

# Technology Assessment



**Technology  
Assessment Program**

**Update on Emerging Genetic Tests  
Currently Available for Clinical Use in  
Common Cancers**

***Prepared for:***  
**Agency for Healthcare  
Research and Quality  
540 Gaither Road  
Rockville, Maryland 20850**

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# **Update on Emerging Genetic Tests Currently Available for Clinical Use in Common Cancers**

## **Technology Assessment Report**

Project ID: GEND0511

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## **Tufts Evidence-based Practice Center**

Gowri Raman, M.D., M.S.  
Esther E. Avendano, B.A.  
Minghua Chen, M.D., M.P.H.

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The information in this report is intended to help health care decision-makers; patients and clinicians, health system leaders, and policymakers, make well-informed decisions and thereby improve the quality of health care services. This report is not intended to be a substitute for the application of clinical judgment. Decisions concerning the provision of clinical care should consider this report in the same way as any medical reference and in conjunction with all other pertinent information, i.e., in the context of available resources and circumstances presented by individual patients.

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## Preface

The Agency for Healthcare Research and Quality (AHRQ), through its Evidence-based Practice Centers (EPCs), sponsors the development of evidence reports and technology assessments to assist public- and private-sector organizations in their efforts to improve the quality of health care in the United States. The reports and assessments provide organizations with comprehensive, science-based information on common, costly medical conditions, and new health care technologies and strategies.

The EPCs systematically review the relevant scientific literature on topics assigned to them by AHRQ and conduct additional analyses when appropriate prior to developing their reports and assessments. To bring the broadest range of experts into the development of evidence reports and health technology assessments, AHRQ encourages the EPCs to form partnerships and enter into collaborations with other medical and research organizations. The EPCs work with these partner organizations to ensure that the evidence reports and technology assessments they produce will become building blocks for health care quality improvement projects throughout the Nation. The reports undergo peer review and public comment prior to their release as a final report.

AHRQ expects that the EPC evidence reports and technology assessments will inform individual health plans, providers, and purchasers as well as the health care system as a whole by providing important information to help improve health care quality.

We welcome comments on this evidence report. Comments may be sent by mail to the Task Order Officer named in this report to: Agency for Healthcare Research and Quality, 540 Gaither Road, Rockville, MD 20850, or by e-mail to [epc@ahrq.hhs.gov](mailto:epc@ahrq.hhs.gov).

Carolyn M. Clancy, M.D.  
Director  
Agency for Healthcare Research and Quality

Jean Slutsky, P.A., M.S.P.H.  
Director, Center for Outcomes and Evidence  
Agency for Healthcare Research and Quality

Stephanie Chang, M.D., M.P.H.  
Director  
Evidence-based Practice Program  
Center for Outcomes and Evidence  
Agency for Healthcare Research and Quality

Kim Marie Wittenberg, M.A.  
Task Order Officer  
Center for Outcomes and Evidence  
Agency for Healthcare Research and Quality

## Peer Reviewers

We wish to acknowledge individuals listed below for their review of this report. This report has been reviewed in draft form by individuals chosen for their expertise and diverse perspectives. The purpose of the review was to provide candid, objective, and critical comments for consideration by the EPC in preparation of the final report. Synthesis of the scientific literature presented here does not necessarily represent the views of individual reviewers.

Diane Allingham-Hawkins, PhD, FCMG, FACMG  
Senior Director, Genetic Test Evaluation and Technical Editing  
Hayes, Inc.  
Lansdale, Pennsylvania

Linda A. Bradley, MD  
Associate Director, Division of Medical Screening and Special Tests  
Women & Infants Hospital Rhode Island  
Providence, Rhode Island

Marta Gwinn, MD, MPH  
Senior Epidemiologist Consultant  
McKing Consulting Corporation  
Atlanta, Georgia

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# **Executive Summary**

## **Introduction**

The Coverage and Analysis Group at the Centers for Medicare and Medicaid Services (CMS) requested that the Technology Assessment Program (TAP) of the Agency for Healthcare Research and Quality (AHRQ) conduct an update of genetic tests for cancer conditions that were identified since the 2011 horizon scan report on Genetic Testing for Cancer. AHRQ assigned this project to the Tufts Medical Center Evidence-based Practice Center (Contract Number: HHS A 290 2007 10055 I, Task Order #11).

The main objective of this report is to provide succinct information on each identified genetic test through grey literature search since 2011. The contents in this report reflect the data of genetic tests that were obtained from manufacturers' Web sites or other commercial Web sites, and should not be considered as verified information or construed as definitive clinical evidence or as recommendations for their routine clinical use.

## **Methods**

We included genetic tests that have applications in the common solid tumors (breast, lung, colorectal, pancreas, etc.) as well as tests that are used in hematologic cancers (leukemia, lymphoma) and are already available in clinical practice. The population of interest was adults with more applicability to the Medicare age group. We included genetic tests that are performed to aid in diagnosing, treating, predicting, prognosticating, monitor patient status and detect cancer recurrence. We also included genetic tests based on at least one of the following selection criteria:

- 1) Genetic tests that have been cleared by FDA or pending clearance by FDA.

2) Genetic tests that are conducted in Clinical Laboratory Improvement Amendments (CLIA) certified labs and require a physician order, but may or may not have been cleared by FDA.

3) Genetic tests offered by Internet sites that specifically require a physician order.

We excluded genetic tests that are performed for cancers that occur in early childhood or adolescence and exclusively result in early death before reaching adulthood and tests marketed directly to consumers (direct-to-consumer genetic tests). For this report, we also excluded tests performed to identify noncancer conditions.

Once the list of current genetic tests was identified, one-page summaries of each test were completed using data extracted from various sources, including laboratory Web sites and test manufacturer Web sites. Data included in these summaries are a more detailed description of the test and its clinical use. The following items are included in the one-page summary: Test name, Description, Purpose (such as Diagnostic, Prognostic, Predictive, Recurrence, Monitoring, and Therapeutic), Availability, Specimen, Diseases, Clinical uses, Source, Marker (Medline Search Terms), Organ (Medline Search Terms), and Exploratory PubMed search.

The horizon scanning has been ongoing as a continuous process since 2005 and the reports were continuously updated until January 2013. The results of grey literature along with one-pagers have been updated weekly. The relevance of the genetic tests was verified at the time of preparation of this report.

## **Results**

We identified 44 new genetic tests for 10 common cancer conditions since the 2011 report, with the largest number of tests being utilized for breast cancer (18 genetic tests). Additionally we added 22 new tests that were identified during peer and public review process. All 44 new genetic tests that we identified were through internet searches alone. Recent grey literature



searches indicate that the largest numbers of new tests were found in the breast cancer category. The one-page description for these newly identified genetic tests for cancer conditions can be found in Appendix A. Of the tests that were identified as tests in our previous reports, the following five tests are excluded for the following reasons: One test (PyloriProbe) has been voluntarily withdrawn from the market, two tests that were identified as those used in the context of aspiration of cervical or breast specimens, one test identified as evaluating genetic material of infectious agent (digene High-Risk HPV HC2 DNA Test), one test (PreGen Plus) has also been withdrawn voluntarily from the market, and one test (OvaSure) identified by our 2011 report has been withdrawn from the market.

## **Discussion**

Since 2011, a total of 66 new genetic tests available for clinical use in 10 common cancer conditions. This report of updating genetic tests for cancer conditions adds potentially important information on emerging tests that are in clinical use. The current report is a valuable source of genetic tests that are in clinical use with specific applicability to older adults. In addition, the yield from this report has helped us to generate topics for conducting systematic reviews of emerging genetic tests. Genetic testing is a rapidly evolving field with the potential to dramatically influence clinical decision-making. Health care providers, patients, payers, decision-makers, and consumers can benefit from staying abreast of newly-released tests.

## Introduction

Recent scientific and technical advances in genomic testing have resulted in the rapid proliferation of lower cost and more efficient genomic technologies. (1;2) The number of available genetic tests that can be used in every day clinical practice is increasing, and the rapid dissemination of information regarding these tests is already occurring through the Internet. The genetic tests are used for a variety of purposes that may include screening, diagnosis, risk stratification, and therapeutic management. In addition, the genetic tests can be used as a clinical decisionmaking tool to aid disease monitoring and prognosis of patients.

Genetic tests are now increasingly being used for the screening and diagnosis of both cancer and noncancer conditions. Those for cancer differ from genetic tests for noncancer conditions in the relatively larger number of tests for somatic mutations. Somatic mutations are genetic mutations that occur in somatic cells after conception. As cancer develops, somatic mutations are common if growth regulators in the cell are damaged by toxins, radiation, random error in cell division, and other factors. Somatic mutations cannot be inherited and only affect the lineage of cells derived from mutated cells. In contrast, mutations in germ cells will affect all the cells in the body, and are often the result of acquired mutations from a parent.

The Coverage and Analysis Group at the Centers for Medicare and Medicaid Services (CMS) requested that the Technology Assessment Program (TAP) of the Agency for Healthcare Research and Quality (AHRQ) conduct an update of the horizon scan of genetic tests for cancer conditions. AHRQ assigned this project to the Tufts Medical Center Evidence-based Practice Center (Contract Number: HHSA 290 2007 10055 I, Task Order #11). The current report presents an update of genetic tests for cancer conditions that were identified since the 2011

horizon scan report on Genetic Testing for Cancer.(3) Issues related to emerging genomic tests include lack of data on test performance, clinical validation, and impact on clinical outcomes. CMS would like the report and the accompanying one-page summaries to serve as a ready reference for their internal discussions in this area as well as the source for decisions on future topic generation for systematic reviews.

The main objective of this report is to provide a broad overview with sufficient information on each identified genetic test, and to provide a preliminary estimate on the amount of published literature available on each genetic test. This report is not meant to be an in-depth review of each test. The contents in this report reflect the data of genetic tests that were obtained from manufacturers' or other commercial Web sites, and should not be construed as verified information or definitive clinical evidence or as recommendations for their routine clinical use.

## **Methods**

### **Genetic test**

Our working definition of genetic tests includes genetic variations, panels of genetic markers, measurements of gene expression and transcription products, biochemical biomarkers, topographic genotyping, and cytogenetic tests. The terms “genetics” and “genomics” are often used interchangeably in the literature, and both can refer to tests for molecular or biochemical biomarkers, as well as cytogenetic and gene-based tests. In general, the genetic tests for cancer conditions have no specific names and are usually named after the disease/condition and/or by the gene and methodology of the specific genetic test. Thus, the name of a genetic test can vary from one laboratory to another. Therefore, the types of genetic tests in this report also include genomic, pharmacogenomic, proteomic, and other tests as reported by the individual manufacturers or laboratories that were identified through Internet searches. We summarized all

genetic tests that provide diagnostic and prognostic information, monitor patient status, or detect disease recurrence.

## **Eligibility criteria**

### ***Inclusion criteria***

We considered genetic tests that have applications in the 10 common solid tumors namely breast, lung, colorectal, pancreas, prostate, ovarian, upper gastrointestinal and liver, genitourinary, endocrine and hematologic cancers (leukemia, lymphoma). We included genetic tests that are already in clinical practice. We included genetic tests based on any one of the following selection criteria:

- 1) Genetic tests that have been cleared by FDA or pending clearance by FDA.
- 2) Genetic tests that are conducted in Clinical Laboratory Improvement Amendments (CLIA) certified labs and require a physician order, but may or may not have been cleared by FDA.
- 3) Genetic tests offered by Internet sites that specifically require a physician order.

The population of interest was adults with more applicability to the Medicare age group. We included genetic tests that are performed to aid in diagnosing, treating, predicting, and prognosticating, and monitoring cancer status or detecting cancer recurrence. Tests conducted for the same gene by multiple laboratories were included only once, except when a test varied explicitly in methodology or description.

### ***Exclusion criteria***

We excluded tests that are performed for cancers that are exclusively early-onset and result in death before reaching adulthood and also excluded were tests marketed directly to consumers (direct-to-consumer genetic tests). We also excluded tests performed for the purpose of identifying noncancer conditions.

## **Clinical Applications of Genetic Tests**

For clinical applications of genetic tests that are covered in this report, we used the following categories to describe various applications:

- 1) Diagnostic: used to confirm or aid in the diagnosis of the particular disease.
- 2) Prognostic: information from the test can be used to determine or predict the aggressiveness of the disease or overall outcome of the disease at the time of initial diagnosis and prior to initiation of treatment.
- 3) Predictive: information from the test can be used to determine or predict the potential risk of eventually developing a disease or a disorder.
- 4) Recurrence: to detect disease recurrence in a patient who has already been diagnosed and treated for cancer.
- 5) Monitoring: test used to monitor tumor and/or patient response to treatment.
- 6) Therapeutic management: information can be used to determine therapeutic decisionmaking.

### **Description of grey literature sources**

The contents in this section were obtained directly from manufacturers' Web sites or other commercial Web sites, and should not be considered as verified information.

1) Genetic Testing Registry (<http://www.ncbi.nlm.nih.gov/gtr>) is a Web site funded by the NIH with an overarching goal to advance public health and research into the genetic basis of health and disease. The NIH Genetic Testing Registry (GTR) is available since 2011 as a central location for voluntary genetic test information by providers. The current contents scope includes test purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts

and credentials. This Web site also includes materials that were previously available at the GeneTests.org.

2) We searched Internet Web sites using the following algorithm. We first searched Google News (<http://www.news.google.com>) for the following: “gene, genetic, genomic, pharmacogenomic, epigenetic” OR “FDA + cleared genetic test.” The news items with their links were automatically deposited into an email system to generate daily email alerts.

Periodically, we visited Web links listed in the news items weekly. We also visited the relevant laboratories that appeared in the news items to identify any new genetic tests. The Web links that identify potentially eligible tests are stored in a spreadsheet.

3) Commercial Web sites were screened to identify genetic tests that are available for routine clinical use. We also identified the Web pages of companies that supply tests such as Roche Diagnostics<sup>®</sup>, or major commercial laboratories in the United States, such as Quest Diagnostics<sup>®</sup>, and LabCorp<sup>®</sup>. A selected list of systematically queried laboratories and their Web sites can be found in Table 1. The Web sites of the major laboratories are visited once quarterly every year. For any potential genetic tests that were mentioned in these Web sites, we conducted focused Internet searches by including the specific test names to find more information, including other manufacturers, suggested uses, and press releases.

**Table 1. Selected list of Web sites that were reviewed to identify new genetic tests for cancers\***

<i>Description</i>	<i>URL</i>
Quest Diagnostics®	<a href="http://www.questdiagnostics.com/">http://www.questdiagnostics.com/</a>
LabCorp®	<a href="http://www.labcorp.com/">http://www.labcorp.com/</a>
Roche Diagnostics®	<a href="http://www.roche-diagnostics.us/">http://www.roche-diagnostics.us/</a>
Athena Diagnostics, Inc	<a href="http://www.athenadiagnostics.com">http://www.athenadiagnostics.com</a>
GeneDx	<a href="http://www.genedx.com">http://www.genedx.com</a>
Abbott Molecular Laboratories	<a href="http://www.abbottmolecular.com">http://www.abbottmolecular.com</a>
Google News	<a href="http://news.google.com">http://news.google.com</a>
FDA News	<a href="http://FDAnews.com">http://FDAnews.com</a>
Genelex Corporation	<a href="http://www.healthanddna.com/">http://www.healthanddna.com/</a>
deCODE Genetics Inc.	<a href="http://www.decode.com/">http://www.decode.com/</a>
Medical Solutions Ltd. (Nottingham)	<a href="http://www.medical-solutions.co.uk/default.aspx">http://www.medical-solutions.co.uk/default.aspx</a>
DiagnoCure	<a href="http://www.diagnocure.com/en/index.php">http://www.diagnocure.com/en/index.php</a>
Epigenomics	<a href="http://www.epigenomics.com/">http://www.epigenomics.com/</a>
Matritech, Inc.	<a href="http://www.matritech.com/">http://www.matritech.com/</a>
Agendia	<a href="http://www.agendia.com/">http://www.agendia.com/</a>
Caris Life Sciences	<a href="http://www.molecularprofiling.com/">http://www.molecularprofiling.com/</a>
Monogram Biosciences	<a href="http://www.monogrambio.com/">http://www.monogrambio.com/</a>
Bostwick Laboratories	<a href="http://www.bostwicklaboratories.com/">http://www.bostwicklaboratories.com/</a>
Arup Laboratories	<a href="http://www.aruplab.com/">http://www.aruplab.com/</a>
Wako Chemicals USA, Inc	<a href="http://www.wakousa.com/">http://www.wakousa.com/</a>
Veridex, LLC	<a href="http://www.veridex.com/">http://www.veridex.com/</a>
Dako (formerly DakoCytomation)	<a href="http://www.dako.com/">http://www.dako.com/</a>
Clariant, Inc	<a href="http://www.clariantinc.com/">http://www.clariantinc.com/</a>
Ambry Genetics	<a href="http://ambrygen.com/">http://ambrygen.com/</a>
Prevention Genetics	<a href="http://www.preventiongenetics.com/">http://www.preventiongenetics.com/</a>
Genomic Health	<a href="http://www.genomichealth.com/">http://www.genomichealth.com/</a>

\* Searches were not limited to these Web sites.

4) Other internet sites: At the direction of experts in the field of genetics, we included tests available at the following Web sites PHG Foundation (phgfoundation.org), EGAPP Reviews (egappreviews.org), and Association for Molecular Pathology (amp.org). To identify additional tests, searches were conducted in major academic /university Web sites such as Mayo Medical Laboratories, Baylor College of Medicine Medical Genetics Laboratories, GeneDx, and Emory Molecular Genetics Laboratory.

