Hereditary Cancer Risk Assessment: Establishing a Comprehensive Safety Net in a Large Multi-specialty Group

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BACKGROUND:
In the United States, it is estimated that 1 in 400 individuals will harbor a deleterious mutation in BRCA1/2 genes associated with hereditary breast and ovarian cancer (HBOC). In addition, approximately 1 in 440 individuals will have a predisposition to early-age colon, uterine, gastric, and ovarian cancer associated with Lynch syndrome and polyposis (MLH1, MSH2, MSH6, PMS2, EPCAM, and APC genes). Benefit has been demonstrated for both oncology and unaffected patients through detection of mutations with impact on surgical, surveillance, and chemoprevention options. The purpose of this study was to highlight our practice-based model across specialties to integrate assessment and testing for hereditary cancer within a 200 physician multi-specialty group.

METHODS:
In June 2012, our breast surgery program implemented a simple process to identify, screen, and evaluate patients for hereditary cancer using a sustainable workflow with the following components:

• A hereditary cancer risk assessment (HCRA) questionnaire was created and evaluated based on NCCN guidelines for HBOC, Lynch, and polyposis syndromes
• HCRA forms were given out to all incoming surgery and mammography patients
• HCRA forms were reviewed by trained nurses; patients with history that met NCCN criteria were offered risk assessment and hereditary cancer testing when appropriate
• All patients were provided with pre-test risk assessment and informed consent
• Test results were reviewed by physician and a detailed patient-specific management plan was communicated to the patient and primary care physician
• Upon presenting results, consultation with a certified community-based genetic counselor was offered to patients and families
• In 2014, we expanded our testing to include panel testing and expanded our screening process to other departments.

RESULTS:
In the eighteen months prior to implementation, four patients received testing and one was positive for a deleterious mutation. Testing results in the 67 months post implementation (through December 2017) revealed 96 deleterious mutations out of 1,015 tested patients which represents 9.46% of tested patients (see figure 1). This is consistent with rates seen in other studies. Among these harmful mutations, 46.8% were positive for a BRCA mutation and 16.7% for a Lynch syndrome mutation. We also detected a patient who carries an APC mutation. The systematic nature of this process allows for a platform on quality ongoing improvement. Since implementation, we have developed more testing sites within the Internal Medicine and OBGYN groups.

CONCLUSIONS:
Comprehensive screening with a systematic process for evaluating hereditary cancers has allowed us to identify and manage a large number of patients with deleterious mutations. This is potentially life-saving for those as well as for close blood relatives. In the first 67 months of this process we were able to identify 96 deleterious mutations (9.46% positive rate) and provide personalized cancer risk management to all 1,015 patients. This approach could be easily implemented in other similar practice environments.