Women At Risk For Hereditary Breast Ovarian Cancer Syndrome: The Value of a Genetics Clinic Referral

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BACKGROUND

Each year more than 250,000 women are diagnosed with breast or ovarian cancer. Of these women, 5-10% develop cancer due to an inherited genetic variant. Prevention or early detection in this subset of women could be identified by genetic testing prior to a cancer diagnosis Because genetic testing is covered by insurance only when certain medical or family history criteria are met, an accurate and complete family history is an essential step prior to genetic testing. The National Cancer Comprehensive Network (NCCN) and the US Preventative Services Task Force (USPSTF) guidelines recommend that primary care providers elicit a screening personal and family history of cancer, and make appropriate referrals to cancer genetics professionals.

The hypothesis of our project is that latent information present (but not recognized by treating providers) stored in the electronic health record (EHR) can identify patients who should be referred to the Genetics Clinic

METHODS

To test the accuracy of family history information by non-genetics providers, we:

- Reviewed the EHR of 56 women (probands) selected from a randomly generated cohort of 1000 women.
  - Age > 30 years
  - Seen in UW Medicine at least 5 times
  - Seen in Genetics Clinic at UW
- Compared family history
  - Before Genetics visit
  - After Genetics visit
- Analyzed genetic testing
  - If pursued
  - Results of testing

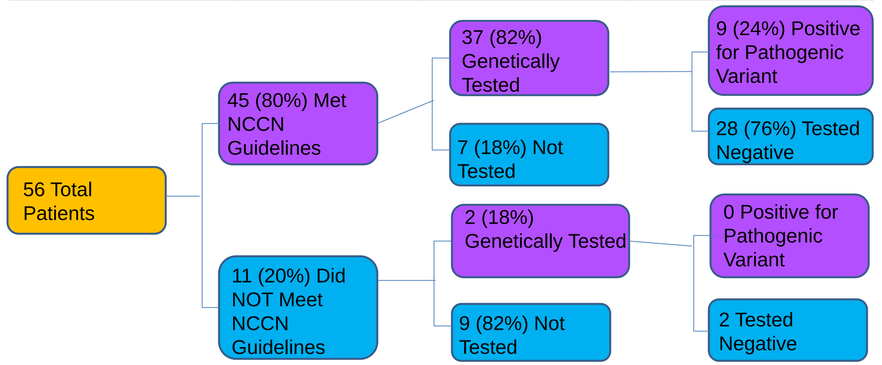
RESULTS

	Reported with Cancer	Confirmed by Genetics	Confirmed Cases with data changed	Percent Confirmed	Percent Changed from Reported	Percent Changed from Confirmed
Mother	22	22	17	100.0%	77.3%	77.3%
Maternal 2nd Degree	32	26	23	81.3%	71.9%	88.5%
Maternal 3rd Degree	8	6	4	75.0%	50.0%	66.7%
Father	14	14	12	100.0%	85.7%	85.7%
Paternal 2nd Degree	25	21	14	84.0%	56.0%	66.7%
Paternal 3rd Degree	9	6	2	66.7%	22.2%	33.3%
Siblings	11	11	7	100.0%	63.6%	63.6%
Totals	121	106	79	87.6%	65.3%	74.5%

	Before Genetics	After Genetics
Total Relatives With Cancer Reported	121	242

	Yes	No	Percent Yes
NCCN Guidelines Met	45	11	80.4%
Genetics Visit Changed Family History	24	32	42.9%
Genetic Testing Perfomed	40	16	71.4%

	Total Tested	Positive Tests	Percent Positive
Genetic Testing	40	9	22.5%



DISCUSSION

Results interpretations:

- 24/56 (43%) of patient's family histories changed significantly after the Genetics visit
- Relatives reported with cancer doubled from 121 to 242 .
- 79/106 (75%) relatives with a confirmed cancer diagnosis had details of the diagnosis changed at the visit .
- 45/56 (80%) women referred to the genetics clinic met NCCN guidelines for genetic testing.
- 9/40 (23%) tested women were positive for a Likely Pathogenic or Pathogenic (LP/P) variant.

Limitations:

- Number of completed primary and secondary reviews of charts

Future work:

- Characterize if women meet NCCN guideline based on family history, personal history, or both.

CONCLUSIONS

This pilot study of 56 women demonstrates the value of a Genetics Clinic visit via:

- Improved rates of identifying the key family history components for identifying women at increased risk of hereditary cancers.
- More appropriate use of genetic testing for risk assessment.

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