

Genetic testing for breast cancer 1 (*BRCA1*) and breast cancer 2 (*BRCA2*) mutations in clinical oncology is becoming more widely employed around the world.^{1,2,3} Testing allows clinicians to determine if unaffected high-risk women carry a *BRCA* mutation, and if preventive care in the form of prophylactic procedures and/or increased surveillance is advisable. As well, identifying carrier status can aid physicians in tailoring the best treatment for affected women with breast or ovarian cancer. However, studies have shown that the uptake of preventive strategies, particularly risk-reducing surgeries, among unaffected women found to carry a *BRCA* mutation is influenced by a number of factors. These include cost of genetic testing, cancer-related distress, patient consultation, perceived benefits and risks of invasive surgery, and level of education. To alleviate these emotional and cognitive barriers, clinicians should ensure women carrying a *BRCA* mutation are well-informed about available treatments and potentially fatal outcomes associated with breast and ovarian cancer.

INTRODUCTION

BRCA1 and *BRCA2* are human caretaker genes encoding for proteins that function to repair damaged DNA. Mutations in either of these genes therefore threaten the stability of genetic material and increase cancer susceptibility.^{1,2,3} Specifically, hereditary breast cancer has been estimated to account for 5–10% of all breast cancers, and carriers of *BRCA1* or *BRCA2* genetic mutations have a 40–80% lifetime risk of developing breast cancer.^{1,2} The frequency of these mutations may vary significantly between populations depending on geographic origin and ethnically defined genetic combinations. For example, approximately 23% of unselected cases of breast cancer in the Bahamian population are attributable to a mutation in the *BRCA1* gene.^{1,2} In Canada, a country with a more ethnically diverse population, the population attributable risk is approximately 1%.

Genetic testing for mutations in the *BRCA1* and *BRCA2* cancer susceptibility genes in clinical practice is an accessible service in North America and Europe for high-risk individuals and patients diagnosed with breast cancer.² A study on Canadian women with a *BRCA* mutation reported an uptake of preventive bilateral mastectomy at 36% and preventive oophorectomy at 61%.¹³ By detecting *BRCA1* and *BRCA2* mutations prior to cancer diagnosis, patients can be offered preventive interventions, including increased surveillance for early

detection and prophylactic surgeries. As well, the standard of treatment for women with breast or ovarian cancer carrying a *BRCA* mutation may vary compared to that of non-carriers.

COST-UTILITY OF GENETIC TESTING

Genetic testing can be an expensive process.^{3,4,5} As a result, many countries and institutions ration this service based on eligibility criteria and risk-assessments. Genetic counselling without testing has been reported to cost approximately \$200, while counselling, testing, and disclosure of carrier status collectively can exceed \$2000.^{3,4,5} However, advances in genetic sequencing have gradually decreased the cost of testing. The factors normally

considered in determining if a patient is eligible for testing involve the probability of a woman carrying a *BRCA* mutation and the cost of the test.³ The probability of a positive test outcome is a function of age, gender, disease status, ethnicity, and family history of relevant conditions. Robinson et al. emphasized the utility of genetic testing and the associated uptake of prophylactic procedures in underserved and underrepresented populations.⁶ In this way, recognizing factors

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that influence the decision-making process and barriers to the uptake of prophylactic surgeries by individuals who are found to carry a *BRCA* mutation has been identified as a subject for further research.^{3,6}

The National Comprehensive Cancer Network has advised that *BRCA1* and *BRCA2* mutation carriers be offered prophylactic bilateral mastectomy, the surgical removal