5) The two currently developing fields of pharmacogenetics (focuses on single genes) and pharmacogenomics (focuses on multiple genes) may provide insights into the inter-individual variability in drug responses. We identified genetic tests from the PharmGKB Web site ([pharmgkb.org](http://pharmgkb.org)) maintained by Stanford University (4).

### **Individual test summaries**

Once the list of current genetic tests was identified, one-page summaries of each test were completed using data extracted from various sources, including laboratory Web sites and test manufacturer Web sites. Data included in these summaries are a more detailed description of the test and its clinical use. The “one-page summary” included the following items:

- 1) **Test name:** The majority of the clinically available genetic tests were identified either by the disease/ conditions or by the disease causing genes without any specific test name. Hence the gene names, protein, and disease/conditions served as the surrogate for the genetic testing identifier. When available, we recorded the specific test name.
- 2) **Description:** Included a brief summary of the genetic or genomic test and its association with the cancer condition.
- 3) **Purpose:** The clinical applications of genetic tests included primary or secondary prevention, diagnostic, prognostic, predictive, recurrence, monitoring, and therapeutic management.
- 4) **Availability:** Included a brief list of laboratories including commercial and academic laboratories in the U.S. and other countries.
- 5) **Specimen:** The specimen was utilized to evaluate the gene-disease condition, which included whole blood, serum, tumor tissue, etc.
- 6) **Diseases:** Included a list of disease conditions for which the genetic test was utilized



- 7) **Clinical uses:** Included genetic test applications in a clinical setting (e.g. routine use, investigational use, etc.).
- 8) **Source:** A list of additional sources that were typically consulted for information about the genetic test application.
- 9) **Marker (Medline Search Terms):** A PubMed search parameter; included the list of possible genetic test names, genes, and biomarkers that were used for Medline search strategy.
- 10) **Organ (Medline Search Terms):** A PubMed search parameter; included a list of specific organ(s) affected by the gene-disease association.
- 11) **Exploratory PubMed search:** The exploratory PubMed search included the name of the genetic or molecular marker, the disease, and the terms “cancer condition [MeSH®]”. For tests that use a panel of genetic or molecular markers, we used the brand name of the panel crossed with the search terms. All searches were repeated on 1/31/2013. For new tests added at the time of draft revision, all searches were repeated on 6/1/2013. These search strategies are exploratory and the number of citations returned is an estimate of the scientific literature available on each test-disease condition. However, this number is preliminary, and depending on the key questions to be addressed in a systematic review, the final yield of eligible citations may change based on search strategy and the application of specific screening criteria.

### **Updating of the reports**

The horizon scanning has been ongoing as a continuous process since 2005 and the identified tests are being continuously updated. We also assessed the relevance and availability of genetic tests identified overtime.

## Results

Overall, the horizon scan reports have identified 178 different genetic tests for 10 common cancer conditions. Our report lists 66 new genetic tests since the 2011 report, with the largest number of tests being utilized for breast cancer (**Table 2**). We identified 44 new tests through grey literature searches, and during peer review process, we added 22 new tests that are currently available in clinical use. The one-page description for these newly identified genetic tests for a variety of common solid tumors and hematological cancers cancer conditions can be found in Appendix A. Tests that were identified in our previous reports are listed in the Appendix Tables 1 and 2. One test (OvaSure) identified by our 2011 report has been withdrawn from the market. In addition, one test (PreGen Plus) identified as a test in clinical use in our 2006 report has also been withdrawn voluntarily from the market.

Of the 104 tests that were identified as tests in development in our 2006 report, only 21 tests matured to full clinical use in 2011. Recent Internet searches indicate that three additional tests are available for clinical uses (Appendix B). Among tests that were in development, four were excluded for the following reasons: one test (PyloriProbe) has been voluntarily withdrawn from the market, two tests that were identified as those used in the context of aspiration of cervical or breast specimens were excluded, and one test was excluded since it was identified as evaluating genetic material of infectious agent (digene High-Risk HPV HC2 DNA Test). The remaining 76 tests are currently being tracked as tests in development or in research.

**Table 2. Genetic tests for cancer found between March 2011 and January 2013**

Test Name	Germline (yes)	Purpose					
		Prognostic	Predictive	Diagnostic	Monitoring	Recurrence	Therapeutic management
<b><i>Breast</i></b>							
<a href="#">Blue Print</a>				X			X
<a href="#">Breast cancer index</a>		X				X	
<a href="#">BreastOncPx™</a>							X
<a href="#">BreastNext™</a>	Yes		X				
Caris Target Now® for Breast Cancer <sup>1</sup>							X
<a href="#">Cytochrome P450 2D6 genotyping</a>		X					X
<a href="#">Inform Dual ISH</a>							X
<a href="#">Her-2 by FISH; Her-2 by ISH</a>		X					X
<a href="#">HER-2 neu (ERBB2)</a>		X					X
<a href="#">HERmark Breast Cancer Assay</a>		X					X
<a href="#">HER2 – DUAL ISH</a>							X
<a href="#">MammaPrint</a>		X				X	X
<a href="#">Mammostrat</a>		X				X	X
<a href="#">MapQuant Dx Genomic grade test</a>							X
OncoVue® <sup>1</sup>		X	X				
<a href="#">OncoType DX™</a>		X				X	X
<a href="#">PAM50 breast Intrinsic Classifier</a>		X					X
<a href="#">Rotterdam Signature 76-Gene Panel</a>		X				X	
<a href="#">SYMPHONY™ Breast Cancer Profile</a>		X		X		X	X
<a href="#">TargetPrint</a>							X
<b><i>Colorectal</i></b>							
Caris Target Now® for Colorectal Cancer <sup>1</sup>							X
<a href="#">ColoNext™</a>	Yes		X				
<a href="#">ColoPrint™</a>		X		X			X
<a href="#">ColoSeq™</a>	Yes		X				
<a href="#">GCC (GUCY2C) Blood Test</a>				X			
<a href="#">Therascreen KRAS RGQ PCR</a>				X			X

<a href="#">PMS2 test</a>			X	
<a href="#">Previstage™ GCC</a>		X		
<a href="#">ResponseDx Colon®</a>				X
<b>Genitourinary</b>				
<a href="#">Onco FISH cervical</a>		X		
<a href="#">UteroFISH</a>				X
<a href="#">UroVysion FISH</a>			X	X
<b>Hematologic</b>				
<a href="#">5q del, 7q del/-7 FISH test</a>		X		X
<a href="#">17 p deletion FISH</a>		X		
<a href="#">Multiple myeloma panel FISH test</a>		X		X
<a href="#">MyPRS Plus</a>		X		X
<b>Lung</b>				
<a href="#">ALK FISH</a>			X	X
<a href="#">Vysis ALK FISH test</a>				X
Caris Target Now® for NSCLC <sup>1</sup>				X
<a href="#">ResponseDx Lung®</a>				X
<b>Ovarian</b>				
Caris Target Now® for Ovarian Surface Epithelial Cancer <sup>1</sup>				X
<a href="#">PreOvar</a>	Yes		X	
<b>Prostate</b>				
<a href="#">Adenomatous polyposis coli (APC)</a>	Yes	X		
<a href="#">Glutathione-S-Transferase (GST-P1)</a>		X		
<a href="#">Oncotype Dx Prostate</a>				X
<a href="#">ProstaVysion</a>		X		X
<b>Other<sup>2</sup></b>				
<a href="#">Cobas® BRAF V600 mutation</a>				
<a href="#">BRAF gene mutation detection</a>	Yes			X
Caris Target Now® for Melanoma Cancer <sup>1</sup>				X
<a href="#">MEN2 (RET) DNA sequencing test</a>	Yes		X	
<a href="#">miRInform™ Pancreas</a>			X	
<a href="#">NeoSite Melanoma</a>		X		
<a href="#">PIK3CA Oncogene mutation detection</a>		X		X
<a href="#">PDGFRA mutation analysis</a>		X		X

<a href="#">ResponseDx Melanoma®</a>		X		X
<a href="#">ResponseDx Gastric®</a>		X		X
<a href="#">RET gene sequencing</a>	Yes		X	
<a href="#">SDHB DNA sequencing test</a>	Yes	X		
<a href="#">1P, 19Q FISH</a>		X	X	
<b>Multiple<sup>3</sup></b>				
<a href="#">BROCA-Cancer Risk Panel</a>	Yes		X	
<a href="#">CancerNext™</a>	Yes		X	
CancerType ID® <sup>1</sup>				
<a href="#">EGFR FISH</a>		X		
<a href="#">OVANEXT</a>	Yes		X	
<a href="#">PANEXIA</a>	Yes		X	
<a href="#">PTEN genetic analysis for cancer</a>		X	X	X
<a href="#">5-FU sensitivity (DPYD, TYMS, and MTHFR)</a>				X

1. One pager tests are not provided owing to the inadequate information available on company Web site
2. Other includes brain, liver, and upper gastrointestinal, respectively
3. Test used for multiple cancers including lung and brain

## Discussion

We performed Internet-based grey literature searches and added a total of 66 new genetic tests available for clinical use in cancer conditions since our 2011 report. Of these, 44 new genetic tests were identified through grey literature searches alone. The remaining 22 tests were identified by peer and public reviewers. A total of 24 tests matured to clinical use of the 104 tests “in development” in our 2006 report. Recent grey literature searches indicate that the largest numbers of new tests were found in the breast cancer category to aid in prognosis or predict response to therapies as well as to individualize therapeutic management. Most of the information for each of the genetic tests was gathered from various public and proprietary Web sites. The laboratories offering genetic testing services provided most of the information on the description of the gene involved with the disease. We searched sites that were identified from our prior horizon scan reports (previous *Genetic Testing for Cancer Conditions* reports) and many other sites identified through Google News searches. In terms of tests that were in development, only few biomarkers (22%) made it to the clinical application stage.

Potential limitations of our report include lack of empirical structure providing guidance on how to conduct optimal grey literature searches of the Internet. The following are caveats to our grey literature searches. Internet searches are not strictly reproducible. Periodically we stored Web links along with access dates. However, for searches conducted within a reasonably short time period, the Web pages will be more or less the same. To overcome such limitations related to Internet searches conducted in Google, we supplemented with periodic review of Web sites of major companies that manufacture genetic and molecular tests, and by searching the FDA Web site. The attempt to horizon scan genomic testing through Web searching has been applied by at

least one other group that focuses on emerging genetic tests with continuous updating (<http://www.hugenavigator.net/GAPPKB/topicFinder.do>).(5) We did not contact companies and, this process limits our ability to identify a test with multiple commercial names (for example, a test that has been licensed from one company to another company, but carries a different commercial name for the same test) or if changes are made to a test that retains the same name (for example, when additional single-nucleotide polymorphisms are added to a test). Future grey literature searches can explore the possible engagement of relevant stakeholders in this field to identify potentially useful Web sites.

Our report indicates that there has been an increase in the number of genetic tests available for clinical use, and we limited emerging genetic tests. Many genetic and molecular markers and panels are being associated with cancer conditions. We have selected those that are available for clinical applications in screening, diagnosis, prognosis, prediction, disease recurrence, therapeutic management, or patient monitoring as tests for cancer conditions. In addition to grey literature searches, our discussion with local experts helped us to identify this list of genetic tests. The tests identified from prior reports have been utilized to generate topics for conducting systematic reviews for various cancer conditions.(6-8)

This report of horizon scan for genetic tests for cancer conditions, with biannual updates, adds important information on emerging tests. The NIH registry was launched in February 2012.(9) Currently, NIH registry is fully effective and future readers are directed to obtain information on emerging genetic tests from their Web site (<http://www.ncbi.nlm.nih.gov/gtr/>).(10) The current report is a valuable source of genetic tests that are in clinical use for common cancer conditions with specific applicability to older adults. Genetic testing is a rapidly emerging field with the potential to dramatically influence clinical

decision-making. Health care providers, patients, payers, decision-makers, and consumers can benefit from staying abreast of newly-released tests.



## References

- (1) Khoury MJ, Dorman JS. The Human Genome Epidemiology Network. *Am J Epidemiol.* 1998;148:1-3.
- (2) Khoury MJ, Millikan R, Little J, Gwinn M. The emergence of epidemiology in the genomics age. *Int J Epidemiol.* 2004;33:936-44.
- (3) Chin, KM, Wessler, B, Chew, P, and Lau, J. Genetic Tests for Cancer. <http://archive.ahrq.gov/clinic/ta/gentests/>. 2006. Evidence Report/Technology Assessment. Access Date: 6/1/13.
- (4) Thorn CF, Klein TE, Altman RB. PharmGKB: the pharmacogenetics and pharmacogenomics knowledge base. *Methods Mol Biol.* 2005;311:179-91.
- (5) Gwinn M, Grossniklaus DA, Yu W, Melillo S, Wulf A, Flome J et al. Horizon scanning for new genomic tests. *Genet Med.* 2011;13:161-65.
- (6) Dahabreh IJ, Terasawa T, Castaldi PJ, Trikalinos TA. Systematic review: Anti-epidermal growth factor receptor treatment effect modification by KRAS mutations in advanced colorectal cancer. *Ann Intern Med.* 2011;154:37-49.
- (7) Raman, G., Trikalinos, T. A., Zintzaras, E., Kitsios, G., Ziogas, D., Ip, S., and Lau, J. Reviews of Selected Pharmacogenetic Tests for Non-Cancer and Cancer Conditions. <http://www.cms.gov/Medicare/Coverage/DeterminationProcess/downloads/id61TA.pdf> . 2008. Access Date: 6/1/13.
- (8) Terasawa T, Dahabreh I, Trikalinos TA. BCR-ABL mutation testing to predict response to tyrosine kinase inhibitors in patients with chronic myeloid leukemia. *PLoS Curr.* 2010;2:RRN1204.
- (9) Kuehn BM. NIH launching genetic test registry. *JAMA.* 2010;303:1685.
- (10) Rubinstein WS, Maglott DR, Lee JM, Kattman BL, Malheiro AJ, Ovetsky M et al. The NIH genetic testing registry: a new, centralized database of genetic tests to enable access to comprehensive information and improve transparency. *Nucleic Acids Res.* 2013;41:D925-D935.

## **Appendix A. One-page summaries of the genetic tests for cancers**

## **BREAST CANCER**

## **Gene Test Information: Breast cancer index, breast cancer**

### **Test Name: Breast cancer index**

**Description:** BioTheranostics Breast Cancer Index<sup>SM</sup> (BCI) is a prognostic biomarker that provides quantitative assessment of the likelihood of distant recurrence in patients diagnosed with estrogen receptor-positive, lymph node-negative breast cancer. In development and validation studies, BCI stratified ~50% of tamoxifen treated ER+, node-negative breast cancer patients into a low risk group for 10-year distant recurrence.

**Purpose:** Prognostic and Recurrence

**Availability:** Bio Theranostics

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue block

**Methodology:** BCI is a molecular assay developed from the combination of two indices: HOXB13:IL17BR and Five cell cycle-associate gene index that assesses tumour grade.

**Diseases:** breast cancer

**Clinical Uses:** BioTheranostics Breast Cancer Index<sup>SM</sup> (BCI) is a prognostic biomarker that provides quantitative assessment of the likelihood of distant recurrence in patients diagnosed with estrogen receptor-positive, lymph node-negative breast cancer.

**Sources:** [www.biotheranostics.com](http://www.biotheranostics.com)

**Marker (Medline Search):** HOXB13 OR IL17BR

**Organ (Medline Search):** breast

**Medline Searches:** (HOXB13[All Fields] OR IL17BR[All Fields]) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**31

**FDA approved:** No

## **Gene Test Information: Blueprint, breast cancer**

### **Test Name: Blueprint® Molecular Subtyping Signature**

**Description:** Blueprint is an 80-gene profile that classifies breast cancer into molecular subtypes. The profile separates tumors into Basal-type, Luminal-type and ERBB2-type subgroups by measuring the functionality of downstream genes for each of these molecular pathways to inform the physician of the potential effect of adjuvant therapy.

**Purpose:** Diagnostic, Therapeutic management of breast cancer

**Availability:** Agendia

**Specimen:** Formalin fixed paraffin-embedded, fresh or frozen breast tumor tissue

**Methodology:** Genomic signature by microarray-based RNA gene expression

**Diseases:** breast cancer

**Clinical Uses:** Blueprint® provides information on the sub-classification of the tumor which guides the choice of therapies and combinations of therapies.

**Sources:** [www.agendia.com](http://www.agendia.com)

**Marker (Medline Search):** Blueprint

**Organ (Medline Search):** breast

**Medline Searches:** Blueprint[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**22

**FDA approved:** No

**Gene Test Information:** BreastNext™

**Test Name:** BreastNext™

**Description:** BreastNext™ utilizes next generation sequencing to offer a comprehensive testing panel for hereditary breast and/or ovarian cancer and targets detection of mutations in 14 genes (ATM, BARD1, BRIP1, CDH1, CHEK2, MRE11A, MUTYH, NBN, PALB2, PTEN, RAD50, RAD51C, STK11 and TP53), excluding BRCA1 and BRCA2. Gross deletion/duplication analysis is performed for all 14 genes.

**Purpose:** Predictive

**Availability:** Ambry genetics

**Specimen:** Blood, DNA, or Saliva

**Methodology:** gene sequencing, deletion/duplication analysis

**Diseases:** Breast cancer; ovarian cancer

**Clinical Uses:** Mutations in BRCA1 and BRCA2 explain hereditary breast cancer occurrence ~25–50% of the time, additional genes associated with hereditary breast cancer are emerging. Studies suggest that mutations in the genes on the BreastNext™ panel may confer an estimated 25–70% lifetime risk for breast cancer.

