Keeping pace with evolving tests for Hereditary Breast and Ovarian Cancer and Lynch Syndrome

In January 2013, Myriad launched Integrated BRACAnalysis®, which incorporates BART™ testing for large gene rearrangements into the test for Hereditary Breast and Ovarian Cancer. In February, sequencing and large rearrangement testing for the MYH gene was added to COLARIS®, the diagnostic test for Lynch syndrome. This posting reviews the timeline of some key developments in testing for these two most common hereditary cancer syndromes. Over the past 15 years, technology has advanced, testing has broadened, and experience has led to changes in clinical practice. As your patients return for follow-up visits, determine whether they have been evaluated according to current standards. Changes in family history, updated professional society guidelines, a new insurance plan and improved testing technology are some of the reasons to update every patient’s history annually.

HEREDITARY BREAST AND OVARIAN CANCER SYNDROME:

1996: BRACAnalysis was launched by Myriad as a sequencing test of BRCA1 and BRCA2, the two genes known to cause Hereditary Breast and Ovarian Cancer.

2002: Since sequencing does not detect unusual rearrangements in genes (such as large deletions or duplications), the 5-site rearrangement panel was routinely added to BRACAnalysis to detect five recurring large rearrangements in BRCA1.

2006: Myriad began offering BART (BRACAnalysis Large Rearrangement Test) as a standalone, full gene rearrangement test for both BRCA1 and BRCA2. Since then, clinicians have ordered BART as a reflex test for selected cases, and BART has been performed automatically when a patient’s personal and family history indicated particularly high risk.

Jan 2013: Myriad began offering Integrated BRACAnalysis, which includes sequencing and comprehensive (full gene) rearrangement testing of the BRCA1 and BRCA2 genes, i.e. BART is included. This enhancement is based on the 2012 NCCN guidelines recommending large rearrangement testing for all patients undergoing BRCA testing.1

LYNCH SYNDROME:

2000: COLARIS was launched by Myriad as a sequencing test for the two most common genes associated with Lynch syndrome, MLH1 and MSH2.

2004: Large rearrangement analysis of these two genes was added.

2005: Sequencing of the MSH6 gene became available.

2008: Sequencing of the MSH6 gene was incorporated into all COLARIS tests.

2011: Myriad enhanced the COLARIS test to include MSH6 large rearrangement testing, as well as tests for mutations in the PMS2 and EPCAM genes. (In New York State, this enhancement went into effect in March 2012.)

Feb 2013: Sequencing and large rearrangement testing for the MYH gene was added to COLARIS.

The field of molecular genetics is evolving constantly. New techniques for analyzing large rearrangements in genes have been developed, and new genes associated with hereditary cancer have been discovered. As “next generation” molecular technologies begin to enable testing of larger “panels” of genes, research and clinical experience will be needed to establish the reliability of these newer technologies, as well as the associated cancer risks and appropriate medical management for mutation carriers of the lesser-known genes. Regular contact with your Myriad Account Executive will keep you up to date on test offerings, test enhancements and current professional society guidelines. Contact Myriad Medical Services at 1-800-469-7423, extension 3850 or helpmed@myriad.com, or your Regional Medical Specialist if you need assistance in determining whether your patient’s past genetic testing meets current standards.
EVOLUTION OF TESTING

HEREDITARY BREAST AND OVARIAN CANCER SYNDROME

- 1996: Sequencing of BRCA1 and BRCA2 genes
- 2000: 5-site Large Rearrangement Panel in BRCA1
- 2004: BART (full gene rearrangements in BRCA1 and BRCA2)
- 2005: Large rearrangements in MLH1 and MSH2
- 2006: MSH6 sequencing available
- 2008: MSH6 sequencing routinely included with COLAPIS
- May 2011: MSH6 large rearrangements, PMS2, EPCAM
- Jan 2013: NRH sequencing and large rearrangements
- Feb 2013: Integrated BRACAnalysis (sequencing and BART)

LYNCH SYNDROME

REFERENCES: