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Impact of genetic testing on risk-reducing behavior in women at risk for hereditary gynecologic cancer syndromes

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Background

Women with hereditary breast and ovarian cancer (HBOC) have an estimated 15-65% lifetime risk of ovarian cancer; similarly, women with Lynch syndrome have a 40-60% lifetime risk of endometrial cancer and a 10-12% lifetime risk of ovarian cancer. The aim of this study was to investigate the impact of genetic testing on risk-reducing behavior for gynecologic malignancies in women being tested for HBOC and Lynch syndrome.

Methods

190 women age ≥30 undergoing genetic testing for HBOC (N=102) or Lynch syndrome (N=88) completed questionnaires at baseline, 1 month, 3 months, and one year after testing. Women evaluated for HBOC were tested for germline BRCAI or BRCA2 gene mutations; those evaluated for Lynch syndrome were tested for germline DNA mismatch repair (MMR) gene mutations. Subjects were asked about personal cancer history, prior surgeries, cancer screening practices, and genetic test results. Gynecologic cancer screening was considered adequate for the HBOC cohort subjects if they had a transvaginal ultrasound (TYUS) in the past 12 months. Adequate screening for Lynch cohort subjects was defined as having had a TVUS and endometrial biopsy in the past 12 months.

Results

Of the 190 women age ≥30 undergoing genetic testing, 145 subjects (87 HBOC; 58 Lynch) had no prior history of gynecologic malignancy, hysterectomy, or salpingo-oophorectomy (BSO) and were thus at risk for gynecologic cancer. Subjects' mean age was 45.4 years and 52% (74/141) reported a personal history of any cancer. At baseline, 40% (34/84) of subjects being tested for HBOC and 46% (26/57) of those tested for Lynch syndrome reported prior gynecologic cancer screening.

One-year follow up questionnaires were available on 74 subjects (48 HBOC; 26 Lynch) none of whom had been diagnosed with a gynecologic cancer in the year following testing. 92% (11/12) of BRCA mutation-positive subjects had undergone either BSO (N=8) or screening with TVUS (N=3). None of the 6 BRCA mutation-negative subjects had prophylactic surgery or gynecologic cancer screening. Of those HBOC subjects with indeterminate or variant genetic test results, 27% (8/30) had either a BSO (N=3) or TVUS (N=5). 100% (5/5) of the MMR mutation-positive had undergone hysterectomy with BSO (N=2) or screening with TVUS and endometrial biopsy (N=3). None of the 3 MMR mutation-negative subjects had prophylactic surgery or gynecologic cancer screening. Of those subjects with indeterminate or variant genetic test results, 17% (3/18) had screening; none had prophylactic surgery.

Conclusions

In the first year after genetic testing, women who tested positive for HBOC or Lynch syndrome increased uptake of prophylactic surgery or screening to reduce their risk of gynecologic cancers. Women with true-negative results do not pursue these unnecessary interventions, whereas those with indeterminate or variant test results do not significantly change their risk-reducing behaviors.

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