**Sources:** [www.ambrygen.com](http://www.ambrygen.com)

**Marker (Medline Search):** (ATM OR BARD1 OR BRIP1 OR CDH1 OR CHEK2 OR MRE11A OR MUTYH OR NBN OR PALB2 OR PTEN OR RAD50 OR RAD51C OR STK11 OR TP53)

**Organ (Medline Search):** breast OR ovarian cancer

**Medline Searches:** (("atm"[All Fields]) OR BARD1[All Fields] OR BRIP1[All Fields] OR CDH1[All Fields] OR CHEK2[All Fields] OR muty[All Fields] OR NBN[All Fields] OR PALB2[All Fields] OR pten[All Fields] OR RAD50[All Fields] OR RAD51C[All Fields] OR STK11[All Fields] OR TP53[All Fields]) AND (("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields]) OR ("ovarian neoplasms"[MeSH Terms] OR ("ovarian"[All Fields] AND "neoplasms"[All Fields]) OR "ovarian neoplasms"[All Fields] OR ("ovarian"[All Fields] AND "cancer"[All Fields]) OR "ovarian cancer"[All Fields])) AND "humans"[MeSH Terms]

**Medline hits=**3202

**FDA approved:** No

## **Gene Test Information: Cytochrome P450 2D6, breast cancer**

### **Test Name: Cytochrome P450 2D6 genotyping**

**Description:** The cytochrome (P450) enzyme catalyse the oxidation of many drugs and chemicals. Individual differences of cytochrome P450 activity can result in the total absence of the metabolism of certain drugs to ultrafast metabolism and can also lead to adverse drug reactions or a lack of therapeutic effect under standard therapy conditions. Specific variants in this gene also influence the metabolism of the breast cancer drug, tamoxifen, in postmenopausal women.

**Purpose:** Predictive and Therapeutic management

**Availability:** Labcorp

**Specimen:** Whole blood

**Methodology:** Polymerase chain reaction (PCR) and allele-specific primer extension. This analysis includes identification of the following CYP2D6 alleles: \*1, \*2, \*3, \*4, \*5, \*6, \*7, \*8, \*9, \*10, \*11, \*12, \*14, \*15, \*17, \*41, \*1XN, \*2XN, \*4XN, \*10XN, \*17XN, \*41XN.

**Diseases:** Breast cancer

**Clinical Uses:** Genetic polymorphism of CYP2D6 could be used to predict the altered enzyme activity and address the potential effects of metabolized drugs.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** cytochrome P450 2D6 and breast cancer

**Organ (Medline Search):** breast

**Medline Searches:** ("cytochrome p-450 cyp2d6"[MeSH Terms] OR ("cytochrome"[All Fields] AND "p-450"[All Fields] AND "cyp2d6"[All Fields]) OR "cytochrome p-450 cyp2d6"[All Fields] OR ("cytochrome"[All Fields] AND "p450"[All Fields] AND "2d6"[All Fields]) OR "cytochrome p450 2d6"[All Fields]) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

Medline hit: 247

**FDA approved:** No

## **Gene Test Information: Dual ISH, breast**

### **Test Name: Inform Dual ISH**

**Description:** The Inform Dual ISH test allows lab personnel to count the number of copies of HER2 genes on chromosome 17 in a small sample of the breast tumor. Copies of the HER2 gene appear black and copies of chromosome 17 appear red. Patients with more than the normal number of copies of the HER2 gene are considered candidates for Herceptin therapy.

**Purpose:** Therapeutic management

**Availability:** Ventana Medical Systems

**Specimen:** Tumour tissue

### **Methodology: Dual in-situ hybridization (ISH)**

**Diseases:** Breast cancer

**Clinical Uses:** A new genetic test that will help health care professionals determine if women with breast cancer are HER2-positive and, therefore, candidates for Herceptin (trastuzumab), a commonly used breast cancer treatment

**Sources:** [www.fda.gov](http://www.fda.gov)

**Marker (Medline Search):** HER-2 chromosome 17

**Organ (Medline Search):** breast

**Medline Searches:** (("genes, erbb-2"[MeSH Terms] OR ("genes"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 genes"[All Fields] OR "her 2 neu"[All Fields]) AND (("chromosomes"[MeSH Terms] OR "chromosomes"[All Fields] OR "chromosomal"[All Fields]) AND 17[All Fields])) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**174

**FDA approved:** Yes



## **Gene Test Information: Her-2 by FISH; Her-2 by ISH, breast**

### **Test Name: Her-2 by FISH; Her-2 by ISH**

**Description:** HER 2 amplification (positive) predicts resistance to standard chemotherapy. HER2 amplification also predicts response to anthracycline agents (adriamycin) in high dose chemotherapy, and response to the humanized monoclonal antibody. A negative test is indicated by a ratio of less than 1.8, equivocal test is indicated by a ratio from 1.8 to 2.2, and a positive test is indicated by a ratio greater than 2.2.

**Purpose:** Prognostic and therapeutic management

**Availability:** PathVysion, a registered trademark of Vysis Inc

**Specimen:** Tumour tissue

**Methodology:** Fluorescence in situ Hybridization; In situ Hybridization

**Diseases:** Breast cancer

**Clinical Uses:** HER-2 Fluorescence in situ Hybridization test are often used in advanced breast cancer to assess prognosis and therapeutic choices

**Sources:** [www.labcorp.com/wps/portal/](http://www.labcorp.com/wps/portal/); [www.abbottmolecular.com](http://www.abbottmolecular.com)

**Marker (Medline Search):** HER-2 AND Fluorescence in situ Hybridization test

**Organ (Medline Search):** Breast

**Medline Searches:** ("genes, erbb-2"[MeSH Terms] OR ("genes"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 genes"[All Fields] OR "her 2 neu"[All Fields]) AND ("receptor, erbb-2"[MeSH Terms] OR ("receptor"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 receptor"[All Fields] OR "erbb2"[All Fields] OR "genes, erbb-2"[MeSH Terms] OR ("genes"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 genes"[All Fields]) AND (("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("research design"[MeSH Terms] OR ("research"[All Fields] AND "design"[All Fields]) OR "research design"[All Fields] OR "test"[All Fields]) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields]))

**Medline hits=**124

**FDA approved:** Not reported

## **Gene Test Information: HER-2 neu (ERBB2), breast**

### **Test Name: HER-2 neu (ERBB2)**

**Description:** HER 2 amplification (positive) predicts resistance to standard chemotherapy. HER2 amplification also predicts response to anthracycline agents (adriamycin) in high dose chemotherapy, and response to the humanized monoclonal antibody.

**Purpose:** Prognostic and therapeutic management.

**Availability:** BayCare Laboratories

**Specimen:** Tumour tissue

**Methodology:** Fluorescence in situ Hybridization

**Diseases:** Breast cancer

**Clinical Uses:** HER-2 neu (ERBB2) is used to help determine prognosis and therapeutic choices for invasive breast cancer.

**Sources:** [www.baycare.org/laboratories](http://www.baycare.org/laboratories)

**Marker (Medline Search):** HER-2 neu (ERBB2) Fluorescence in situ Hybridization test

**Organ (Medline Search):** Breast

**Medline Searches:** ("genes, erbb-2"[MeSH Terms] OR ("genes"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 genes"[All Fields] OR "her 2 neu"[All Fields]) AND ("receptor, erbb-2"[MeSH Terms] OR ("receptor"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 receptor"[All Fields] OR "erbb2"[All Fields] OR "genes, erbb-2"[MeSH Terms] OR ("genes"[All Fields] AND "erbb-2"[All Fields]) OR "erbb-2 genes"[All Fields]) AND (("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("research design"[MeSH Terms] OR ("research"[All Fields] AND "design"[All Fields]) OR "research design"[All Fields] OR "test"[All Fields]) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields]))

**Medline hits=**124

**FDA approved:** Yes

## **Gene Test Information: HERmark, Breast Cancer**

### **Test Name: HERmark Breast Cancer Assay**

**Description:** HERmark Breast Cancer Assay is used to help determine prognosis and therapeutic choices for metastatic breast cancer. Clinical Practice Guidelines recommend determining HER2 status in patients with all invasive breast cancer, but caution that current HER2 testing methods such as central immunohistochemistry and Fluorescence in situ Hybridization test may be inaccurate in approximately 20% of cases. According to the HERmark Web site, their method precisely quantifies HER2 total protein and HER2 homodimer levels in formalin-fixed, paraffin-embedded tissue sections and outperformed Fluorescence in situ Hybridization at determining patient outcomes in patients with metastatic breast cancer.

**Purpose:** Prognostic and therapeutic management

**Availability:** HERmark®, a registered trademark of biosciences monogram

**Specimen:** Formalin-fixed, paraffin-embedded tissue sections

**Methodology:** Not reported

**Diseases:** breast cancer

**Clinical Uses:** prognosis and therapeutic choices

**Sources:** [www.labcorp.com](http://www.labcorp.com); [www.monogrambio.com](http://www.monogrambio.com)

**Marker (Medline Search):** HERmark

**Organ (Medline Search):** Breast

**Medline Searches:** HERmark[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits =**7

**FDA approved:** Not reported

## **Gene Test Information: HER2 DUAL ISH, Breast Cancer**

### **Test Name: HER2 – DUAL ISH**

**Description:** HER 2 overexpression occurs in 18-20% of breast cancers and indicates resistance to standard chemotherapy. Patients with HER2 positive breast cancer can benefit from Trastuzumab, a humanized monoclonal antibody against the extracellular domain of HER2. Therefore, demonstration of HER2 gene amplification and/or protein overexpression in the tumor can aid in selecting patients for trastuzumab therapy. Chromogenic in situ hybridization signals do not fade over time allowing tissue samples to be archived and reviewed later.

**Purpose:** Therapeutic management (adjunctive with HER IHC and HER FISH)

**Availability:** Labcorp

**Specimen:** Formalin-fixed Tumor tissue of breast

**Methodology:** Chromogenic in situ hybridization (ISH)

**Diseases:** Breast Cancer

**Clinical Uses:** Evaluation aids in identifying breast cancer patients with HER2 overexpression and in selecting patients for trastuzumab therapy.

**Sources:** [www.labcorp.com/integratedoncology](http://www.labcorp.com/integratedoncology)

**Marker (Medline Search):** HER2 AND Chromogenic in situ hybridization

**Organ (Medline Search):** Breast cancer

**Medline Searches:** HER2[All Fields] AND (dual[All Fields] AND ("in situ hybridization"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields]) OR "in situ hybridization"[All Fields])) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits = 53**

**FDA approved:** Yes

## **Gene Test Information: MammaPrint, breast cancer**

### **Test Name: MammaPrint**

**Description:** MammaPrint a 70-gene profile that classifies breast cancer into Low Risk or High Risk of recurrence, by measuring genes representative of all the pathways of cancer metastases which were selected for their predictive relationship to 10-year recurrence probability. MammaPrint is indicated for women who have stage I or II breast cancer, are lymph node positive or negative, are ER-positive or negative and tumor size of less than five centimeters.

**Purpose:** Prognosis, recurrence, predictive, and therapeutic management of breast cancer

**Availability:** Agendia.com

**Specimen:** Formalin fixed paraffin-embedded, fresh or frozen breast tumor tissue

**Methodology:** Genomic signature by microarray-based RNA gene expression

**Diseases:** Breast cancer

### **Clinical Uses:**

MammaPrint determines if the patient is a candidate for chemotherapy..

**Sources:** [www.agendia.com](http://www.agendia.com)

Marker: MammaPrint

Organ: Breast

**Medline Searches:** MammaPrint[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=74**

**FDA approved:** Yes

## **Gene Test Information: Mammostrat, breast cancer**

### **Test Name: Mammostrat**

**Description:** Mammostrat is a novel test for estimating the risk for recurrence in hormone-receptor positive, early stage breast cancer that is independent of proliferation and grade. Five biomarkers are combined with a defined mathematical algorithm resulting in a risk index. Mammostrat is clinically validated and has been studied on more than 4,500 total patients in numerous independent cohorts that include the NSABP B14 and B20 trials.

**Purpose:** Prognostic, recurrence, and therapeutic management

**Availability:** Clarinet

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue block

**Methodology:** Not reported

**Diseases:** Breast cancer

**Clinical Uses:** Clinicians and patients are faced with difficult choices as to whether to add toxic adjuvant chemotherapy in addition to standard endocrine treatment. Mammostrat may help clinicians understand the inherent aggressiveness of the tumor and the likelihood of tumor recurrence.

**Sources:** [www.clarientinc.com](http://www.clarientinc.com)

**Marker (Medline Search):** Mammostrat

**Organ (Medline Search):** breast

**Medline Searches:** Mammostrat[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**3

**FDA approved:** No

## **Gene Test Information: MapQuant Dx Genomic grade test, breast cancer**

### **Test Name: MapQuant Dx Genomic grade test**

**Description:** Tumor grade is a decision factor in most national and international guidelines to breast cancer treatment. It is generally recommended to treat high-grade “grade 3” breast carcinoma with chemo therapy because they are chemosensitive and will often recur otherwise. By contrast, most low-grade “grade 1” tumors should not be treated with chemotherapy because they have a good prognosis and are often chemo-insensitive. A key clinical issue is how to treat the 50% of breast cancers tested today as uncertain/Intermediate “grade 2” by current methods. MapQuant DX genomic grade test directly measures the expression of 97 genes that best characterize high-grade vs. Low-grade tumors. It can resolve these grade 2 tumors into either grade 1 or grade 3 tumors in 80% of cases.

**Purpose:** Therapeutic management of breast cancer

**Availability:** IPSOGEN

**Specimen:** Blood

**Methodology:** Not reported

**Diseases:** Breast cancer

**Clinical Uses:** This test may be useful when tumor grade information can be decisive for prescribing chemotherapy.

**Sources:** [www.ipsogen.com](http://www.ipsogen.com)

**Marker (Medline Search):** MapQuant

**Organ (Medline Search):** breast

**Medline Searches:** MapQuant[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits** =4

**FDA approved:** No

## **Gene Test Information: OncoType DX, breast cancer**

### **Test Name: OncoType DX TM**

**Description:** Oncotype that analyses the patterns of 21 genes is being applied as a quantification tool for likelihood of breast cancer recurrence within 10 years of newly diagnosed, stage I or II, lymph node-negative, hormone receptor-positive breast cancer in women who will be treated with tamoxifen.

**Purpose:** Prognosis, recurrence, and therapeutic management

**Availability:** Genomic Health

**Specimen:** Paraffin-preserved tissue

**Methodology:** RT-qPCR

**Diseases:** Breast cancer

**Clinical Uses:** Oncotype is being applied as a quantification tool for likelihood of breast cancer recurrence in 10 years in women with newly diagnosed breast cancer. It is also intended to assist in making decisions regarding adjuvant chemotherapy based on recurrence likelihood.

**Sources:** [www.genomichealth.com](http://www.genomichealth.com)

Marker: OncoType DX

Organ: Breast

**Medline Searches:** OncoType[All Fields] AND DX[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**118

**FDA approved:** No



**Gene Test Information: BreastOncPx™, breast cancer**

**Test Name: BreastOncPx™ (Breast Cancer Prognosis Gene Expression Assay)**

**Description:** BreastOncPx™, a 14-gene signature assay provides prognostics information for lymph node-negative (N-), estrogen receptor-positive (ER+) breast cancer patients and is associated with risk of distant metastasis. It helps identify higher-risk patients who might benefit from additional therapy.

**Purpose:** Therapeutic management

**Availability:** US Labs

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue block

**Methodology:** 14-gene signature assay

**Diseases:** Breast cancer

**Clinical Uses:** BreastOncPx™ that provides prognostic information to aid identifying higher-risk patients who might benefit from additional therapy.

**Sources:** [www.uslabs.net](http://www.uslabs.net)

**Marker (Medline Search):** BreastOncPx™ and breast cancer

**Organ (Medline Search):** breast

**Medline Searches:** mark[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits** =476

**FDA approved:** No

**Gene Test Information: PAM50 breast Intrinsic Classifier, breast cancer**

**Test Name: PAM50 breast Intrinsic Classifier**

**Description:** PAM50 Breast Cancer Intrinsic Classifier examining 50 genes and sorts breast cancer into four subtypes. Each subtype responds differently to standard therapies, and knowing the subtype allows doctors to tailor treatment for each patient.

**Purpose:** Prognostic and therapeutic management

**Availability:** University of Utah

**Specimen:** Tumor tissue

**Methodology:** RT-qPCR

**Diseases:** Breast cancer

**Clinical Uses:** PAM50 assay can aid profiling for both prognosis and prediction of benefit from adjuvant tamoxifen and has been found superior to immunohistochemistry.

**Sources:** [www.hci.utah.edu](http://www.hci.utah.edu)

**Marker (Medline Search):** PAM50 breast Intrinsic Classifier

**Organ (Medline Search):** breast cancer

**Medline Searches:** PAM50[All Fields] AND ("breast"[MeSH Terms] OR "breast"[All Fields]) AND Intrinsic[All Fields] AND Classifier[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=3**

**FDA approved:** No

## **Gene Test Information: Rotterdam Signature, breast cancer**

**Test Name:** Rotterdam Signature 76-Gene Panel

**Description:** The Rotterdam Signature test is a 76-gene expression assay. Sixty genes are intended to evaluate estrogen-receptor positive samples and 16 genes to evaluate estrogen-receptor negative samples. In a validation study that tested the signature on samples from 148 women, 50 fell into the low-risk group and 98 into the high-risk group. The test had 88% specificity and 39% sensitivity for the low-risk group, with a hazard ratio for distant relapse within 5 years of 5.74 comparing the high-risk group to the low-risk group.

