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A Clinical Trial to Increase the Identification, Genetic Counseling Referral and Genetic Testing of Women at risk for Hereditary Breast and/or Ovarian Cancer

Conference Abstract

A clinical trial to increase the identification, genetic counseling referral and genetic testing of women at risk for hereditary breast and/or ovarian cancer

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Background: Approximately 1/300 individuals in the general population are at risk for hereditary breast and ovarian cancer due to an inherited mutation in the BRCA1/BRCA2 genes. *BRCA* mutations are associated with dramatically increased risks for breast cancer, especially at younger ages, in addition to ovarian cancer. Enhanced screening and risk reduction strategies can significantly reduce associated morbidity and mortality. The United States Preventive Services Task Force (USPSTF) recommends identifying women at-risk for *BRCA* mutations for receipt of genetic counseling and if appropriate, testing. The Breast Cancer Genetics Referral Screening Tool (B-RST) is a validated screen endorsed by USPSTF to facilitate this process. This implementation study seeks to evaluate the most effective means of follow-up for screen-positive women to maximize the number who are referred to, and receive, cancer genetic counseling (CGC) services.

Methods: B-RST (v3.0) was used in three Emory Healthcare breast-imaging centers. Screen-positive women were randomized into three methods of follow-up (patient initiated, physician notification, or staff phone call). Primary outcomes were to compare the number of screen positive individuals who were referred for, scheduled, and completed a CGC appointment among the three groups.

Results: Of 3,419 women approached, 63% participated and 579 (27%) screened positive. Appointments were scheduled by 7% of Group 1 participants, 17% of Group 2 individuals, and 11% of Group 3 ($p<0.001$). Scheduling challenges included physician non-response to notification and unsuccessful direct contact. Of those scheduled, 73% completed the CGC appointment.

Conclusions: Genomic medicine is receiving increased attention in the public health arena. Screening with B-RST 3.0 in mammography settings can improve identification of individuals at-risk for *BRCA* mutations and facilitate referral to CGC services. Despite B-RST's ability to easily and accurately identify individuals appropriate for CGC, additional strategies are needed to facilitate completion of CGC in routine clinical practice.

Key words: cancer genetic counseling, hereditary breast and ovarian cancer, BRCA

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