

Genetic Testing of Individuals Diagnosed with Breast or Ovarian Cancer who Meet Testing Guidelines: Trends in Utah, 2008-2012

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BACKGROUND

Hereditary Breast and Ovarian Cancer (HBOC) and Lynch syndrome (LS) are the two most common hereditary cancer syndromes, affecting approximately 1 in 500 people nationwide. In 2014, The Utah Department of Health received a grant from the CDC to establish a statewide cancer genomics program. This program has three main objectives within the state of Utah:

- 1) Education – increase provider and public awareness of HBOC and LS and appropriate recognition of at-risk families
- 2) Surveillance – assess the amount of appropriate genetic counseling and genetic testing for HBOC and LS being conducted
- 3) Policy – assess and improve insurance coverage of genetic counseling, testing, and management of HBOC and LS

INTRODUCTION

Many individuals who meet guidelines for *BRCA1/2* testing do not pursue testing. This can be due to multiple factors, including patient preference, lack of awareness among patients and providers, financial limitations, or other barriers.

Population-based data can be utilized in unique ways to identify genetic testing uptake among at-risk individuals. Some individuals meet National Comprehensive Cancer Network (NCCN) guidelines for *BRCA1/2* testing based on their personal cancer diagnosis alone. Prior to 2013, only one commercial laboratory offered complete *BRCA1/2* testing in the United States. By collecting data from the state cancer registry and commercial laboratory, it is possible to identify trends in *BRCA1/2* testing and assess for gaps in access.

Utah has a unique resource in the Utah Population Database (UPDB) that matches available health and other records to genealogy data. By querying the state cancer registry and UPDB, it is possible to identify how many individuals in Utah meet the NCCN guidelines for *BRCA1/2* testing based on family history. This data can help inform needs assessments for genetic counseling and testing access and education throughout the state.

METHOD

- Data on the number of diagnoses in each of the following categories was pulled from the Utah Cancer Registry from 2008-2012:
 - Women diagnosed with ovarian cancer
 - Women diagnosed with breast cancer at age 45 or younger
 - Men diagnosed with breast cancer
- The number of *BRCA1/2* tests (comprehensive, multi-site and single site) ordered in Utah within a year of the individual's diagnosis was obtained from the commercial laboratory for corresponding years.
- Data from the Utah Cancer Registry and UPDB was queried to identify the number of individuals currently living in Utah who meet NCCN guidelines for *BRCA1/2* testing.

RESULTS

- In Utah, the proportion of women diagnosed with ovarian cancer who underwent testing within a year of diagnosis was 13.9% in 2008 and increased to 32.9% by 2012 (Fig 1).
- Among women diagnosed with breast cancer at/under age 45, 44% underwent testing in 2008 compared to 78% in 2012 (Fig 2).
- 8.3% of men diagnosed with breast cancer underwent testing within a year of their diagnosis in 2008 compared to 61.5% in 2012 (Fig 3).

Figure 1: Number of *BRCA1/2* tests among women with ovarian cancer

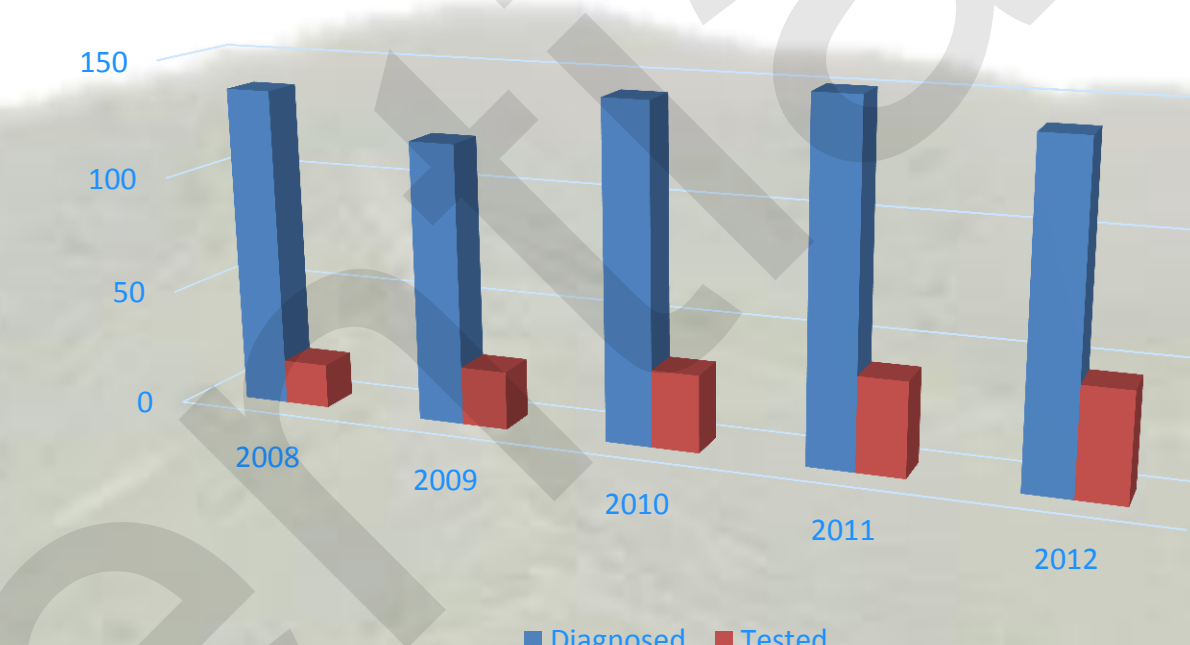


Figure 2: Number of *BRCA1/2* tests among women with breast cancer diagnosed at/under age 45