**Purpose:** Prognostic, predictive, and recurrence

**Availability:** Veridex, LLC

**Specimen:** Tumor tissue

**Methodology:** Not reported

**Diseases:** breast cancer

**Clinical Uses:** Identifies women at high and low risk of disease recurrence.

**Sources:** [www.hci.utah.edu](http://www.hci.utah.edu)

**Marker (Medline Search):** Rotterdam signature

**Organ (Medline Search):** breast cancer

**Medline Searches:** rotterdam[All Fields] AND ("Signature"[Journal] OR "signature"[All Fields]) AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**7

**FDA approved:** No

## **Gene Test Information: SYMPHONY™, Breast Cancer**

### **Test Name: SYMPHONY™ Breast Cancer Profile**

**Description:** SYMPHONY™ provides complete tumor profiling and is used to support therapeutic choices for breast cancer. SYMPHONY includes four assays to support breast cancer treatment decisions: MammaPrint® determines the risk of recurrence. Blueprint™ determines molecular subtypes and TargetPrint® determines estrogen receptor (ER), progesterone receptor (PR), and HER2 status. TheraPrint™ identifies alternative types of therapy for metastatic disease.

**Purpose:** Diagnostic, prognostic, recurrence, and therapeutic management

**Availability:** Agendia

**Specimen:** formalin-fixed, paraffin-embedded tissue sections

**Methodology:** Panel of several genomic tests; microarray-based RNA gene expression methodology

**Diseases:** breast cancer

**Clinical Uses:** SYMPHONY provides genomic information assisting with therapeutic decisions even for cases that have been otherwise classified as indeterminate, such as grade 2, small tumors, HER2 and/or lymph node positive. MammaPrint® determines if the patient is a candidate for chemotherapy. TargetPrint® determines if the patient is a candidate for hormonal therapy. Blueprint® provides information on the sub-classification of the tumor which guides the choice of therapies and combinations of therapies. TheraPrint® identifies alternative types of therapy for metastatic disease.

**Sources:** [www.agendia.com](http://www.agendia.com)

Marker (Medline search): MammaPrint AND Blueprint AND TargetPrint AND TheraPrint

Organ (Medline search): breast cancer

**Medline Searches:** MammaPrint[Title] AND Blueprint[Title] AND TargetPrint[Title] AND breast[Title] AND cancer[Title]

**Medline hits=1**

**FDA approved:** No

**Gene Test Information: TargetPrint, breast cancer**

**Test Name: TargetPrint®, ER/PR/HER2 Expression Assay**

**Description:** TargetPrint is a microarray-based gene expression test which offers a quantitative assessment of the patient's level of estrogen receptor (ER), progesterone receptor (PR) and HER2/neu overexpression within her breast cancer. TargetPrint is offered in conjunction with MammaPrint to provide the physician an even more complete basis for treatment decisions.

**Purpose:** Therapeutic management

**Availability:** Agendia

**Specimen:** Formalin fixed paraffin-embedded, fresh or frozen breast tumor tissue

**Methodology:** Panel of three separate single gene readouts by microarray-based RNA gene expression

**Diseases:** Breast cancer

**Clinical Uses:** TargetPrint delivers an added benefit to the diagnostic process.

Immunohistochemistry provides a semi-quantitative positive or negative result, whereas the gene expression result provided by TargetPrint allows physicians to integrate the absolute level of ER, PR and HER2 gene expression into treatment planning. TargetPrint determines if the patient is a candidate for hormonal therapy.

**Sources:** [www.agendia.com](http://www.agendia.com)

**Marker (Medline Search):** TargetPrint and breast cancer

**Organ (Medline Search):** breast

**Medline Searches:** TargetPrint[All Fields] AND ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])

**Medline hits=**4

**FDA approved:** No

## **COLORECTAL CANCER**

**Gene Test Information:** ColoNext™

**Test Name:** ColoNext™

**Description:** ColoNext™ utilizes next generation sequencing to offer a comprehensive testing panel for hereditary colon cancer and targets detection of mutations in 14 genes (APC, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11, and TP53). Gross deletion/duplication analysis is performed for all 14 genes.

**Purpose:** Predictive

**Availability:** Ambry genetics

**Specimen:** Blood, DNA, or Saliva

**Methodology:** gene sequencing, deletion/duplication analysis

**Diseases:** Colon Cancer

**Clinical Uses:** ColoNext™ is a next-generation cancer panel that simultaneously analyzes selected genes associated with a wide range of cancers. While mutations in each gene on this panel may be individually rare, they may collectively account for a significant amount of hereditary cancer susceptibility.

**Sources:** [www.ambrygen.com/tests/colonext](http://www.ambrygen.com/tests/colonext)

**Marker (Medline Search):** (APC OR BMPR1A OR CDH1 OR CHEK2 OR EPCAM OR MLH1 OR MSH2 OR MSH6 OR MUTYH OR PMS2 OR PTEN OR SMAD4 OR STK11 OR TP53)

**Organ (Medline Search):** colon

**Medline Searches:** (APC[All Fields] OR ("bone morphogenetic protein receptors, type i"[MeSH Terms] OR "type i bone morphogenetic protein receptors"[All Fields] OR "bmp1a"[All Fields]) OR CDH1[All Fields] OR CHEK2[All Fields] OR ("tumor-associated antigen GA733"[Supplementary Concept] OR "tumor-associated antigen GA733"[All Fields] OR "epcam"[All Fields]) OR MLH1[All Fields] OR MSH2[All Fields] OR MSH6[All Fields] OR MUTYH[All Fields] OR PMS2[All Fields] OR PTEN[All Fields] OR SMAD4[All Fields] OR STK11[All Fields] OR TP53[All Fields]) AND ("colonic neoplasms"[MeSH Terms] OR ("colonic"[All Fields] AND "neoplasms"[All Fields]) OR "colonic neoplasms"[All Fields] OR ("colon"[All Fields] AND "cancer"[All Fields]) OR "colon cancer"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=**3646

**FDA approved:** No

## **Gene Test Information: ColoPrint®, Colon Cancer**

### **Test Name: ColoPrint® Colon Cancer Gene Expression Test**

**Description:** ColoPrint is an 18-gene profile that classifies colon cancer into Low Risk or High Risk of relapse, by measuring genes representative of the metastatic pathways of colon cancer metastases which were selected for their predictive relationship to 5-year distant metastases probability.

ColoPrint is indicated for stage II colon cancer, and provides relapse risk stratification independent of clinical and pathologic factors such as T4-stage and MSI status.

**Purpose:** Prognosis, recurrence, and therapeutic management of colon cancer

**Availability:** Agendia

**Specimen:** Fresh tumor tissue

**Methodology:** Genomic signature by microarray-based RNA gene expression

**Diseases:** Colon cancer

**Clinical Uses:** ColoPrint determines if the patient is a candidate for chemotherapy.

**Sources:** [www.Agendia.com](http://www.Agendia.com)

**Marker (Medline Search):** Coloprint

**Organ (Medline Search):** Colon

**Medline Searches:** Coloprint[All Fields] AND ("colonic neoplasms"[MeSH Terms] OR ("colonic"[All Fields] AND "neoplasms"[All Fields]) OR "colonic neoplasms"[All Fields] OR ("colon"[All Fields] AND "cancer"[All Fields]) OR "colon cancer"[All Fields])

**Medline hits=**3

**FDA approved:** No



## **Gene Test Information: ColoSeq™ - Lynch and Polyposis Syndrome Panel**

### **Test Name: ColoSeq™ - Lynch and Polyposis Syndrome Panel**

**Description:** ColoSeq™ is a comprehensive genetic test for hereditary colon cancer that uses next-generation sequencing to detect mutations in multiple genes associated with Lynch syndrome (hereditary non-polyposis colorectal cancer, HNPCC), familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP), hereditary diffuse gastric cancer (HDGC), Cowden syndrome, Li-Fraumeni syndrome, Peutz-Jeghers syndrome, Muir-Torre syndrome, Turcot syndrome, and Juvenile Polyposis syndrome. The assay sequences all exons, introns, and flanking sequences of the 13 genes. Large deletions, duplications, and mosaicism are also detected by the assay and reported.

**Purpose:** Prediction and diagnoses of hereditary Colon Cancer

**Availability:** University of Washington Laboratory Medicine- Genetics Lab

**Specimen:** Blood

**Methodology:** RNA, Gene Sequencing

**Diseases:** Lynch syndrome (hereditary non-polyposis colorectal cancer, HNPCC), familial adenomatous polyposis (FAP), MUTYH-associated polyposis (MAP), hereditary diffuse gastric cancer (HDGC), Cowden syndrome, Li-Fraumeni syndrome, Peutz-Jeghers syndrome, Muir-Torre syndrome, Turcot syndrome, and Juvenile Polyposis syndrome

**Clinical Uses:** To determine if a person is at risk of colon cancer.

**Sources:** [www.web.labmed.washington.edu](http://www.web.labmed.washington.edu)

**Marker:** MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, CDH1, PTEN, STK11, TP53, SMAD4, and BMPR1A.

**Organ:** Colon, Rectum

**Medline Searches:** (MLH1[All Fields] OR MSH2[All Fields] OR MSH6[All Fields] OR PMS2[All Fields] OR ("tumor-associated antigen GA733"[Supplementary Concept] OR "tumor-associated antigen GA733"[All Fields] OR "epcam"[All Fields]) OR APC[All Fields] OR MUTYH[All Fields] OR CDH1[All Fields] OR PTEN[All Fields] OR STK11[All Fields] OR TP53[All Fields] OR SMAD4[All Fields] OR ("bone morphogenetic protein receptors, type i"[MeSH Terms] OR "type i bone morphogenetic protein receptors"[All Fields] OR "bmpr1a"[All Fields])) AND ("colorectal neoplasms, hereditary nonpolyposis"[MeSH Terms] OR ("colorectal"[All Fields] AND "neoplasms"[All Fields] AND "hereditary"[All Fields] AND "nonpolyposis"[All Fields]) OR "hereditary nonpolyposis colorectal neoplasms"[All Fields] OR ("lynch"[All Fields] AND "syndrome"[All Fields]) OR "lynch syndrome"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=1698**

**FDA approved: No**

## **Gene Test Information: GCC (GUCY2C) Blood Test, Colorectal Cancer Staging Test**

### **Test Name: Guanylyl Cyclase C (GUCY2C)**

**Description:** Guanylyl Cyclase C (GCC or GUCY2C) a gene coding for a protein found in cells, lining the intestine from the duodenum to the rectum. It is involved in water transport, crypt morphology and suppression of tumorigenesis. It is not normally found in tissue in other parts of the body, and therefore, GCC detected outside of the intestine, indicates presence of colorectal cancer metastases. Early studies have indicated that the presence of GCC in the blood may be an early indicator of micrometastases that would otherwise escape detection by the current standard methods of monitoring. Earlier detection provides an opportunity for more immediate treatment or surgical intervention to potentially improve patient outcomes and survival rates.

**Purpose:** Diagnostic test

**Availability:** Diagnocure

**Specimen:** Whole Blood

**Methodology:** Reverse Transcriptase–Polymerase Chain Reaction

**Diseases:** Colorectal Cancer

**Clinical Uses:** Diagnostic test for recurrence by identification of micrometastasis in the blood

**Sources:** [www.diagnocure.com](http://www.diagnocure.com)

**Marker (Medline Search):** Guanylyl Cyclase C AND Blood

**Organ (Medline Search):** Colorectal cancer

**Medline Searches:** ("enterotoxin receptor"[Supplementary Concept] OR "enterotoxin receptor"[All Fields] OR "guanylyl cyclase c"[All Fields]) AND ("blood"[Subheading] OR "blood"[All Fields] OR "blood"[MeSH Terms]) AND ("colorectal neoplasms"[MeSH Terms] OR ("colorectal"[All Fields] AND "neoplasms"[All Fields]) OR "colorectal neoplasms"[All Fields] OR ("colorectal"[All Fields] AND "cancer"[All Fields]) OR "colorectal cancer"[All Fields])

**Medline hits=** 22

**FDA approved:** No

**Gene Test Information: ResponseDX: Colon®**

**Test Name: ResponseDX: Colon®**

**Description:** ResponseDX: Colon® panel utilizes testing of multiple genes including KRAS mutation, BRAF mutation, ERCC1 expression, MSI, c-Met expression, EGFR expression, VEGFR2 expression, NRAS mutation, PIK3CA mutation, and Thymidylate synthetase.

**Purpose:** Therapeutic Management

**Availability:** Response Genetics

**Specimen:** Formalin-fixed, paraffin embedded (FFPE) biopsy specimen

**Methodology:** PCR-based tests

**Diseases:** Metastatic colorectal cancer

**Clinical Uses:** Predict disease prognosis and select patients who might benefit from alternative therapies and aids in selection of metastatic colorectal cancer patients that might benefit from EGFR-targeted monoclonal antibody therapies.

**Sources:** <http://www.responsegenetics.com/products-services/respondedx-colon/>

**Marker (Medline Search):** (KRAS OR BRAF OR ERCC1 OR MSI OR c-MET OR EGFR OR VEGFR2 OR PIK3CA OR NRAS)

**Organ (Medline Search):** Colon

**Medline Searches:** (("proto-oncogene proteins p21(ras)"[MeSH Terms] OR ("proto-oncogene"[All Fields] AND "proteins"[All Fields] AND "p21(ras)"[All Fields]) OR "proto-oncogene proteins p21(ras)"[All Fields] OR "kras"[All Fields]) OR BRAF[All Fields] OR ERCC1[All Fields] OR ("N-methylsuccinimide"[Supplementary Concept] OR "N-methylsuccinimide"[All Fields] OR "msi"[All Fields]) OR c-MET[All Fields] OR EGFR[All Fields] OR VEGFR2[All Fields] OR PIK3CA[All Fields] OR NRAS[All Fields]) AND ("colonic neoplasms"[MeSH Terms] OR ("colonic"[All Fields] AND "neoplasms"[All Fields]) OR "colonic neoplasms"[All Fields] OR ("colon"[All Fields] AND "cancers"[All Fields]) OR "colon cancers"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=** 1437

**FDA approved:** No

**Gene Test Information: *Therascreen* KRAS RGQ PCR, colorectal cancer**

**Test Name:** *Therascreen* KRAS RGQ PCR

**Description:** *Therascreen* KRAS RGQ PCR Kit is intended to detect 7 mutations in codons 12 and 13 of the KRAS gene. The kit utilizes two technologies — ARMS and Scorpions — for detection of mutations in real-time PCR.

**Purpose:** Diagnosis and therapeutic management

**Availability:** QIAGEN

**Specimen:** Whole blood

**Methodology:** Real-time PCR.

**Diseases:** Colorectal cancer

**Clinical Uses:** The *therascreen* KRAS RGQ PCR kit is being developed as a companion diagnostic to aid clinicians, through detection of KRAS mutations, in the identification of patients with metastatic colorectal cancer (mCRC) who are more likely to benefit from cetuximab.

**Sources:** [www.qiagen.com](http://www.qiagen.com)

**Marker (Medline Search):** *therascreen* kras rgq

**Organ (Medline Search):** colorectal

**Medline Searches:** *therascreen*[All Fields] AND ("proto-oncogene proteins p21(ras)"[MeSH Terms] OR ("proto-oncogene"[All Fields] AND "proteins"[All Fields] AND "p21(ras)"[All Fields]) OR "proto-oncogene proteins p21(ras)"[All Fields] OR "kras"[All Fields]) AND rgq[All Fields] AND ("colorectal neoplasms"[MeSH Terms] OR ("colorectal"[All Fields] AND "neoplasms"[All Fields]) OR "colorectal neoplasms"[All Fields] OR ("colorectal"[All Fields] AND "cancer"[All Fields]) OR "colorectal cancer"[All Fields])

**Medline hits=**1

**FDA approved:** yes

## **Gene Test Information: PMS2, hereditary nonpolyposis colorectal cancer**

**Test Name:** PMS2

**Description:** PMS2 test covers all coding nucleotides of gene Postmeiotic Segregation Increased, *S. Cerevisiae*, 2(PMS2), plus at least two and typically 20 flanking intronic nucleotides upstream and downstream of each coding exon, covering the conserved donor and acceptor splice sites, as well as typically 20 flanking nucleotides in the 5' and 3' UTR. This test can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures.

**Purpose:** Diagnosis

**Availability:** LabCorp

**Specimen:** Whole blood, DNA is accepted.

**Methodology:** DNA sequencing

**Diseases:** Hereditary nonpolyposis colorectal cancer

**Clinical Uses:** can confirm a clinical diagnosis of HNPCC and allow early diagnosis in family members, guiding preventive measures

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** PMS2 gene sequencing and

**Organ (Medline Search):** hereditary nonpolyposis colorectal cancer

**Medline Searches:** PMS2[All Fields] AND ("genes"[MeSH Terms] OR "genes"[All Fields] OR "gene"[All Fields]) AND sequencing[All Fields] AND ("colorectal neoplasms, hereditary nonpolyposis"[MeSH Terms] OR ("colorectal"[All Fields] AND "neoplasms"[All Fields] AND "hereditary"[All Fields] AND "nonpolyposis"[All Fields]) OR "hereditary nonpolyposis colorectal neoplasms"[All Fields] OR ("hereditary"[All Fields] AND "nonpolyposis"[All Fields] AND "colorectal"[All Fields] AND "cancer"[All Fields]) OR "hereditary nonpolyposis colorectal cancer"[All Fields])

**Medline hits=**28

**FDA approved:** No

## **Gene Test Information: Previstage™ GCC, Colorectal Cancer Staging Test**

### **Test Name: Previstage™ Guanylyl Cyclase C (GCC or GUCY2C)**

**Description:** Guanylyl Cyclase C (GCC or GUCY2C) a gene coding for a protein found in cells, lining the intestine from the duodenum to the rectum. It is involved in water transport, crypt morphology and suppression of tumorigenesis. It is not normally found in tissue in other parts of the body, and therefore, GCC detected outside of the intestine, indicates presence of colorectal cancer metastases. GCC mRNA has shown to be highly accurate in detecting the spread and recurrence of colorectal cancer, respectively in lymph nodes and blood, thereby representing a significant improvement over traditional detection methods.

