Breast surgeons have been ordering and counseling patients for BRCA1/2 testing for 20 years but genetic testing is rapidly expanding—with both demand in the genetic testing. ASBG Consensus guidelines recognize that breast surgeons are ideally positioned to provide access and counseling, but acknowledge that support by genetic counselors is beneficial in some circumstances.

This study was designed to determine if remote genetic counselor access impacts physician decision making for appropriate patient selection, genes tested, and clinical management.

Genetic counselors are an invaluable but limited resource with variable supply across the country. There are less than 1000 genetic counselors in the clinical setting focused on hereditary cancer risk with reported monthly caseloads ranging from 2-96 patients. The number of patients undergoing testing is rapidly expanding and currently hundreds of thousands of patients are tested annually. This study was designed to determine how a new model to support expanded genetic testing—real-time physician access to a remote genetic counselor—impacts physician decision making for appropriate patient selection, genes tested, and clinical management.

Methods

This multi-center prospective study involved 13 community-based breast cancer practices experienced with hereditary cancer testing, who did not have a genetic counselor as part of their practice. Participating practices on average ordered 10 tests per month. Physicians identified genetic testing candidates and discussed each case with a remote GC. Physicians could also review cases with GCs after test results were received and prior to conducting patient counseling. Pre and Post test surveys about each patient were completed by the testing physician. A standard pricing fee regardless of panel size or genes selected was employed for this study.

A total of 192 patients were evaluated, with a median age of 52. Reasons for genetic testing included family history of cancer (72%), recent personal diagnosis of breast cancer (58%) and impact on medical care for family members (42%). Risk assessment was performed in 98% of patients. 65% of patients met NCCN genetic testing guidelines. Risk tools included BRCAPRO and Hughes Risk. Pathogenic mutations were found in 14% of patients.

Physicians changed their test selection after conferring with the GC 21% of the time. Physicians sought post-test consultations 47% of the time. Post-test remote GC consultation resulted in a meaningful change in patient management 15% of the time.

Conclusions

- Community-based breast surgeons are assessing risk and using standardized tools to select appropriate patients for genetic testing.
- Remote genetic counseling support helps physicians choose the best gene panel test, aids in counseling challenging cases, and impacts clinical management of patients and unaffected family members.
- Breast surgeons see patients multiple times over their lifetime and can make follow up recommendations and referrals as new genetic risk information emerges.
- Remote GC support increases physician confidence in utilization of expanded panels.
- Remote genetic counseling provided to specialty breast surgeons is an efficient use of genetic resources and may provide a model for “as needed” or “on demand” genetic counseling as genetic testing volume and use of larger panels increase.

Case Study

Patient Background: 40-year-old female presents with DCIS. Family history significant for maternal aunt with breast cancer in her 50s and maternal uncle with prostate cancer in his 50s.

Testing and Counseling: Patient meets NCCN criteria for BRCA1/2 testing based on being diagnosed <45 with breast cancer. Ordered guidelines breast panel with reflex to larger panel if negative. The main reason for testing is surgical decision making.

Outcome: CHEK2 pathogenic result. While breast conserving surgery and increased surveillance is an option, patient chose bilateral mastectomy and saw a local genetic counselor. After reviewing the case a month later with a remote GC, the surgeon learned that NCCN management guidelines now incorporate colon cancer risk and early screening and referred patient to GI specialist.

Close relatives are at risk for a CHEK2 mutation and were referred for genetic testing to determine if they inherited this mutation and associated cancer risks.

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