

# Defining Expectations of Future Biopsy and Frequency of Benign or Malignant Results of BRCA1/2 Positive Patients Undergoing Active Surveillance

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## ABSTRACT

### INTRODUCTION AND BACKGROUND

- At our institution, the Preventive Care Program for Women's Cancers was started in 2003 for women at increased risk of developing breast cancer.
- When a woman is diagnosed with a genetic mutation known to be associated with breast cancer, she may elect to undergo active surveillance or prophylactic surgery.
- BRCA1 and BRCA2 are the most common genetic mutations and women with these mutations are at the highest risk for developing breast cancer.<sup>[1]</sup>
- The overall lifetime risk of breast cancer in women with BRCA1 and BRCA2 genetic mutations is 72% and 69%, respectively, compared to 12.5% in women with average risk factors.<sup>[2]</sup>
- Studies have shown that decision aids about surveillance options and association with breast cancer risk reduction, may help patients choose among different options.<sup>[3,4]</sup>
- In women who choose active surveillance, information regarding how frequently they can expect to undergo biopsy and the frequency of a benign or malignant result is useful in defining realistic future expectations in this high-risk group.

### OBJECTIVES

- To report on the asymptomatic patient with BRCA1 and BRCA2 deleterious mutations, who enrolled in our prevention program, frequency of biopsies, rate of benign or malignant results, and decision for prophylactic surgery.
- To provide patients with BRCA1/2 mutations with surveillance expectations when enrolled in our Preventive Care Program for Women's Cancer.

## METHODS

- Since the initiation of the Prevention Care Program for Women's Cancers, 2,641 patients have enrolled in this IRB approved database.
- A retrospective cross-sectional study was conducted using this population.
- From February 2003 through August 2018, women identified as increased risk for developing breast cancer were recruited for enrollment in this study.
- Demographics, family history of cancers, and genetic testing data were collected. In addition, events regarding genetic mutations, method of detection of suspicious lesions, number of biopsies, results of those biopsies, prophylactic surgery, and cancer diagnosis were recorded.

	BRCA1	BRCA2	p-value
	n=74	n=78	
Age†	35.7 (11.1)	41.6 (12.9)	0.01
Race			
Caucasian	67 (90.5)	70 (90.9)	0.99
African American	6 (8.1)	6 (7.8)	
Asian	1 (1.4)	1 (1.3)	
Prophylactic Surgery	32 (44.4)	20 (26.7)	0.02
Length of Follow-Up (days)*	1,065 (354-2,020)	1,309 (672-2,637)	0.36
Active Surveillance	40 (54.1)	54 (69.2)	0.05
Length of Follow-up (days)*	648 (353 – 1278)	570 (349-1381)	0.65
Biopsy	24 (32.4)	27 (34.6)	0.78
Malignant Result	0 (0.0)	3 (11.1)	0.11

† Mean and Standard Deviation Provided

\* Median and Interquartile Range Provided

## RESULTS

- 152 patients** with asymptomatic BRCA1 and BRCA2 deleterious mutations; mean age at enrollment was 38.7 (BRCA1 35.7 vs BRCA2 41.6, p=0.004).
- 52 (34.2%) underwent prophylactic surgery and 94 (61.8%) underwent active surveillance [BRCA1 40 (54.1%) and BRCA2 54 (69.2%), p=0.05].
- Prophylactic surgery:** 39 (75.0%) did not require biopsy, 12 (23.1%) had abnormal imaging with subsequent biopsy with a benign result, and 1 (1.9%) had a biopsy with a malignant result.
- Active surveillance:** 59 (62.3%) did not require biopsy, 34 (36.2%) had a biopsy with a benign result, and 1 (1.1%) had a biopsy with a malignant result.
  - 1 biopsy: 28 (29.8%)
  - 2 biopsies: 6 (6.4%)
  - 3 biopsies: 1 (1.1%)
- Method of detection of lesion requiring biopsy included 22 (47.8%) magnetic resonance imaging, 12 (26.1%) mammogram, 2 (4.4%) physical exam, and 1 (2.2%) ultrasound.

## CONCLUSIONS

- Women with BRCA mutations enroll in breast cancer prevention clinics to learn more about their breast cancer risk, methods of prevention, access to participation in surveillance and early detection of breast cancer.
- With BRCA mutations being the most common genetic mutation in breast cancer, this study highlights the effectiveness of screening this high-risk population. While this group of women undergo biopsies more frequently due to increased screening, the majority do not require a biopsy during their surveillance.
- For those who do require a biopsy, this result is typically benign. This additional information can be offered to women enrolling in prevention clinics to further allow them to make more informed decisions about pursuing surveillance in this high-risk group and establish realistic expectations of potential future need for tissue sampling.

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