

# Timing of *BRCA* Genetic Testing and Extent of Breast Cancer Surgery in Women with Deleterious Mutations

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## Background

Deleterious *BRCA* mutations confer increased cancer risks, including higher risks of new primary cancers, but surgical interventions can reduce these risks by at least 95%.<sup>1,2</sup>

Women with breast cancer who are found to be positive for *BRCA* mutations may opt for more extensive surgeries at the time of cancer treatment for the purpose of prophylaxis. Others may not have testing until well after their cancer diagnosis.

Our study explored which surgical interventions women with deleterious *BRCA* mutations choose, and whether there were differences in surgery choice depending on knowledge of *BRCA* results at the time of cancer treatment.

## Methods

A phone survey including *BRCA* positive women was created by the Michigan Department of Community Health and conducted by eight Michigan centers with board-certified/eligible genetics providers who had provided genetic counseling to the respondents. Seven of the eight facilities attempted inclusion of all of patients meeting criteria, while one facility contacted a random subset of their eligible patients.

Women in this analysis met the following criteria:

- Counseled at one of eight facilities with genetics providers from Oct. 1, 2007 – Sept. 30, 2009
- Had *BRCA* testing, either before or after the counseling visit
- Were positive for a deleterious *BRCA* mutation

### Survey Surgery Q's

#### Prophylactic Surgery

*Prompt: A prophylactic measure is something done to prevent cancer before any symptoms occur. These surgeries may occur at the same time as cancer treatment. For example, the removal of an unaffected breast during surgery to remove cancer in the opposite breast is prophylactic.*

1) Have you ever had a prophylactic mastectomy to prevent breast cancer?

- Was this after you received your *BRCA* test results?

2) Are you considering a prophylactic surgery in the future?

- What type of prophylactic surgery are you thinking of having?

#### Cancer Treatment Surgery

*Prompt: This section is about cancer diagnoses and treatment.*

1) If you have ever had surgery to treat breast cancer, what is the most extensive type of surgery you have had?

- Did you receive your *BRCA* results before this cancer surgery?

## Results

Out of 96 respondents who tested positive for a *BRCA* mutation, 48 (50.0%) had been diagnosed with breast cancer, and three of these individuals were diagnosed with breast cancer after having genetic counseling and testing (Table 1).

**Table 1. Cancer History of *BRCA* positive respondents**

Cancer Event	<i>BRCA</i> Positive Patients N = 96 # (%)
New Cancer*	
New breast cancer	3 (3.1)
New ovarian cancer	4 (4.2)
No new cancer	89 (92.9)
Any cancer history**	
Breast	44 (45.8)
Ovarian	10 (10.4)
Both breast and ovarian	4 (4.2)
No cancer history	38 (39.6)

\*Diagnosed since *BRCA* testing

\*\*Diagnosed at any time, prior to or after counseling and testing

Forty-five *BRCA* positive respondents with a history of breast cancer reported the most extensive breast cancer treatment surgery undergone, including bilateral mastectomy (17, 37.8%), unilateral mastectomy (17, 37.8%) and lumpectomy (11, 24.4%).

There was a significant difference in the surgeries undergone by *BRCA* positive women who knew and who did not know their *BRCA* status at the time of surgery (Fisher's exact test,  $p < 0.01$ ), shown in Figure 1.

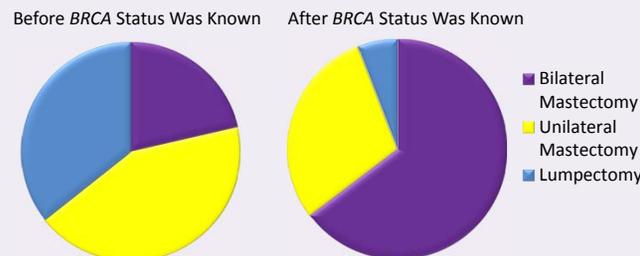
**Fig 1. Breast cancer surgery and knowledge of mutation status among *BRCA* positive women**

Of women who had NOT YET known their *BRCA* positive status:

- 21.4% (6) had bilateral mastectomies
- 42.9% (12) had unilateral mastectomies
- 35.7% (10) had lumpectomies

Of women who KNEW their *BRCA* positive status:

- 64.7% (11) had bilateral mastectomies
- 29.4% (5) had unilateral mastectomies
- 5.9% (1) had lumpectomies



## Results

The survey also addressed prophylactic surgeries, including both the removal of an unaffected breast at the time of cancer treatment, and independent of any cancer surgery. Of all respondents with a *BRCA* positive result, 41.7% had a prophylactic mastectomy after learning their mutation status (Table 2).

**Table 2. Prophylactic Surgeries among *BRCA* positive women**

<i>BRCA</i> Result	Prophylactic Mastectomies (row %)		
	Knew <i>BRCA</i> Status at Surgery:	<i>BRCA</i> Status Unknown at surgery:	No Mastectomy:
Positive	40 (41.7)	11 (11.5)	45 (46.9)

While 45 (46.9%) of positive women had not yet had a prophylactic mastectomy, only 13 (13.5%) of these women had not had an oophorectomy, which also provides breast cancer risk-reduction. Of these 13 women with no surgical history, 11 (84.6%) planned to have a future risk-reducing surgery.

## Conclusion

- *BRCA* positive women who knew their mutation status at the time of surgery tended to choose more extensive breast cancer surgeries (majority bilateral mastectomy)
- Women with cancer who did not learn their *BRCA* positive status until after cancer treatment chose lumpectomies nearly as often as unilateral mastectomies; both were more common than bilateral mastectomies
- Prophylactic surgery uptake appears to be high among *BRCA* positive respondents
- Knowledge of *BRCA* positive status at the time of treatment may enable cancer patients to avoid additional prophylactic breast surgeries at a later time
- When genetic counseling and testing are indicated, efforts should be made to provide these services prior to surgical cancer treatment decision-making

## References & Acknowledgements

1. Petrucelli N, Daly MB, Feldman GL. *BRCA1* and *BRCA2* Hereditary Breast and Ovarian Cancer. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1998 Sep 4 [Updated 2013 Sep 26]; Available from: <http://www.ncbi.nlm.nih.gov/books/NBK1247/>
2. National Comprehensive Cancer Network Clinical Practice Guidelines in Oncology. "Genetic/Familial High-Risk Assessment: Breast and Ovarian." Version 1.2014. Accessed May 2014. <http://www.nccn.org/index.asp>

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