OVERLAP BETWEEN LYNCH SYNDROME AND HEREDITARY BREAST AND OVARIAN CANCER SYNDROME AMONG FAMILY HISTORIES IN PATIENTS TESTED FOR HEREDITARY CANCER SYNDROMES

Jennifer Saam, MS, CGC, PhD, Chris Arnell, MBA, Kelsey Moyes, MStat, Ingrid Marino, MS, CGC, Rick Wenstrup, MD
Myriad Genetic Laboratories, Inc., Salt Lake City, UT

BACKGROUND

Patients with a personal and family history of cancer may be candidates for hereditary cancer syndrome testing. Currently, health care providers are faced with the difficulty of predicting which syndrome is more likely based on the available cancer history information. This strategy is based on the premise that different cancer syndromes have distinct cancer profiles. Lynch syndrome (LS) and hereditary breast and ovarian cancer (HBOC) are two of the most common cancer syndromes and, while there are cancers more common in one than the other (colon (CRC), endometrial (EC) for LS and breast (BR) for HBOC), these syndromes also have overlapping cancers (ovarian (OC), pancreatic (PC)).

METHODS

To determine the overlap between syndromes in a large population of patients, we analyzed a commercial laboratory database of patients tested for LS and HBOC in a clinical setting from 2006 to 2013. Cancer history of patients tested for HBOC were analyzed using the 2012 NCCN criteria for Lynch syndrome and patients tested for LS were analyzed using the 2013 NCCN criteria for HBOC. A subset of 9000 patients tested for both syndromes either sequentially or in parallel was also analyzed. Patients tested for a specific family mutation, the common Ashkenazi Jewish mutations, or a single gene for LS were excluded.

RESULTS

• Of the patients tested for HBOC, 6.9% met 2012 NCCN clinical criteria for Lynch syndrome. The secondary bar graph shows how patients met the LS criteria. (Figure 1)
• Of the patients tested for LS, 30% met 2013 NCCN clinical criteria for HBOC. The secondary bar graph shows how patients met HBOC criteria. (Figure 2)
• Among the 9000 patients tested for both syndromes (either concurrently or in sequence), 3.3% were positive for BRCA1/2 mutations and 3.5% were positive for LS mutations (MLH1, MSH2, MSH6, PMS2, and EPCAM). (Figure 3)

CONCLUSIONS

• A personal and family history analysis of patients tested for HBOC and LS demonstrates that it is not unusual for patients to meet criteria for both syndromes.
• Patients tested for both syndromes show patients were equally likely to be positive for LS as HBOC.
• Failure to test patients for all appropriate genes may result in the underdiagnoses of patients with hereditary cancer syndromes. With the availability of panel testing, covering multiple genes involved in hereditary cancer, patients may receive a more accurate diagnosis and appropriate medical management.
• Future studies of patients receiving panel testing may indicate that cancer syndromes overlap more than was previously understood.

Presented at CGA-ICC - October 7, 2013