**Purpose:** Predictive test, Prognostic marker

**Availability:** Diagnocure

**Specimen:** lymph nodes

**Methodology:** Reverse Transcriptase–Polymerase Chain Reaction

**Diseases:** Colorectal Cancer

**Clinical Uses:** Predictive test for risk stratification of recurrence and prognostic marker for recurrence

**Sources:** [www.diagnocure.com](http://www.diagnocure.com)

**Marker (Medline Search):** Guanylyl Cyclase C

**Organ (Medline Search):** Colorectal cancer

**Medline Searches:** ("enterotoxin receptor"[Supplementary Concept] OR "enterotoxin receptor"[All Fields] OR "guanylyl cyclase c"[All Fields]) AND ("lymph nodes"[MeSH Terms] OR ("lymph"[All Fields] AND "nodes"[All Fields]) OR "lymph nodes"[All Fields] OR ("lymph"[All Fields] AND "node"[All Fields]) OR "lymph node"[All Fields]) AND ("colorectal neoplasms"[MeSH Terms] OR ("colorectal"[All Fields] AND "neoplasms"[All Fields]) OR "colorectal neoplasms"[All Fields] OR ("colorectal"[All Fields] AND "cancer"[All Fields]) OR "colorectal cancer"[All Fields])

**Medline hits=** 29

**FDA approved:** No

**GENITOURINARY**



## **Gene Test Information: Onco FISH, cervical**

### **Test Name: Onco FISH cervical**

**Description:** Among the many chromosomal changes observed in cervical cancer, the most consistent abnormality is detected in chromosome arm 3q.2. Studies have shown that at least 90% of invasive cervical cancer cases have a gain in the 3q arm.<sup>3,4</sup> Additional research has demonstrated a correlation between the gain in the 3q26 copy number as the severity and stage of cervical disease progression.

**Purpose:** Predictive

**Availability:** Ikonisys

**Specimen:** Liquid cytology specimens

**Methodology:** Fluorescence in situ Hybridization

**Diseases:** cervical cancer (precancerous to malignancy cervical cancer)

**Clinical Uses:** Using this technology to look at the progression of individual patients, it has been shown that the sensitivity of the 3q26 loci for predicting progression from CIN1/CIN2 to CIN3 was 100% and the specificity, i.e., the prediction of regression, was 70%.

**Sources:** [www.ikonisys.com](http://www.ikonisys.com)

**Marker (Medline Search):** 3q26 AND Fluorescence in situ Hybridization

**Organ (Medline Search):** cervical

**Medline Searches:** 3q26[All Fields] AND ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("uterine cervical neoplasms"[MeSH Terms] OR ("uterine"[All Fields] AND "cervical"[All Fields] AND "neoplasms"[All Fields]) OR "uterine cervical neoplasms"[All Fields] OR ("cervical"[All Fields] AND "cancer"[All Fields]) OR "cervical cancer"[All Fields])

**Medline hits=**24

**FDA approved:** Not Reported

## **Gene Test Information: UteroFISH, Uterine**

### **Test Name: UteroFISH**

**Description:** UteroFISH helps distinguish between atypia or hyperplasia on an endometrial biopsy diagnosis which can have a large affect on how physicians will treat their patients. An abnormal UteroFISH result indicates a high risk (>89%) for cancer or atypical hyperplasia, and a true positive rate of 81% in patients with cancer/atypia.

**Purpose:** provide help on how physicians will treat their patients, therapeutic purpose

**Availability:** Gynecor

**Specimen:** biopsy

**Methodology:** Fluorescence in situ Hybridization

**Diseases:** Uterine cancer

**Clinical Uses:** The UteroFISH test results may aid to establish atypia or cancer. The test result infromation can save a woman from having to undergo an unnecessary hysterectomy.

**Sources:** [www.gynecor.com](http://www.gynecor.com)

**Marker (Medline Search):** Utero Fluorescence in situ Hybridization

**Organ (Medline Search):** Uterine cancer

**Medline Searches:** ("uterus"[MeSH Terms] OR "uterus"[All Fields] OR "utero"[All Fields]) AND ("in situ hybridization, fluorecence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("uterine neoplasms"[MeSH Terms] OR ("uterine"[All Fields] AND "neoplasms"[All Fields]) OR "uterine neoplasms"[All Fields] OR ("uterine"[All Fields] AND "cancer"[All Fields]) OR "uterine cancer"[All Fields])

**Medline hits=**58

**FDA approved:** No

**Gene Test Information: UroVysion FISH, bladder cancer**

**Test Name: UroVysion Fluorescence in situ hybridization (FISH) analysis**

**Description:** The UroVysion Bladder Cancer Kit (UroVysion Kit) is designed to detect aneuploidy for chromosomes 3, 7, 17, and loss of the 9p21 locus via fluorescence in situ hybridization (FISH) in urine specimens from persons with hematuria suspected of having bladder cancer. FISH analysis is used in conjunction with cystoscopy to monitor for recurrence among those with previously diagnosed bladder cancer.

**Purpose:** Diagnosis and monitoring for recurrence

**Availability:** Abbott, Baycare Laboratories

**Specimen:** Urine

**Methodology:** Fluorescence in situ hybridization (FISH)

**Diseases:** Bladder cancer

**Clinical Uses:** FISH analysis is a surveillance tool in established primary and secondary bladder adenocarcinoma.

**Sources:** [www.abbottmolecular.com](http://www.abbottmolecular.com); [www.baycare.org/laboratories](http://www.baycare.org/laboratories)

**Marker (Medline Search):** Fluorescence in situ hybridization

**Organ (Medline Search):** bladder cancer

**Medline Searches:** ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("urinary bladder neoplasms"[MeSH Terms] OR ("urinary"[All Fields] AND "bladder"[All Fields] AND "neoplasms"[All Fields]) OR "urinary bladder neoplasms"[All Fields] OR ("bladder"[All Fields] AND "cancer"[All Fields]) OR "bladder cancer"[All Fields])

**Medline hits=**469

**FDA approved:** Yes

## **HEMATOLOGICAL**

**Gene Test Information: 5q del, 7q del/-7 FISH test, Acute myeloid leukemia and myelodysplastic syndrome**

**Test Name: 5q del, 7q del/-7 FISH test**

**Description:** Chromosomal abnormalities are detected in 40-60% of patients with de novo myelodysplastic syndromes (MDS). MDS with interstitial deletion of a segment of the long arm of chromosome 5q [del(5q)] as an isolated cytogenetic abnormality is characterized by bone marrow erythroid hyperplasia, atypical megakaryocytes, thrombocytopenia, refractory anemia, and low risk of progression to acute myeloid leukemia (AML) compared with other types of MDS. In published studies Presence of -7/7q- was associated with shorter overall survival than absence of such aberrations. FISH 7q could be beneficial in patients with intermediate WHO morphologic risk stratification and no evidence of -7/7q- by cytogenetics.

**Purpose:** Prognostic and therapeutic management.

**Availability:** Arup laboratories and other laboratories such as community, academic, and other commercial laboratories.

**Specimen:** Bone marrow or whole blood

**Methodology:** Fluorescence in-situ hybridization

**Diseases:** Acute myelogenous leukemia with myelodysplastic syndrome

**Clinical Uses:** The 5q del, 7q del/-7 FISH test may aid in prognosis of acute myeloid leukemia and myelodysplastic syndrome. It helps a subset of patients get treatment tailored to their unique genetic profile

**Sources:** [www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov), [www.aruplab.com](http://www.aruplab.com)

**Marker (Medline Search):** 5q del OR -7/7q del

**Organ (Medline Search):** Acute myeloid leukemia AND myelodysplastic syndrome

**Medline Searches:** ((5q[All Fields] AND del[All Fields]) OR (7q[All Fields] AND del[All Fields])) AND (("leukemia, myeloid, acute"[MeSH Terms] OR ("leukemia"[All Fields] AND "myeloid"[All Fields] AND "acute"[All Fields]) OR "acute myeloid leukemia"[All Fields] OR ("acute"[All Fields] AND "myeloid"[All Fields] AND "leukemia"[All Fields])) AND ("myelodysplastic syndromes"[MeSH Terms] OR ("myelodysplastic"[All Fields] AND "syndromes"[All Fields]) OR "myelodysplastic syndromes"[All Fields] OR ("myelodysplastic"[All Fields] AND "syndrome"[All Fields]) OR "myelodysplastic syndrome"[All Fields]))

**Medline hits=**111

**FDA approved:** No

## Gene Test Information: 17p deletion

### Test Name: 17p deletion

**Description:** The deletion of 17p is a bad prognosis factor for patients with Chronic Lymphocytic Leukemia. del(17p) typically involves TP53 locus and del(11q) contains ATM gene, both of which are tumor suppressors. Loss of p53 function or its activator, the ATM gene, is associated with treatment resistance and clinically aggressive disease.

**Purpose:** Prognosis of Chronic Lymphocytic Leukemia

**Availability:** Multiple Laboratories, including Arup Laboratories and Abbott Laboratories

**Specimen:** Blood

**Methodology:** Fluorescence in situ hybridization (FISH)

**Diseases:** Chronic lymphocytic leukemia (CLL)

**Clinical Uses:** Predicts poor prognosis of Chronic Lymphocytic Leukemia

**Sources:** [www.aruplab.com](http://www.aruplab.com), [www.abbottmolecular.com](http://www.abbottmolecular.com)

**Marker:** 17p deletion

**Organ:** Chronic Lymphocytic Leukemia

**Medline Searches:** (17[All Fields] AND p[All Fields] AND ("sequence deletion"[MeSH Terms] OR ("sequence"[All Fields] AND "deletion"[All Fields]) OR "sequence deletion"[All Fields] OR "deletion"[All Fields])) AND ("chronic lymphocytic leukaemia"[All Fields] OR "leukemia, lymphocytic, chronic, b-cell"[MeSH Terms] OR ("leukemia"[All Fields] AND "lymphocytic"[All Fields] AND "chronic"[All Fields] AND "b-cell"[All Fields]) OR "b-cell chronic lymphocytic leukemia"[All Fields] OR ("chronic"[All Fields] AND "lymphocytic"[All Fields] AND "leukemia"[All Fields]) OR "chronic lymphocytic leukemia"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=**59

**FDA approved:** No

## **Gene Test Information: Multiple myeloma panel by FISH**

### **Test Name: Multiple myeloma panel FISH test**

**Description:** Chromosomal abnormalities are important prognostic indicators in multiple myeloma. In this test, fluorescence in situ hybridization (FISH) panel is performed on bone marrow or tissue containing neoplastic plasma cells for multiple myeloma prognosis-specific genomic abnormalities: for example, *CKS1B* (1q gain), *ASS1* (+9), *CCND1/IGH* (*IGH/CCND1* fusion or +11), *IGH* rearrangement, *PML* (+15) and *p53* (17p deletion).

**Purpose:** Prognostic and therapeutic management.

**Availability:** Arup laboratories and other laboratories such as community, academic, and other commercial laboratories.

**Specimen:** Bone marrow or whole blood

**Methodology:** Fluorescence in-situ hybridization

**Diseases:** Multiple myeloma

**Clinical Uses:** The FISH test may aid in prognosis of multiple myeloma. It helps a subset of patients get treatment tailored to their unique genetic profile

**Sources:** [www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov), [www.aruplab.com](http://www.aruplab.com)

**Marker (Medline Search):** CD138+ OR CD138 positive

**Organ (Medline Search):** Multiple myeloma

**Medline Searches:** ("syndecan-1"[MeSH Terms] OR "syndecan-1"[All Fields] OR "cd138"[All Fields]) AND ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("multiple myeloma"[MeSH Terms] OR ("multiple"[All Fields] AND "myeloma"[All Fields]) OR "multiple myeloma"[All Fields])

**Medline hits=**35

**FDA approved:** No

**Gene Test Information: MyPRS Plus****Test Name: MyPRS Plus**

**Description:** MyPRS Plus analyzes all of the nearly 25,000 genes in a patient's genome to determine the gene expression profile that is associated with their condition. In the case of myeloma, the gene expression profile is made up of the 70 most relevant genes which aid in the prediction of the patient's outcome.

**Purpose:** Prognosis and Therapeutic management of Myeloma

**Availability:** Signal Genetics

**Specimen:** Bone Marrow Aspirate

**Methodology:** Gene Sequencing

**Diseases:** Myeloma

**Clinical Uses:** Helps patients and physicians determine the best treatment for patients with Myeloma.

**Sources:** [www.signalgenetics.com](http://www.signalgenetics.com),  
[www.anthem.com/medicalpolicies/policies/mp\\_pw\\_c139353.htm](http://www.anthem.com/medicalpolicies/policies/mp_pw_c139353.htm)

**Marker (Medline Search):** Not reported

**Organ (Medline Search):** Bone

**Medline Searches:** MyPRS AND Plus AND myeloma

**Medline hits= No PubMed citations found.**

**FDA approved:** No



**Lung**

## **Gene Test Information: ALK FISH, Lung cancer**

### **Test Name: ALK FISH**

**Description:** A rearrangement of anaplastic lymphoma kinase (ALK) is reported to be associated with the development of non-small-cell lung cancer (NSCLC)

**Purpose:** Diagnosis and therapeutic management

**Availability:** LabCorp

**Specimen:** Fixed cell pellet from a cytogenetic analysis

**Methodology:** Polymerase chain reaction (PCR), direct DNA sequencing, fluorescence in situ hybridization (FISH)

**Diseases:** Lung cancer

**Clinical Uses:** ALK FISH probe is used to identify gene rearrangements involving the ALK gene in patients with NSCLC who are eligible for treatment with crizotinib.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker:** ALK fluorescence in situ hybridization

**Organ:** lung

**Medline Searches:** ALK[All Fields] AND ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("lung neoplasms"[MeSH Terms] OR ("lung"[All Fields] AND "neoplasms"[All Fields]) OR "lung neoplasms"[All Fields] OR ("lung"[All Fields] AND "cancer"[All Fields]) OR "lung cancer"[All Fields])

**Medline hits=**65

**FDA approved:** Yes

**Gene Test Information: ResponseDX:Lung®**

**Test Name: ResponseDX:Lung®**

**Description:** ResponseDX:Lung® panel utilizes testing of multiple genes including ROS1 rearrangements, EGFR mutation, EML4-ALK rearrangement, ALK, ERCC1 expression, RRM1 expression, c-MET expression, TS expression, KRAS mutation, and PIK3CA mutation.

**Purpose:** Therapeutic Management

**Availability:** Responsegenetics.com

**Specimen:** Formalin-fixed, paraffin embedded (FFPE) biopsy specimen

**Methodology:** Fluorescence in situ hybridization (FISH) and polymerase chain reaction (PCR)

**Diseases:** Non-small cell lung cancers

**Clinical Uses:** Patients with lung cancer who are being considered for treatment with the tyrosine kinase inhibitor (TKI) crizotinib

**Sources:** [www.responsegenetics.com](http://www.responsegenetics.com)

**Marker (Medline Search):** ROS1 OR EGFR mutation OR EML4-ALK OR ALK OR ERCC1 OR RRM1 OR c-MET OR TS OR KRAS mutation OR PIK3CA mutation

**Organ (Medline Search):** Non-small cell lung cancers

**Medline Searches:** (ROS1[All Fields] OR EGFR[All Fields] OR EML4-ALK[All Fields] OR ALK[All Fields] OR ERCC1[All Fields] OR RRM1[All Fields] OR c-MET[All Fields] OR TS[All Fields] OR ("proto-oncogene proteins p21(ras)"[MeSH Terms] OR ("proto-oncogene"[All Fields] AND "proteins"[All Fields] AND "p21(ras)"[All Fields]) OR "proto-oncogene proteins p21(ras)"[All Fields] OR "kras"[All Fields]) OR PIK3CA[All Fields]) AND (Non-small[All Fields] AND ("cells"[MeSH Terms] OR "cells"[All Fields] OR "cell"[All Fields]) AND ("lung neoplasms"[MeSH Terms] OR ("lung"[All Fields] AND "neoplasms"[All Fields]) OR "lung neoplasms"[All Fields] OR ("lung"[All Fields] AND "cancers"[All Fields]) OR "lung cancers"[All Fields])) AND "humans"[MeSH Terms]

**Medline hits=** 1970

**FDA approved:** No

## **Gene Test Information: Vysis ALK FISH test, Lung cancer**

### **Test Name: Vysis ALK FISH test**

**Description:** Anaplastic lymphoma kinase (ALK) encodes a receptor tyrosine kinase, and ALK gene rearrangement (ALK+) is implicated in the oncogenesis of non-small cell lung carcinomas (NSCLCs), especially adenocarcinomas. The ALK inhibitor crizotinib was approved in August 2011 by the US Food and Drug Administration (FDA) for treating late-stage NSCLCs that are ALK+, with a companion fluorescent in situ hybridization (FISH) test using the Vysis ALK Break Apart fluorescence in-situ hybridization (FISH) Probe Kit. The Abbott ALK test has been designed to identify those patients - about 3 to 5 percent of NSCLC patients - who would be candidates for the new drug.

