Genetic Counseling for *BRCA1/2* Mutations: Women's Experiences, Preferences, and Psychosocial Outcomes of Counseling

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Abstract

AbstractOne in 8 women will be diagnosed with breast cancer in her lifetime, but only 5-10% of women who are diagnosed have a *BRCA1* and *BRCA2* (BReast CAncer) genetic mutation. These mutations naturally occur in biological family units, and women with these mutations live with an increased rate of breast and ovarian cancers. The standard of genetic counseling care in the U.S. is individualized counseling, in which one at-risk family member is tested at a time. A new and potentially more relevant genetic counseling approach, family-based genetic counseling tests all at-risk family members at one time, as one cohesive patient group. The current study explored lived experiences and preferences (i.e., individualized, family-based) of women who have tested positive for these mutations and psychosocial outcomes of genetic counseling. A sample of 60 *BRCA1/2*-positive women was recruited through multiple online support groups. Participants completed an online questionnaire outlining demographic characteristics, genetic counseling information, and HRQoL outcomes. A subset of this sample (n=34) were interviewed to gain experiential insight into their genetic counseling experience and preferences for genetic counseling. Six themes emerged: sources affecting perceived risk, preventive concerns and decisions, experiences in healthcare, emotional reactions to genetic counseling, future recommendations, and family support and communication. Three interesting subthemes were also identified, including 1) "pre-vivor", how women of this demographic describe themselves to others, 2) "testing intuition", the idea of knowing one's

genetic test results were positive before receiving them, and 3) the "hard truth" that prophylactic surgeries are the only true option, whereas biannual surveillance and chemoprevention just buy time. Preferentially, women would have chosen family-based genetic counseling instead of the standard individualized counseling if given the choice. Anxiety ($p<.01$) and stress ($p<.01$) were found to be significantly worse in the current sample compared to the general female population. Other HRQoL domains differed, with physical role limitations ($p<.05$), energy ($p<.05$), social functioning ($p<.05$), and general health perceptions ($p<.01$) being worse in the current sample than in the general population, but physical functioning ($p<.01$) and emotional wellbeing ($p<.05$) were significantly better than in the general population. These findings can inform future research and practice focused on improving women's experiences and psychosocial health in those being tested for *BRCA1/2* genetic mutations.

Keywords: *BRCA1*, *BRCA2*, hereditary mutations, breast cancer, ovarian cancer, family-based genetic counseling