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.12 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.7 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 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.5,6

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