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Editorial

Epithelial Ovarian Cancer in Women

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EDITORAL

Although genetic testing is recommended for women with epithelial ovarian cancer (EOC), little is known about patient preferences for various testing options. We measured relative preferences for attributes of testing in women with EOC referred for genetic counseling. Low owed price, high chance of detection injurious mutations and high chance of a EOC result area unit most well-liked by patients with EOC considering genetic testing.

Personalized medicine is transforming contemporary cancer care. Germline genetic testing is an expanding and increasingly visible means to modify personal cancer risk. However, the evidence base surrounding how patients with cancer value various features of genetic testing is under-developed. Nearly one-quarter of epithelial ovarian cancers (EOC) now have an identifiable hereditary cause. Although the majority of hereditary EOC is associated with mutations in either BRCA1 or BRCA2 (65–85%), genes in the homologous recombination and mismatch repair pathways have also been implicated in hereditary ovarian tumorigenesis. Hereditary EOCs are also associated with elevated risks of other malignancies, including breast, uterine, and colorectal cancers. As such, in March 2014 the Society of Gynecologic Oncology (SGO) issued guidelines stipulating that all women with a diagnosis of EOC should be offered genetic counseling, with consideration given to genetic testing.

The results of genetic testing may have a significant impact on the physical, emotional and financial well-being of those who are tested and their families. The identification of hereditary genetic defects in women with EOC may lead to directed gene testing in family members and enhanced screening recommendations or medical or surgical prophylaxis for associated cancers.

Patients' concerns about the potential financial difficulties associated with testing and downstream medical monitoring as well as concerns about insurance or employment discrimination may also impact their willingness to undergo genetic testing. Meanwhile, a battery of new genetic tests is flooding the market, with no clear guidance for patients or clinicians regarding the appropriate order of testing. Testing for single genes that are part of well-recognized syndromes, such as the BRCA1/2 genes, is most likely to provide a definitive result with actionable clinical care recommendations.

However, a number of currently marketed genetic panels include up to 35 genes. Results from these panels may identify deleterious mutations in additional genes known to increase the risk of certain malignancies. Variants of uncertain significance (VUS), whose clinical relevance is unknown, may also be identified during testing. As a result, up to 40% of women who are tested with the more comprehensive panels will be identified as carriers of VUS, for which no clear pathologic implications or clinical management strategies have been identified.

Given the introduction of numerous genetic tests with varying attributes, our study aimed to elicit the preferences of women with EOC referred for genetic counseling in order to tailor genetic counseling and test selection. We quantified tradeoffs among 5 attributes of genetic testing strategies that would be considered acceptable to patients. We were specifically interested in the influences of out-of pocket costs, the likelihood of identifying a deleterious mutation, and the likelihood of identifying a VUS. This is in line with a recent study demonstrating that more than three-fourths of ovarian cancer patients surveyed expressed a desire to obtain their genetic results, even if the information was not currently actionable.

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