syndrome accounts for the majority of hereditary colorectal cancer cases, but only tells part of the story.



Ambry collaborated in a study of >500 patients with colorectal cancer, in which 10.4.% were identified by ColoNext to have a pathogenic mutation linked to increased cancer risks, which can have significant implications for medical management recommendations.[1]

1. Cragun D, *et al.* Clin Genet 2014 Dec;86(6):510-20.
2. Ambry, Data on File

Finding Answers Through Quality Genetic Testing

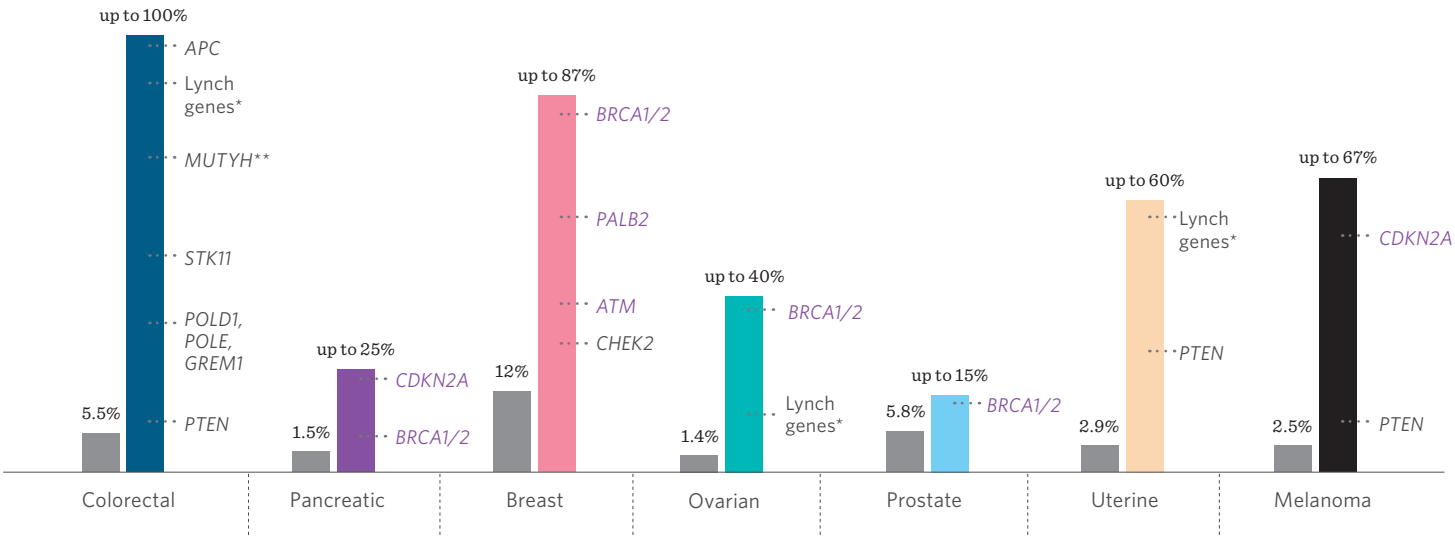
Ambry's hereditary gastrointestinal cancer testing options:

Test	Description	Turnaround Time
ColoNext	Providing more precise information to identify patients with hereditary colorectal cancer and guide personalized medical management recommendations NCCN® management guidelines available for all genes	14-21 days
PancNext	Providing information to identify patients with hereditary pancreatic cancer NCCN® management guidelines available for most genes Combined testing option including hereditary pancreatitis genes also available	14-21 days
CancerNext	Comprehensive panel covering a broad range of tumor types, giving you more information to make better treatment and management decisions NCCN® management guidelines available for most genes	14-21 days

Ambry continually participates in important game-changing studies to expand our knowledge of hereditary cancers. Please visit our website to see the most updated lists of genes included on our panels and additional testing options available: ambrygen.com/hereditary-cancer-panels

Gastrointestinal Cancer Genes and Associated Risks

POTENTIAL LIFETIME CANCER RISKS FOR PATIENTS WITH A HEREDITARY CANCER SYNDROME



General population

Purple genes are on PancNext and CancerNext only

* Lynch genes: *MLH1, MSH2, MSH6, PMS2, EPCAM*

** *MUTYH* biallelic mutations

Over 1 Million Tests Completed

MOVING SCIENCE FORWARD

Purposeful Confirmatory Testing

Many labs validate their tests based on certain limited studies. That's why we participated and led the largest study of its kind (20,000 cases) guiding us to utilize confirmatory testing when we see specific well-defined thresholds. Our mission is to get it right the first time.

Understanding Disease Better Through Free Data Sharing

Identifying an individual's genetic information is nothing new—it's what we do with it that is unique. When labs share genomic information, we can together accelerate the understanding of human disease. Through AmbryShare, we leverage de-identified information to collaborate with others and help people everywhere find answers.

Free Testing for Family Members

We offer specific site analysis (SSA) at no additional cost for family members following single gene or multigene panel testing of the first family member (proband) within 90 days of the original Ambry report date.

Ambry's Translational Genomics (ATG) Lab

As an advanced diagnostic lab, it's our responsibility to ensure the results you get from us are accurate and that classification is as complete and robust as possible. Our ATG lab is a unique laboratory that provides an additional service at no additional cost for you and your patients to generate more precise data potentially bringing clarity to some variants of unknown significance (VUS). This helps to actively drive down the rate of VUS results and can give you an increased understanding of your patient's results, so you can better provide medical management recommendations and improve health outcomes.

About Ambry

Just as no two fingerprints are alike, the way disease presents itself in every individual is different. Since 1999, our mission has always been about understanding disease better, so treatments and cures can be found faster. Every sample that arrives in our lab is viewed as a person with a life and a story that is unique to only them. By providing advanced confirmation genetic testing for inherited and non-inherited diseases, we can help you make more informed and responsible treatment decisions with your patients.

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