**Purpose:** Therapeutic management

**Availability:** Abbott

**Specimen:** Not reported

**Methodology:** Fluorescence in-situ hybridization

**Diseases:** Non-small-cell lung cancer

**Clinical Uses:** The Vysis ALK FISH test uses Abbott's fluorescence in situ hybridization (FISH) technology to detect rearrangements of the ALK gene on the 2p23 chromosome. It helps a subset of lung-cancer patients get treatment tailored to their unique genetic profile

**Sources:** [www.AbbottALK.com](http://www.AbbottALK.com)

**Marker (Medline Search):** ALK fluorescence in-situ hybridization and lung cancer

**Organ (Medline Search):** lung

**Medline Searches:** ALK[All Fields] AND ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND ("lung neoplasms"[MeSH Terms] OR ("lung"[All Fields] AND "neoplasms"[All Fields]) OR "lung neoplasms"[All Fields] OR ("lung"[All Fields] AND "cancer"[All Fields]) OR "lung cancer"[All Fields])

**Medline hits=**64

**FDA approved:** Yes

## Gene Test Information: PreOvar

### Test Name: *PreOvar*

**Description:** PreOvar™ tests for the KRAS-variant, and will help identify ovarian cancer patients whose female relatives should also be evaluated for the KRAS-variant. PreOvar™ may also help assess the relative risk of developing ovarian cancer for women who have a family history of ovarian cancer without a living proband (ancestor with the disease). The KRAS-Variant is present in 6-10% of the general population and 25% of non-selected women with epithelial ovarian cancer. Additionally, the KRAS-variant was identified in over 60% of Hereditary Breast and Ovarian Cancer (HBOC) patients that were previously classified as “uninformative,” or negative for other known genetic markers of ovarian cancer risk.

**Purpose:** Prediction of the risk of developing ovarian cancer.

**Availability:** Mira DX

**Specimen:** Blood, Saliva

**Methodology:** RNA

**Diseases:** Ovarian Cancer

**Clinical Uses:** Test determines if KRAS-variant may put someone at increased risk for developing ovarian cancer.

**Sources:** [www.miradx.com/preovar](http://www.miradx.com/preovar)

**Marker:** KRAS-variant

**Organ:** Ovaries

**Medline Searches:** (("proto-oncogene proteins p21(ras)"[MeSH Terms] OR ("proto-oncogene"[All Fields] AND "proteins"[All Fields] AND "p21(ras)"[All Fields]) OR "proto-oncogene proteins p21(ras)"[All Fields] OR "kras"[All Fields]) AND variant[All Fields]) AND ("ovarian neoplasms"[MeSH Terms] OR ("ovarian"[All Fields] AND "neoplasms"[All Fields]) OR "ovarian neoplasms"[All Fields] OR ("ovarian"[All Fields] AND "cancer"[All Fields]) OR "ovarian cancer"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=**9

**FDA approved:** No

## **PROSTATE**

**Gene Test Information: Adenomatous polyposis coli (APC), Prostate cancer**

**Test Name: Adenomatous polyposis coli (APC)**

**Description:** Prostate cancer is the most common cancer in men and the second leading cause of cancer-related deaths in the US. Due to the reportedly high false-negative rate of initial biopsy results after elevated PSA level, new approaches for improved detection in prostate cancer are needed. Several studies have shown that hypermethylation of the promoter regions of the GST-P1 and APC genes occurs at a significantly higher frequency in prostate cancer samples than in benign conditions of the prostate gland.

**Purpose:** Prognostic

**Availability:** Labcorp

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue

**Methodology:** Quantitative methylation-specific polymerase chain reaction (PCR)

**Diseases:** Prostate cancer

**Clinical Uses:** Gene methylation assays of Adenomatous polyposis coli (APC) may be used as an adjunct to histopathology for patients where prostate disease is considered.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** adenomatous polyposis coli and polymerase chain reaction

**Organ (Medline Search):** prostate

**Medline Searches:** ("adenomatous polyposis coli"[MeSH Terms] OR ("adenomatous"[All Fields] AND "polyposis"[All Fields] AND "coli"[All Fields]) OR "adenomatous polyposis coli"[All Fields]) AND ("polymerase chain reaction"[MeSH Terms] OR ("polymerase"[All Fields] AND "chain"[All Fields] AND "reaction"[All Fields]) OR "polymerase chain reaction"[All Fields]) AND ("prostatic neoplasms"[MeSH Terms] OR ("prostatic"[All Fields] AND "neoplasms"[All Fields]) OR "prostatic neoplasms"[All Fields] OR ("prostate"[All Fields] AND "cancer"[All Fields]) OR "prostate cancer"[All Fields])

Medline hits=14

**FDA approved:** No

## **Gene Test Information: Glutathione-S-Transferase (GST-P1), Prostate cancer**

### **Test Name: Glutathione-S-Transferase (GST-P1)**

**Description:** Prostate cancer is the most common cancer in men and the second leading cause of cancer-related deaths in the US. Due to the reportedly high false-negative rate of initial biopsy results after elevated PSA level, new approaches for improved detection in prostate cancer are needed. Several studies have shown that hypermethylation of the promoter regions of the GST-P1 and APC genes occurs at a significantly higher frequency in prostate cancer samples than in benign conditions of the prostate gland.

**Purpose:** Prognostic

**Availability:** Labcorp

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue

**Methodology:** Quantitative methylation-specific polymerase chain reaction (PCR)

**Diseases:** Prostate cancer

**Clinical Uses:** Hypermethylation of the promoter regions of the GST-P1 and APC genes can aid in prognosticating for prostate cancer

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** Glutathione-S-Transferase and polymerase chain reaction

**Organ (Medline Search):** Prostate

**Medline Searches:** ("glutathione transferase"[MeSH Terms] OR ("glutathione"[All Fields] AND "transferase"[All Fields]) OR "glutathione transferase"[All Fields] OR "glutathione s transferase"[All Fields]) AND ("polymerase chain reaction"[MeSH Terms] OR ("polymerase"[All Fields] AND "chain"[All Fields] AND "reaction"[All Fields]) OR "polymerase chain reaction"[All Fields]) AND ("prostatic neoplasms"[MeSH Terms] OR ("prostatic"[All Fields] AND "neoplasms"[All Fields]) OR "prostatic neoplasms"[All Fields] OR ("prostate"[All Fields] AND "cancer"[All Fields]) OR "prostate cancer"[All Fields])

**Medline hits=**118

**FDA approved:** No



## **Gene Test Information: Oncotype DX Prostate**

### **Test Name: Oncotype DX Prostate**

**Description:** The Oncotype DX test for prostate cancer is a genomic test that determines the risk of the cancer before treatment begins. The test predicts how likely it is that the cancer is low risk and contained within the prostate, or higher risk and more likely to grow and spread. With this information, the patient and their doctor can choose the most appropriate treatment option. For example, a lower risk prostate cancer with more favorable pathology, one that may not need invasive treatment and can be safely managed through close and careful monitoring – a treatment approach called active surveillance.

This genomic test measures biology through the expression of 17 genes across multiple key biological pathways in prostate cancer which can predict the aggressiveness of prostate cancer providing a individualized risk assessment.

**Purpose:** Therapeutic management of early-stage prostate cancer

**Availability:** Genomic Health, Inc.

**Specimen:** Tissue

**Methodology:** Immunohistochemistry

**Diseases:** Prostate Cancer

**Clinical Uses:** The results of this test are combined with PSA level and Gleason Score to determine the best treatment (Active Surveillance or Immediate Treatment) for patients with early-stage prostate cancer.

**Sources:** [www.genomichealth.com/OncotypeDX](http://www.genomichealth.com/OncotypeDX)

**Marker:** FAM13C, KLK2, AZGP1, SRD5A2, BGN, COL1A1, SFRP4, ARF, ATP5E, CLTC, GPS1, PGK1, FLNC, GSN, TPM2, GSTM2, TPX2

**Organ:** Prostate

**Medline Searches:** 4-dehydrogenase"[All Fields] OR "srd5a2"[All Fields]) OR BGN[All Fields] OR COL1A1[All Fields] OR SFRP4[All Fields] OR ARF[All Fields] OR ATP5E[All Fields] OR CLTC[All Fields] OR GPS1[All Fields] OR PGK1[All Fields] OR FLNC[All Fields] OR GSN[All Fields] OR TPM2[All Fields] OR GSTM2[All Fields] OR TPX2[All Fields]) AND ("prostatic neoplasms"[MeSH Terms] OR ("prostatic"[All Fields] AND "neoplasms"[All Fields]) OR "prostatic neoplasms"[All Fields] OR ("prostate"[All Fields] AND "cancer"[All Fields]) OR "prostate cancer"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=**402

**FDA approved:** No

## **Gene Test Information: ProstaVysion, Prostate Cancer**

### **Test Name: ProstaVysion**

**Description:** ProstaVysion is a prognostic genetic panel for prostate cancer. This test examines two major mechanisms of prostate carcinogenesis: ERG gene fusion/translocation and the loss of the PTEN tumor suppressor gene. This test is a tissue-based panel. By examining these two markers, ProstaVysion is able to provide a molecular analysis of prostate cancer aggressiveness and long-term patient prognosis. ERG gene fusions are found in 40% of primary prostate cancers and are associated with a more aggressive phenotype. Deletion of PTEN occurs in both localized prostate cancers and 60% of metastases.

**Purpose:** Prognosis and therapeutic management

**Availability:** Bostwick Labs

**Specimen:** Not reported

**Methodology:** Not reported

**Diseases:** Prostate cancer

**Clinical Uses:** ProstaVysion panel markers provide prognostic information and aid in therapeutic choices

**Sources:** [www.bostwicklaboratories.com](http://www.bostwicklaboratories.com)

Marker: ERG AND PTEN loss

Organ: Prostate cancer

**Medline Searches:** erg[All Fields] AND pten[All Fields] AND loss[All Fields] AND ("prostatic neoplasms"[MeSH Terms] OR ("prostatic"[All Fields] AND "neoplasms"[All Fields]) OR "prostatic neoplasms"[All Fields] OR ("prostate"[All Fields] AND "cancer"[All Fields]) OR "prostate cancer"[All Fields])

**Medline hits=**19

**FDA approved:** Not reported

**Other**

**Gene Test Information: Cobas® BRAF V600 mutation, Melanoma**

**Test Name: Cobas® BRAF V600 Mutation**

**Description:** Somatic mutations in the BRAF oncogene are frequently found in human cancers. These mutations are common in melanomas, colorectal cancer, lung cancer, ovarian cancer, and thyroid gland cancer.

**Purpose:** Therapeutic management

**Availability:** Roche diagnostics

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) melanoma tissue from humans.

**Methodology:** Polymerase chain reaction (PCR)

**Diseases:** Melanoma.

**Clinical Uses:** It is designed to help select patients for treatment with vemurafenib, an oral medicine designed to treat patients whose melanoma tumors that harbor a mutated form of the BRAF gene.

**Sources:** [www.molecular.roche.com](http://www.molecular.roche.com)

**Marker (Medline Search):** BRAF gene mutation detection

**Organ (Medline Search):** Melanoma

**Medline Searches:** (BRAF[All Fields] AND v600[All Fields] AND ("mutation"[MeSH Terms] OR "mutation"[All Fields]) AND ("melanoma"[MeSH Terms] OR "melanoma"[All Fields])) AND ("PLX4032"[Supplementary Concept] OR "PLX4032"[All Fields] OR "vemurafenib"[All Fields])

**Medline hits=**32

**FDA approved:** No

## **Gene Test Information: BRAF gene mutation detection, Multiple**

### **Test Name: BRAF gene mutation detection**

**Description:** Somatic mutations in the BRAF oncogene are frequently found in human cancers. These mutations are common in melanomas, colorectal cancer, lung cancer, ovarian cancer, and thyroid gland cancer.

**Purpose:** Therapeutic management

**Availability:** Labcorp and other laboratories

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue.

**Methodology:** Amplification refractory mutation system (ARMS) and real-time polymerase chain reaction (PCR) using Scorpions technology.

**Diseases:** Multiple cancers (melanomas, colorectal cancer, lung cancer, ovarian cancer and thyroid gland cancer).

**Clinical Uses:** More than 90% of mutations are the V600E (1799T>A) mutation. Recent studies have shown that metastatic colorectal cancer patients with this BRAF mutation do not have a strong response to anti-EGFR therapies such as cetuximab and panitumumab. This assay detects any amino acid change that has an A nucleotide at position 1799 in exon 15 of the BRAF gene regardless of any other common changes (could detect V600E/V600K/V600D), allowing identification of patients who are likely to benefit from such treatment.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** BRAF gene mutation detection

**Organ (Medline Search):** Melanoma

**Medline Searches:** BRAF[All Fields] AND ("genes"[MeSH Terms] OR "genes"[All Fields] OR "gene"[All Fields]) AND ("mutation"[MeSH Terms] OR "mutation"[All Fields]) AND detection[All Fields] AND ("melanoma"[MeSH Terms] OR "melanoma"[All Fields])

**Medline hits=**19

**FDA approved:** No

## **Gene Test Information: NeoSITE Melanoma**

### **Test Name: NeoSITE Melanoma**

**Description:** NeoSITE Melanoma is a second-generation FISH assay that aids diagnostic discrimination between nevi and melanoma by informing of chromosomal gains or losses in four regions predictive of malignancy. This modified version improves classification of morphologically borderline lesions and detection of spitzoid melanoma, to control for tetraploidy, and to include 8q24 and 9p21 markers.

**Purpose:** Diagnostic of melanoma.

**Availability:** NeoGenomics

**Specimen:** Paraffin Block, Cut Slides

**Methodology:** Fluorescence in situ hybridization (FISH)

**Diseases:** Melanoma

**Clinical Uses:** To aid in the discrimination between nevi and melanoma.

**Sources:** [www.neogenomics.com/neosite-melanoma.htm](http://www.neogenomics.com/neosite-melanoma.htm)

**Marker (Medline Search):** RREB1 OR 6p25; MYC OR 8q24 OR p16(CDKN2A) OR 9p21 OR CCND1 OR 11Q13 OR Cen 9 OR Centromere

**Organ (Medline Search):** Melanoma

**Medline Searches:** (RREB1[All Fields] OR (6p25:[All Fields] AND MYC[All Fields]) OR 8q24[All Fields] OR p16[All Fields] AND ("genes, p16"[MeSH Terms] OR ("genes"[All Fields] AND "p16"[All Fields]) OR "p16 genes"[All Fields] OR "cdkn2a"[All Fields] OR "cyclin-dependent kinase inhibitor p16"[MeSH Terms] OR ("cyclin-dependent"[All Fields] AND "kinase"[All Fields] AND "inhibitor"[All Fields] AND "p16"[All Fields]) OR "cyclin-dependent kinase inhibitor p16"[All Fields]) OR 9p21[All Fields] OR ("cyclin d1"[MeSH Terms] OR ("cyclin"[All Fields] AND "d1"[All Fields]) OR "cyclin d1"[All Fields] OR "ccnd1"[All Fields]) OR 11Q13[All Fields] OR (Cen[All Fields] AND 9[All Fields]) OR ("centromere"[MeSH Terms] OR "centromere"[All Fields])) AND ("melanoma"[MeSH Terms] OR "melanoma"[All Fields]) AND "humans"[MeSH Terms]

**Medline hits=**1076

**FDA approved:** No

**Gene Test Information: MEN2 (RET) DNA sequencing test, pheochromocytoma**

**Test Name:** MEN2 rearranged during transfection (RET) DNA sequencing test

**Description:** MEN type 2 (MEN2) conditions represent at least four different syndromes that associate pheochromocytoma with medullary thyroid carcinoma, hyperparathyroidism, and a number of other manifestations. MEN2 rearranged during transfection (RET) DNA sequencing test detects mutations in the coding sequences of the RET genes for diagnosis.

**Purpose:** Diagnosis.

**Availability:** Athena diagnostics

**Specimen:** Whole blood

**Methodology:** Polymerase chain reaction (PCR) and DNA sequencing

**Diseases:** Pheochromocytoma

**Clinical Uses:** MEN2 rearranged during transfection (RET) DNA sequencing test detects mutations in the coding sequences of the RET genes and may aid clinical diagnoses of pheochromocytoma and associated clinical manifestations

**Sources:** [www.athenadiagnostics.com](http://www.athenadiagnostics.com)

**Marker:** MEN2

**Organ:** thyroid

**Medline Searches:** MEN2[All Fields] AND ("phaeochromocytoma"[All Fields] OR "pheochromocytoma"[MeSH Terms] OR "pheochromocytoma"[All Fields])

**Medline hits=**84

**FDA approved:** No

## **Gene Test Information: miRInform™ Pancreas**

### **Test Name: miRInform™ Pancreas**

**Description:** miRInform™ Pancreas is a molecular diagnostic test, performed on fine needle aspirate (FNA) biopsies of solid lesions, that aids in the pre-operative diagnosis and disease management of pancreatic ductal adenocarcinoma (PDAC) patients.