Figure 3: Number of *BRCA1/2* tests among men with breast cancer

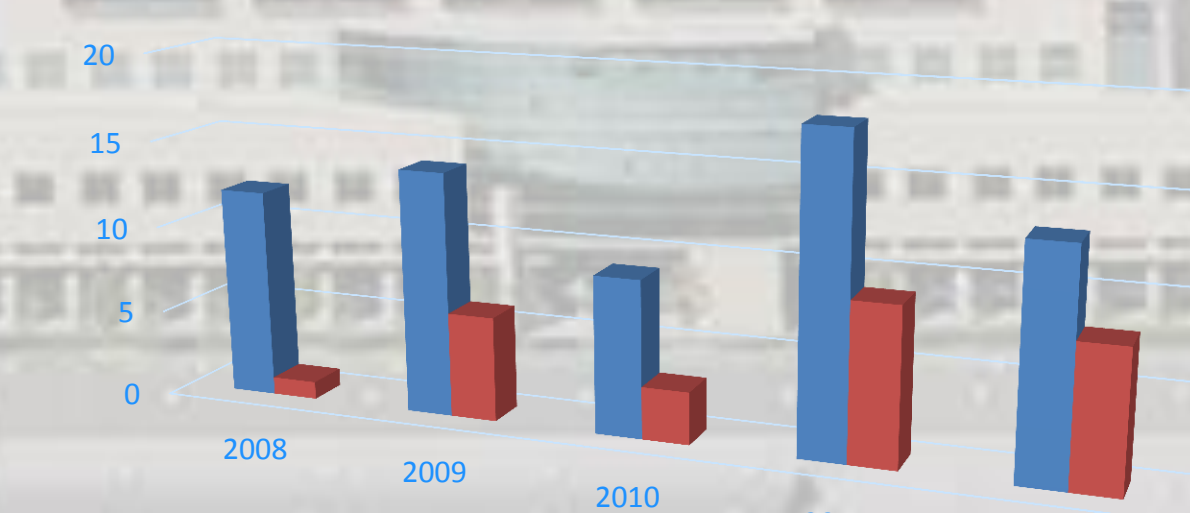


Table 1: Numbers of Affected Individuals Meeting 2015 NCCN *BRCA1/2* Testing Criteria Currently living in Utah

- 1) Personal history of breast cancer and one or more of the following:
 - a) Diagnosed ≤ 45 years old: 3,543
 - b) Diagnosed ≤ 60 years old with triple negative: 267
 - c) Diagnosed ≤ 50 years old with one of the following criteria:

Criteria	Total Alive/In Utah
An additional breast cancer primary	542
≥1 close blood relative with breast cancer at any age	1,863
≥1 close blood relative with pancreatic cancer	347
≥1 close blood relative with prostate cancer (gleason score ≥7)	790
An unknown or limited family history	50

- d) Diagnosed at any age with one of the following criteria:

Criteria	Total Alive/In Utah
≥1 close relatives with breast cancer diagnosed ≤ 50	2,439
≥2 close relatives with breast cancer diagnosed at any age	2,890
≥1 close relatives with Ovarian cancer	1,474
≥2 close relatives with Pancreatic or prostate	993
A close relative with male breast cancer	79

- 2) Personal history of ovarian cancer 1,104

- 3) Personal history of male breast cancer 88

Table 2: Affected and Unaffected Individuals meeting 2015 NCCN *BRCA1/2* testing criteria currently living in Utah (2016)

Testing Criteria (NCCN Guidelines 2.2015)	Sex	Degree of relative affected	Total Alive/In Utah
Affected with Breast/Ovarian/Prostate or Pancreatic cancer and meets one or more of the testing criteria	F	1-3	9,374
	M	1-3	2,771
	Total		12,145
Unaffected and 1st or 2nd relative meets testing criteria	F	1	41,375
	F	2	120,285
	M	1	41,735
	M	2	124,348
	U	2	1
Unaffected who has 3 relatives (breast, ovarian) one relative diagnosed ≤ 50 years with breast cancer one relative is 3 rd degree	Total		327,744
	F	1-3	1,147
	M	1-3	1,191
	Total		2,338

DISCUSSION

- The appropriate utilization of *BRCA1/2* testing in Utah for individuals with a breast or ovarian cancer diagnosis meeting NCCN guidelines increased dramatically during the time period studied.
- However, these data indicate that barriers to testing persist, particularly among patients with ovarian cancer.
- Further educational and outreach efforts are warranted, and research should further explore the barriers to testing within the Utah population.
- The large number of unaffected relatives meeting NCCN *BRCA1/2* testing criteria in Utah further highlights the importance of starting testing with an affected relative whenever possible. Further testing in many families is unnecessary when a negative test result exists on an affected relative.
- Further exploration of ways to increase access to genetic counseling services throughout the state (i.e. telephone/telemedicine based counseling, etc.) are needed to serve the large demand.

ACKNOWLEDGMENTS AND REFERENCES

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