

Table 2. BRCA Testing Results by Personal History of Cancer, USPSTF Family History Criteria, October 2007– March 2011

	Personal Cancer History*			No Personal History*‡		Known Familial Mutation
	Ovarian cancer	Breast cancer at ≤ 50 years	Breast cancer at > 50 years	Met USPSTF criteria	Did not meet USPSTF criteria	
Negative	153 (73.6)	1,352 (86.8)	676 (90.1)	432 (90.8)	301 (92.9)	345 (52.8)
Positive	44 (21.2)	135 (8.7)	29 (3.9)	23 (4.8)	8 (2.5)	298 (45.6)
Variant	11 (5.3)	71 (4.6)	45 (6.0)	21 (4.4)	15 (4.6)	10 (1.5)
Total	208	1,558	750	476	324	653

* Excluding males and those with a known familial mutation

‡ No personal history of breast and/or ovarian cancer

NCCN guidelines recommend counseling and testing for women with ovarian cancer. Counseling and testing for women with early-age-onset breast cancer is dependent on age, hormone receptor status and family history. As shown in Table 2, over 20% of women with a history of ovarian cancer at any age who received testing were found to have a deleterious BRCA mutation. In women with a personal history of breast cancer at or before the age of 50, 8.7% were positive for a deleterious mutation.

The USPSTF guidelines for genetic counseling referral are intended for women with a family history of cancer; the guidelines do not address personal cancer history. USPSTF family history criteria are used here as an indicator of substantial family history. In women without a personal cancer history or known familial mutation who presented for counseling, those with a family history meeting these criteria had a higher rate of deleterious mutation (4.8%) than those without such a family history (2.5%).

According to NCCN and USPSTF guidelines, those with known familial mutations should always be referred for counseling and testing. Patients with a first degree relative with a known mutation have a 50% probability of inheriting that mutation. In this cohort, 45.6% of patients with any relative with a known mutation tested positive for the family’s known (deleterious) mutation.

These data include genetic counseling visits from October 1, 2007 – March 31, 2011. Special thanks to the following institutions whose de-identified patient information was included in these analyses: Beaumont Health System Cancer Genetics Program, Henry Ford Health System, InformedDNA, Karmanos Cancer Institute Genetics Service, Michigan State University Division of Clinical Genetics, Oakwood Healthcare System’s Genetic Risk Assessment for Cancer Clinic, Providence Hospital Medical Genetics, Spectrum Health Cancer Genetics Program, University of Michigan Cancer Genetics Clinic, and University of Michigan Breast and Ovarian Cancer Risk and Evaluation Program.

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