**Purpose:** Aids in the pre-operative diagnosis and disease management of pancreatic ductal adenocarcinoma (PDAC) patients

**Availability:** Asuragen, Inc

**Specimen:** Fine needle aspirate (FNA) biopsies of solid lesions

**Methodology:** Reverse transcription quantitative polymerase chain reaction (RT-qPCR) analysis

**Diseases:** Pancreatic Cancer

**Clinical Uses:** Helps physicians diagnose indeterminate, atypical and suspicious cytopathology result and plan for best treatment.

**Sources:** [www.asuragen.com](http://www.asuragen.com)

**Marker:** miRNA

**Organ:** Pancreas

**Medline Searches:** (("micrornas"[MeSH Terms] OR "micrornas"[All Fields] OR "mirna"[All Fields]) AND ("pancreatic neoplasms"[MeSH Terms] OR ("pancreatic"[All Fields] AND "neoplasms"[All Fields]) OR "pancreatic neoplasms"[All Fields] OR ("pancreatic"[All Fields] AND "cancer"[All Fields]) OR "pancreatic cancer"[All Fields])) AND "humans"[MeSH Terms]

**Medline hits=**237

**FDA approved:** No



## **Gene Test Information: PIK3CA Oncogene mutation detection, Liver**

### **Test Name: PIK3CA Oncogene mutation detection**

**Description:** Somatic mutations in the PIK3CA oncogene are frequently found in human cancers. They are common in liver cancer, breast cancer, colorectal cancer, and ovarian cancer. These mutations may indicate prognosis and drug response. This assay detects four PIK3CA mutations (H1047R, E542K, E545D, E545K) in exon 9 and 20, allowing determination of whether there is a correlation between PIK3CA mutation status and drug response to PIK3 inhibitors like EGFR-targeted MoAbs.

**Purpose:** Prognosis and therapeutic management

**Availability:** LabCorp

**Specimen:** Formalin-fixed, paraffin-embedded (FFPE) tissue

**Methodology:** Amplification refractory mutation system (ARMS) and realtime polymerase chain reaction (PCR) using Scorpions technology.

**Diseases:** Multiple cancers (liver cancer, breast cancer, colorectal cancer, and ovarian cancer)

**Clinical Uses:** PIK3CA mutation status may aid to assess drug response like EGFR-targeted MoAbs.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker:** PIK3CA Oncogene mutation detection

**Organ:** Liver cancer

**Medline Searches:** PIK3CA[All Fields] AND ("oncogenes"[MeSH Terms] OR "oncogenes"[All Fields] OR "oncogene"[All Fields]) AND ("mutation"[MeSH Terms] OR "mutation"[All Fields]) AND detection[All Fields] AND ("neoplasms"[MeSH Terms] OR "neoplasms"[All Fields] OR "cancer"[All Fields])

**Medline hits=**25

**FDA approved:** No

## **Gene Test Information: PDGFRA mutation analysis, Gastrointestinal stromal tumors**

### **Test Name: PDGFRA mutation analysis**

**Description:** Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumor of the gastrointestinal tract, located mostly in the stomach (60%) and small intestine (35%). Approximately 80% of GISTs have a mutation in c-KIT and 5% to 10% of GISTs have a mutation on PDGFRA. Most known mutations in the PDGFRA gene are associated with imatinib response with the exception of D842V mutation. In a subset of intestinal high risk GISTs lacking c-KIT/PDGFRA mutations, 7% have a mutation in BRAF. Kinase inhibitors targeting BRAF may be effective therapeutic options in this molecular GIST subset for therapeutic management.

**Purpose:** Predictive and therapeutic management

**Availability:** LabCorp

**Specimen:** Formalin-fixed, paraffin-embedded tissue

**Methodology:** Polymerase chain reaction (PCR) and DNA sequencing

**Diseases:** Gastrointestinal stromal tumors

**Clinical Uses:** In a subset of intestinal high risk GISTs lacking c-KIT/PDGFRA mutations, 7% have a mutation in BRAF. Kinase inhibitors targeting BRAF may be effective therapeutic options in this molecular GIST subset for therapeutic management.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** PDGFRA mutation analysis

**Organ (Medline Search):** Gastrointestinal stromal tumors

**Medline Searches:** ("receptor, platelet-derived growth factor alpha"[MeSH Terms] OR ("receptor"[All Fields] AND "platelet-derived"[All Fields] AND "growth"[All Fields] AND "factor"[All Fields] AND "alpha"[All Fields]) OR "platelet-derived growth factor alpha receptor"[All Fields] OR "pdgfra"[All Fields]) AND ("mutation"[MeSH Terms] OR "mutation"[All Fields]) AND ("analysis"[Subheading] OR "analysis"[All Fields]) AND ("gastrointestinal stromal tumors"[MeSH Terms] OR ("gastrointestinal"[All Fields] AND "stromal"[All Fields] AND "tumors"[All Fields]) OR "gastrointestinal stromal tumors"[All Fields])

**Medline hits=**212

**FDA approved:** No

**Gene Test Information: ResponseDX:Melanoma®**

**Test Name: ResponseDX:Melanoma®**

**Description:** ResponseDX:Melanoma® panel utilizes testing of multiple genes including BRAF mutation, and NRAS mutation.

**Purpose:** Prognosis, Therapeutic Management

**Availability:** Responsegenetics.com

**Specimen:** Formalin-fixed, paraffin embedded (FFPE) biopsy specimen

**Methodology:** Fluorescence in situ hybridization (FISH) and polymerase chain reaction (PCR)

**Diseases:** Melanoma

**Clinical Uses:** Patients with melanoma who are being considered for treatment with the tyrosine kinase inhibitor (TKI) and EGFR antagonists cetuximab and panitumumab.

**Sources:** [www.responsegenetics.com](http://www.responsegenetics.com)

**Marker (Medline Search):** BRAF OR NRAS

**Organ (Medline Search):** Melanoma

**Medline Searches:** (BRAF[All Fields] OR NRAS[All Fields]) AND ("melanoma"[MeSH Terms] OR "melanoma"[All Fields])

**Medline hits =** 1226

**FDA approved:** No

**Gene Test Information: ResponseDx: Gastric®**

**Test Name: ResponseDX: Gastric®**

**Description:** ResponseDX: Gastric® panel utilizes testing of multiple genes including HER2 gene amplification, ERCC1 expression, and Thymidilate Synthetase expression.

**Purpose:** Prognostic, therapeutic management

**Availability:** Response Genetics

**Specimen:** Formalin-fixed, paraffin embedded (FFPE) biopsy specimen

**Methodology:** PCR-based tests

**Diseases:** Advanced gastric cancers

**Clinical Uses:** Amplification of the HER2 gene is associated with increased disease recurrence and a worse prognosis. ERCC1 expression predicts the best therapeutic combination of agents including platinum and select patients who might benefit from platinum-based therapies. Thymidylate synthetase (TS) expression predicts the best therapeutic combination of agents including pemetrexed or 5-FU and select patients who might benefit from pemetrexed-based therapies.

**Sources:** [www.responsegenetics.com](http://www.responsegenetics.com)

**Marker (Medline Search):** (HER2 OR ERCC1 OR Thymidylate synthetase)

**Organ (Medline Search):** Gastric cancer

**Medline Searches:** (HER2[All Fields] OR ERCC1[All Fields] OR ("thymidylate synthase"[MeSH Terms] OR ("thymidylate"[All Fields] AND "synthase"[All Fields]) OR "thymidylate synthase"[All Fields] OR ("thymidylate"[All Fields] AND "synthetase"[All Fields]) OR "thymidylate synthetase"[All Fields])) AND ("stomach neoplasms"[MeSH Terms] OR ("stomach"[All Fields] AND "neoplasms"[All Fields]) OR "stomach neoplasms"[All Fields] OR ("gastric"[All Fields] AND "cancer"[All Fields]) OR "gastric cancer"[All Fields])

**Medline hits =** 677

**FDA approved:** No

## **Gene Test Information: RET gene sequencing, thyroid carcinoma**

Test Name: RET gene sequencing, thyroid carcinoma

**Description:** this test can be used to identify genetic variations in the RET gene that are causative for MEN2. Germline mutations in the RET gene on chromosome 10 are causative for multiple endocrine neoplasia, type 2 (MEN2), a monogenic, autosomal-dominant hereditary cancer syndrome. The vast majority (>95%) of MEN2 cases have RET gene mutations in exons 10, 11, 13, 14, 15, or 16. MEN2 is characterized by the development of medullary thyroid carcinoma (MTC) and sometimes includes pheochromocytoma (PHEO) and hyperparathyroidism (HPT).

**Purpose:** Diagnosis

**Availability:** Labcorp

**Specimen:** whole blood

**Methodology:** Polymerase chain reaction (PCR) of targeted RET gene exons, DNA sequencing of those PCR products.

**Diseases:** Thyroid carcinoma, pheochromocytoma, and hyperparathyroidism

**Clinical Uses:** This test is performed on an affected individual (proband) in the suspected or defined MEN2 family. If a RET gene mutation is identified in the proband, testing for the specific family mutation may be offered to appropriate unaffected, at-risk relatives.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** RET gene mutation

**Organ (Medline Search):** thyroid cancer OR parathyroid cancer OR pheochromocytoma

**Medline Searches:** "ret"[All Fields] AND ("genes"[MeSH Terms] OR "genes"[All Fields] OR "gene"[All Fields]) AND ("mutation"[MeSH Terms] OR "mutation"[All Fields]) AND (("thyroid neoplasms"[MeSH Terms] OR ("thyroid"[All Fields] AND "neoplasms"[All Fields]) OR "thyroid neoplasms"[All Fields] OR ("thyroid"[All Fields] AND "cancer"[All Fields]) OR "thyroid cancer"[All Fields]) OR ("parathyroid neoplasms"[MeSH Terms] OR ("parathyroid"[All Fields] AND "neoplasms"[All Fields]) OR "parathyroid neoplasms"[All Fields] OR ("parathyroid"[All Fields] AND "cancer"[All Fields]) OR "parathyroid cancer"[All Fields]) OR ("phaeochromocytoma"[All Fields] OR "pheochromocytoma"[MeSH Terms] OR "pheochromocytoma"[All Fields]))

**Medline hits=**135

**FDA approved:** No

## **Gene Test Information: SDHB DNA sequencing test, pheochromocytoma**

### **Test Name: SDHB DNA sequencing test**

**Description:** MEN type 2 (MEN2) conditions represent at least four different syndromes that associate pheochromocytoma with medullary thyroid carcinoma, hyperparathyroidism, and a number of other manifestations. It detects mutations in the coding sequences of the succinate dehydrogenase complex, subunit B (SDHB) genes. Recent investigations have found mutations in succinate dehydrogenase subunit B (SDHB), the gene coding for subunit B of the respiratory chain complex II. Mutations in the SDHB gene, with additional loss of the wild-type allele, result in loss of function of respiratory complex II and appear to correlate with extra-adrenal location of pheochromocytomas. Also, a link has been established between malignant behaviour and inactivating mutations of SDHB.

**Purpose:** Prognostic

**Availability:** Athena diagnostics

**Specimen:** Whole blood

**Methodology:** Polymerase chain reaction (PCR) and DNA sequencing

**Diseases:** Pheochromocytoma

**Clinical Uses:** SDHB DNA sequencing test detects mutations in the coding sequences of SDHB genes and may aid in prognosticating malignant behaviour of pheochromocytoma.

**Sources:** [www.athenadiagnostics.com](http://www.athenadiagnostics.com)

**Marker (Medline Search):** SDHB gene

**Organ (Medline Search):** Pheochromocytoma

**Medline Searches:** SDHB[All Fields] AND ("phaeochromocytoma"[All Fields] OR "pheochromocytoma"[MeSH Terms] OR "pheochromocytoma"[All Fields])

**Medline hits=**234

**FDA approved:** No

**Gene Test Information: 1P, 19Q FISH, Oligoglioma**

**Test Name:** 1P, 19Q FISH

**Description:** The oligodendroglioma (OG) type of glial cell tumors accounts for 2-5% of primary brain neoplasms and 4-15% of gliomas diagnosed worldwide. Allelic losses on 1p, or on 1p and 19q, correlate with chemotherapy response and good prognosis in OG patients

**Purpose:** Diagnostic and Prognostic

**Availability:** Labcorp

**Specimen:** Fixed-cell pellet from a cytogenetic analysis

**Methodology:** Fluorescence in situ hybridization (FISH)

**Diseases:** Oligoglioma

**Clinical Uses:** Confirmation/identification of brain cancer-related alterations and response to chemotherapy

**Sources:** [www.labcorp.com](http://www.labcorp.com)

Marker: 1p 19q AND fluorescence in situ hybridization

Organ: Oligodendroglioma

**Medline Searches:** ("oligodendroglioma"[MeSH Terms] OR "oligodendroglioma"[All Fields]) AND 1p[All Fields] AND 19q[All Fields] AND ("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields]))

**Medline hits=**85

**FDA approved:** No

**MULTIPLE**



**Gene Test Information:** BROCA - Cancer Risk Panel

**Test Name:** BROCA - Cancer Risk Panel

**Description:** BROCA is useful for the evaluation of patients with a suspected hereditary cancer predisposition, with a focus on syndromes that include breast or ovarian cancer as one of the cancer types. Depending on the causative gene involved, these cancers may co-occur with other cancer types (such as colorectal, endometrial, pancreatic, endocrine, or melanoma). If mutations in BRCA1 or BRCA2 are suspected, these should be evaluated with a separate test.

**Purpose:** Prediction of the risk of breast and/ or ovarian cancer, as well as other cancers.

**Availability:** University of Washington Laboratory Medicine- Genetics Lab

**Specimen:** Blood

**Methodology:** RNA, Gene Sequencing

**Diseases:** Multiple Cancers, including breast and ovarian cancer

**Clinical Uses:** To determine if patient is at increase risk of developing cancer (specifically breast or ovarian cancer) and to develop a medical management plan to reduce the risk of cancer.

**Sources:** [www.web.labmed.washington.edu](http://www.web.labmed.washington.edu)

**Marker:** APC, ATM, ATR, BAP1, BARD1, BMPR1A, BRIP1, CDH1, CDK4, CDKN2A,CHEK1, CHEK2, FAM175A (Abraxas),GALNT12, GEN1, GREM1, HOXB13, MLH1, MRE11A, MSH2 (+E PCAM), MSH6, MUTYH, NBN, PALB2, PMS2, PRSS1, PTEN, RAD50, RAD51, RAD51C, RAD 51D, RET, SMAD4, STK11, TP53,TP53BP1, VHL, and XRCC2

**Organ:** Multiple Organs, including Breast, Ovarian

**Medline Searches:** (APC[All Fields] OR ("atm"[All Fields]) OR ATR[All Fields] OR BAP1[All Fields] OR BARD1[All Fields] OR ("bone morphogenetic protein receptors, type i"[MeSH Terms] OR "type i bone morphogenetic protein receptors"[All Fields] OR "bmp1a"[All Fields]) OR BRIP1[All Fields] OR CDH1[All Fields] OR CDK4[All Fields] OR ("genes, p16"[MeSH Terms] OR ("genes"[All Fields] AND "p16"[All Fields]) OR "p16 genes"[All Fields] OR "cdkn2a"[All Fields] OR "cyclin-dependent kinase inhibitor p16"[MeSH Terms] OR ("cyclin-dependent"[All Fields] AND "kinase"[All Fields] AND "inhibitor"[All Fields] AND "p16"[All Fields]) OR "cyclin-dependent kinase inhibitor p16"[All Fields]) OR CHEK2[All Fields] OR FAM175A[All Fields] OR GALNT12[All Fields] OR GEN1[All Fields] OR GREM1[All Fields] OR HOXB13[All Fields] OR MLH1[All Fields] OR MRE11A[All Fields] OR MSH2[All Fields] OR MSH6[All Fields] OR muty[All Fields] OR NBN[All Fields] OR PALB2[All Fields] OR PMS2[All Fields] OR PRSS1[All Fields] OR pten[All Fields] OR RAD50[All Fields] OR RAD51[All Fields] OR RAD51C[All Fields] OR

RAD51D[All Fields] OR "ret"[All Fields]) OR SMAD4[All Fields] OR STK11[All Fields] OR TP53[All Fields] OR VHL[All Fields] OR XRCC2[All Fields]) AND ("neoplasms"[MeSH Terms] OR "neoplasms"[All Fields] OR "cancer"[All Fields]) AND ((Clinical Trial[ptyp] OR Comparative Study[ptyp]) AND "humans"[MeSH Terms])

**Medline hits=** 3041

**FDA approved:** No

**Gene Test Information:** CancerNext™

**Test Name:** CancerNext™

**Description:** CancerNext™ utilizes next generation sequencing to offer a comprehensive testing panel for hereditary colon cancer and targets detection of mutations in 22 genes (APC, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11, and TP53). Gross deletion/duplication analysis is performed for all 22 genes.

**Purpose:** Predictive

**Availability:** Ambry genetics

**Specimen:** Blood, DNA, or Saliva

**Methodology:** gene sequencing, deletion/duplication analysis

**Diseases:** Breast, Colon, Uterine, Ovarian, Other

**Clinical Uses:** CancerNext™ is a next-generation cancer panel that simultaneously analyzes selected genes associated with a wide range of cancers. While mutations in each gene on this panel may be individually rare, they may collectively account for a significant amount of hereditary cancer susceptibility.

**Sources:** [www.ambrygen.com](http://www.ambrygen.com)

**Marker (Medline Search):** (APC, ATM, BARD1, BRIP1, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, SMAD4, STK11, and TP53)

**Organ (Medline Search):** breast OR ovarian cancer OR uterine OR colon

**Medline Searches:** (APC[All Fields] OR "atm"[All Fields]) OR BARD1[All Fields] OR BRIP1[All Fields] OR ("bone morphogenetic protein receptors, type i"[MeSH Terms] OR "type i bone morphogenetic protein receptors"[All Fields] OR "bmpr1a"[All Fields]) OR CDH1[All Fields] OR CHEK2[All Fields] OR ("tumor-associated antigen GA733"[Supplementary Concept] OR "tumor-associated antigen GA733"[All Fields] OR "epcam"[All Fields]) OR MLH1[All Fields] OR MRE11A[All Fields] OR MSH2[All Fields] OR MSH6[All Fields] OR MUTYH[All Fields] OR NBN[All Fields] OR PALB2[All Fields] OR PMS2[All Fields] OR PTEN[All Fields] OR RAD50[All Fields] OR RAD51C[All Fields] OR SMAD4[All Fields] OR STK11[All Fields] OR TP53[All Fields]) AND (("colon"[MeSH Terms] OR "colon"[All Fields]) OR ("breast"[MeSH Terms] OR "breast"[All Fields]) OR ("uterus"[MeSH Terms] OR "uterus"[All Fields] OR "uterine"[All Fields]) OR ("ovary"[MeSH Terms] OR "ovary"[All Fields])) AND "humans"[MeSH Terms]

**Medline hits=**6434

**FDA approved:** No

## **Gene Test Information: EGFR FISH, Multiple (Lung and brain cancer)**

### **Test Name: EGFR FISH**

**Description:** High EGFR copy number or ratio of EGFR: control >2.0 detected by FISH has been shown to correlate with response, progression-free survival, and overall survival after treatment with EGFR TKIs in patients with non-small-cell lung cancer (NSCLC)

**Purpose:** Prognostic

**Availability:** LabCorp, Bioscience Healthcare

**Specimen:** Fixed cell pellet from a cytogenetic analysis

**Methodology:** Fluorescence in situ hybridization (FISH)

**Diseases:** Multiple (lung cancer and brain cancer)

**Clinical Uses:** High EGFR copy number or ratio of EGFR: control >2.0 detected by FISH has been shown to correlate with response, progression-free survival, and overall [survival](#) after treatment with EGFR TKIs in patients with non-small-cell lung cancer (NSCLC). It is used for prognostic of lung and brain cancer.

**Sources:** [www.labcorp.com](http://www.labcorp.com)

**Marker (Medline Search):** EGFR fluorescence in situ hybridization

**Organ (Medline Search):** lung and brain cancer

**Medline Searches:** (("in situ hybridization, fluorescence"[MeSH Terms] OR ("situ"[All Fields] AND "hybridization"[All Fields] AND "fluorescence"[All Fields]) OR "fluorescence in situ hybridization"[All Fields] OR ("fluorescence"[All Fields] AND "situ"[All Fields] AND "hybridization"[All Fields])) AND EGFR[All Fields]) AND (("lung"[MeSH Terms] OR "lung"[All Fields]) OR ("brain"[MeSH Terms] OR "brain"[All Fields]))

**Medline hits=**334

**FDA approved:** No

## **Gene Test Information: OVANEXT**

### **Test Name: OVANEXT**

**Description:** OvaNext™ is a next generation (next-gen) sequencing panel that simultaneously analyses 19 genes that contribute to increased risk for breast, ovarian, and/or uterine cancers

**Purpose:** Prediction of risk of breast, ovarian, or uterine cancer.

**Availability:** Ambry Genetics

**Specimen:** Blood, DNA, Saliva

**Methodology:** Next Generation Gene Sequencing

**Diseases:** Breast, Ovarian, and/or Uterine Cancer

**Clinical Uses:** To determine if a woman has an increase chance of developing breast, ovarian, and/or uterus cancer.

**Sources:** <http://www.ambrygen.com/tests/ovanext>

**Marker:** ATM, BARD1, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, STK11, and TP53

**Organ:** Breast, Ovaries, Uterus

Medline Searches: (("breast"[MeSH Terms] OR "breast"[All Fields]) OR ("ovary"[MeSH Terms] OR "ovary"[All Fields]) OR ("uterus"[MeSH Terms] OR "uterus"[All Fields])) AND ("neoplasms"[MeSH Terms] OR "neoplasms"[All Fields]) AND (("Ann Thorac Med"[Journal] OR "atm"[All Fields]) OR BARD1[All Fields] OR BRIP1[All Fields] OR CDH1[All Fields] OR CHEK2[All Fields] OR ("tumor-associated antigen GA733"[Supplementary Concept] OR "tumor-associated antigen GA733"[All Fields] OR "epcam"[All Fields]) OR MLH1[All Fields] OR MRE11A[All Fields] OR MSH2[All Fields] OR MSH6[All Fields] OR MUTYH[All Fields] OR NBN[All Fields] OR PALB2[All Fields] OR PMS2[All Fields] OR PTEN[All Fields] OR RAD50[All Fields] OR RAD51C[All Fields] OR STK11[All Fields] OR TP53[All Fields]) AND "humans"[MeSH Terms]

medline hits= 3067

**FDA approved:** No

## **Gene Test Information: PANEXIA**

### **Test Name: PANEXIA**

**Description:** PANEXIA® detects mutations in genes that result in an increased risk of pancreatic cancer, offering insight about the risk of future hereditary cancers for patients and their families. PANEXIA, via a simple blood test, analyzes the PALB2 and BRCA2 genes, the two genes most commonly identified in families with hereditary pancreatic cancer.

The PANEXIA test results provide information for patients and their family members about the inherited risks of pancreatic cancer as well as breast, ovarian, and other cancers. This knowledge may allow at-risk family members the opportunity to lower their risks for some of these cancers through surveillance, preventative options, or lifestyle choices.

**Purpose:** Prediction of an increase risk of developing pancreatic and/or breast cancer.

**Availability:** Myriad Genetics

**Specimen:** Blood

**Methodology:** Gene sequencing

**Diseases:** Pancreatic or/and Breast Cancer

**Clinical Uses:** To determine if a person has an increase risk of developing pancreatic and/or breast cancer. The test determines the presence of the PALB2 and BRCA2 genes. The results of the test enable the development of a patient-specific medical management plan to reduce the risk of cancer

**Sources:** [www.myriad.com](http://www.myriad.com)

**Marker:** PALB2, BRCA2

**Organ:** Pancreas, Breast

**Medline Searches:** (PALB2[All Fields] OR ("genes, brca2"[MeSH Terms] OR ("genes"[All Fields] AND "brca2"[All Fields]) OR "brca2 genes"[All Fields] OR "brca2"[All Fields] OR "brca2 protein"[MeSH Terms] OR ("brca2"[All Fields] AND "protein"[All Fields]) OR "brca2 protein"[All Fields])) AND (("pancreatic neoplasms"[MeSH Terms] OR ("pancreatic"[All Fields] AND "neoplasms"[All Fields]) OR "pancreatic neoplasms"[All Fields] OR ("pancreatic"[All Fields] AND "cancer"[All Fields]) OR "pancreatic cancer"[All Fields]) OR ("breast neoplasms"[MeSH Terms] OR ("breast"[All Fields] AND "neoplasms"[All Fields]) OR "breast neoplasms"[All Fields] OR ("breast"[All Fields] AND "cancer"[All Fields]) OR "breast cancer"[All Fields])) AND "humans"[MeSH Terms]

**Medline hits=**4530

**FDA approved:** No

## **Gene Test Information: PTEN genetic analysis**

### **Test Name: PTEN**

**Description:** Somatic (noninherited) mutations in the PTEN gene are among the most common genetic changes found in human cancers. These mutations are acquired during a person's lifetime and are present only in tumor cells. PTEN gene mutations have been reported in many types of cancer, such as prostate cancer, endometrial cancer, glioblastomas and astrocytomas, and in melanoma. Mutations in the PTEN gene result in an altered protein that has lost its tumor suppressor function. The loss of this protein's function likely permits certain cells to divide uncontrollably, contributing to the growth of cancerous tumors. In some cases, the presence of PTEN gene mutations is associated with more advanced stages of tumor growth.

**Purpose:** Diagnostic, prognosis, and therapeutic management

**Availability:** Academic and Commercial laboratories

**Specimen:** Tumor tissue

**Methodology:** Deletion/duplication analysis; Next generation gene sequencing; Mutation analysis; FISH analysis

**Diseases:** Multiple cancers including prostate cancer, endometrial cancer, glioblastomas and astrocytomas, and melanoma

**Clinical Uses:** To detect and type mutations in the PTEN tumor suppressor gene for diagnosis of Cowden disease and Bannayan-Riley-Rucvalcaba syndrome and for prognosis and therapy selection in range of cancer types, particularly endometrial carcinoma, glioblastoma multiforme, melanoma, and prostatic carcinoma.

**Sources:** [www.ghr.nlm.nih.gov/gene/PTEN](http://www.ghr.nlm.nih.gov/gene/PTEN), [www.mdanderson.org](http://www.mdanderson.org)

**Marker (Medline Search):** PTEN

**Organ (Medline Search):** Cancer or Neoplasms

**Medline Searches:** pten[All Fields] AND ("neoplasms"[MeSH Terms] OR "neoplasms"[All Fields] OR "cancer"[All Fields])

**Medline hits=5396**

**FDA approved:** No

## **Gene Test Information: 5-Fluorouracil Sensitivity gene mutations**

### **Test Name: 5-FU sensitivity (DPYD, TYMS, and MTHFR)**

**Description:** 5-fluorouracil (5-FU) is a fluoropyrimidine drug and is the most frequently used chemotherapeutic drug in the treatment of colorectal cancer and other solid tumors. The dihydropyrimidine dehydrogenase (DPD) enzyme, encoded by the DPYD gene, is responsible for the degradation and inactivation of greater than 80 percent of 5-FU. TYMS gene mutations result in reduced expression of TS and may be associated with higher clinical responsiveness to 5-FU therapy and possibly an increased risk of toxicity. Methylene tetrahydrofolate reductase (MTHFR) is involved in the metabolism of folate and forms the reduced folate cofactor needed for TS inhibition.

**Purpose:** Therapeutic management

**Availability:** Academic and Commercial Laboratories

**Specimen:** Tumor tissue

**Methodology:** Multiplex PCR

**Diseases:** Colorectal cancer and other solid tumors

**Clinical Uses:** Genetic variation may contribute to risk of toxicity and/or altered therapeutic benefit

**Sources:** [www.aruplab.com](http://www.aruplab.com), [www.bcm.edu](http://www.bcm.edu)

**Marker (Medline Search):** Fluorouracil

**Organ (Medline Search):** MTHFR or TYMS or DPYD

**Medline Searches:** ("fluorouracil"[MeSH Terms] OR "fluorouracil"[All Fields] OR "5 fluorouracil"[All Fields]) AND (("methylene tetrahydrofolate reductase (nadph2)"[MeSH Terms] OR ("methylene tetrahydrofolate"[All Fields] AND "reductase"[All Fields] AND "(nadph2)"[All Fields]) OR "methylene tetrahydrofolate reductase (nadph2)"[All Fields] OR "mthfr"[All Fields]) OR TYMS[All Fields] OR DPYD[All Fields])

**Medline hits=269**

**FDA approved:** No (but labeled for 5-FU)



## **Appendix B. Genetic Tests For Cancer From Prior Horizon Scan Reports**

**Table B-1. Genetic tests for cancer found between January, 2006 and February 2011**

Test Name	Germline (Y)	Purpose				
		Prognostic / Predictive	Diagnostic	Monitoring	Recurrence	Therapeutic management
<i>Breast</i>						
Breast Profile						X
deCODE BreastCancer™		X	X			
GeneSearch™ BLN Assay		X	X			
Her2 Neu Overexpression						X
Her2 Pro™			X			X
MammaPrint®		X	X			
SPOT-Light ®HER2 CISH Kit						X
Tamoxitest™						X
TOP2A FISH pharmDx™ Kit		X		X		
<i>Colorectal</i>						
BRAF mutation	Y	X	X			X
ColonSentry™						
Colopath®/ColorectAlert™			X			
Cytokeratin 20(CK 20)			X			
KRAS Mutation Analysis		X				
Oncotype DX® colon cancer assay					X	
Septin-9 DNA methylation biomarker			X			
UGT1A1 Molecular Assay™						X
<i>Genitourinary</i>						
ImmunoCyt™/uCyt+™				X		
NMP22®BladderChek®			X			
<i>Hematologic</i>						
G6PD						X
Heme Profile		X				
JAK2			X			X
KIT Asp816Val Mutation Analysis			X			X
<i>Lung</i>						

Test Name	Germline (Y)	Purpose					
		Prognostic / Predictive	Diagnostic	Monitoring	Recurrence	Therapeutic management	
CellCorrect KvA-40 Lab® Kit			X				
EGFR Mutation Analysis	Y					X	
ELSA-CYFRA 21-1		X	X				
ERCC1	Y					X	
KRAS Mutation Analysis		X				X	
MESOMARK®			X			X	
<i>Ovarian</i>							
OVA1™		X					
OvaCheck™			X				
OvaSure™			X				
<i>Prostate</i>							
Bayer Immuno 1™ Complexed PSA			X	X			
deCODE Prostate Cancer			X				
Hybritech Tandem-R free PSA test			X				
ProgenSA® PCA3 Assay		X	X			X	
Prostate-63			X				
uPM3™ test; PCA3Plus™ test		X	X				
<i>Other**</i>							
DakoCytomation's c-Kit (9.7) pharmDx™			X			X	
LBA®AFP-L3							
MGMT methylation testing						X	
<i>Multiple***</i>							
CellSearch®		X		X	X		
CupPrint			X				
DPD deficiency						X	
EGFR™ assay						X	
miRview™			X				
Pathwork® Tissue of Origin test			X				
PI3K		X					

Test Name	Germline (Y)	Purpose					
		Prognostic / Predictive	Diagnostic	Monitoring	Recurrence	Therapeutic management	
TheraGuide™						X	
Tumor Profile		X					

\*\* Other includes brain, liver, and upper gastrointestinal, respectively

\*\*\* Tests used for multiple cancers including breast, colorectal, lung, ovarian, prostate

**Table B-2. Genetic tests for cancer found until December, 2005**

NAME	Germline (y)	Breast	Prostate	Lung	Colorectal	Pancreas	Ovarian	Esophagus	Liver	Lymphoma	Leukemia
Acid phosphatase, total and prostatic			x								
Adrenocorticotrophic hormone				x		x					
Alpha fetoprotein				x	x	x			x		
AML1/ETO translocation											x
B-cell gene rearrangement										x	
BCL-1/JH gene rearrangement										x	
BCL-2 translocation										x	
BCR/ABL gene rearrangement											x
Beta human chorionic gonadotropin				x		x	x		x		
Beta-2 microglobulin										x	
Bladder tumor antigen											
BRCA Analysis	Y	x				x	x				
Calcitonin											
Cancer antigen 125		x		x	x	x	x				
Cancer antigen 15-3		x									
Cancer antigen 19-9					x	x			x		
Cancer antigen 27.29		x		x		x	x		x		
Carcinoembryonic antigen		x		x	x	x	x				
Cathepsin D		x									
CBFB/MYH11 fusion protein											x
CD 117, c-kit											
CD 20										x	
CD 25										x	
CD 33											x
CD 52											x
Chromosome 18q assay					x						
Colaris					x						
Colaris AP					x						
Cyclin-D1	Y									x	
E-cadherin	Y		x								
Epidermal growth factor receptor		x	x	x	x						
Estrogen/progesterone receptor		x									
Fecal globin					x						
FLT 3 mutation											x
HER-2/neu		x					x				
5-HIAA											
Human papillomavirus hybrid capture											
IgVH mutation analysis											x
Immunocyt											

NAME	Germine (y)	Breast	Prostate	Lung	Colorectal	Pancreas	Ovarian	Esophagus	Liver	Lymphoma	Leukemia
Kappa/lambda light chain										x	
LAP						x					
Lipid associated sialic acid		x		x	x	x	x		x	x	x
Melaris						x					
MIB-1 antibody		x								x	
Micrometastasis detection		x									
Microsatellite instability					x						
MLH1, MSH2, MSH6 mutations	Y				x						
Neuron specific enolase				x		x					
Nuclear matrix proteins											
Oncotype Dx		x									
p53 tumor suppressor gene	Y	x	x		x						
PML/RARA translocation											x
PreGen-26					x						
PreGen-Plus					x						
Prostate-specific antigen			x								
T-cell receptor gene rearrangment										x	
TEL/AML1 gene fusion											x
Thyroglobulin											
Tumor antigen 90 immune complex											
Urokinase plasminogen activator		x									
Urovysion											
ZAP-70											x

**Table B-3. Tests that matured to clinical use since 2006**

Name	Breast	Prostate	Lung	Colorectal	Pancreas	Ovarian	Esophagus	Liver	Lymphoma	Leukemia	Other
AFP-L3								x			
APC		x									
BladderChek											x
CeMinex CellCorrect Lab			x								
c-Kit pharmDx											x
Colorectalert				x							
cPSA		x									
CupPrint											x
Cyfra 21-1			x								
DNA methylation (oncomethylome)		x	x			x					x
DNA methylation (second code)		x	x	x							
EGFR			x								
Immunocyt/ uCyt											x
Iressa test (EGFR mutation analysis)			x								
K-ras			x	x							
MammaPrint	x										
MESOMARK											x
MGMT				x							
NMP22											x
Ovachek						x					
OncoVue	x										

Name	Breast	Prostate	Lung	Colorectal	Pancreas	Ovarian	Esophagus	Liver	Lymphoma	Leukemia	Other
Prostate 63		x									
TUO test											x
uPM3		x									