Genetic Screening Intervention: Identifying Carrier Status with a History of Cancer

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ABSTRACT

It is estimated 5-10% of all breast cancer can be attributed to a hereditary predisposition. A study in 2003 determined 9% of women with a significant family history warranted a genetic conversation.

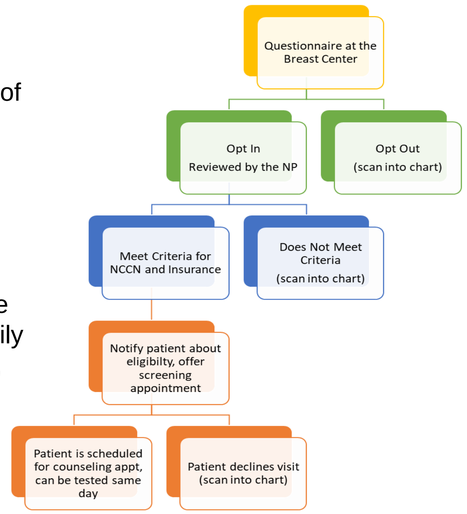
In 2016, a multidisciplinary team created a cancer genetic screening and high risk for breast cancer program. These programs were created to provide cancer risk assessment, genetic cancer screening, genetic cancer evaluation and testing, and develop a treatment plan with the patient.

OBJECTIVES

Screen women and men in the Breast Center for a personal and family history of cancer, not just breast cancer.

Determine who meets criteria, per NCCN, and insurance eligibility.

Have a consultation with the NP about personal and family history, develop a pedigree, test if appropriate.



METHODS

A multidisciplinary team was constructed to develop a risk assessment, genetic screening, evaluation, testing, and treatment plan protocol. This program began January 4, 2016.

The first year of the program, demonstrated our patient population is at a 5.7% risk of having a hereditary cancer predisposition. In addition, our program found a 7.2% high risk breast cancer population. During the second year, we had a hereditary cancer predisposition of 6.6% and a high risk breast cancer population of 9.4%. During this third year, our population of hereditary cancer predisposition is at 7.0% and a high risk population of 6.4%.

During the two years and ten months of the program, a new question arose. How many patients positive for a genetic mutation had a personal diagnosis of a Hereditary Breast and Ovarian Cancer (HBOC) classified cancer? An HBOC classified cancer includes breast, ovarian, prostate, melanoma, and pancreatic cancers (CDC, 2015).

A total of 69 positive mutations were identified. Of those, 26 patients had a personal history of a breast, ovarian, prostate, pancreatic, or melanoma cancer. **This equates to a 38% of our positivity rate is associated with HBOC classified cancer.** Not all the mutations identified are associated with HBOC.

3-year results

BRCA1	18
BRCA2	7
CHEK2	6
PALB2	1
ATM	4
PTEN	1
MSH6	1
SMAD4	1
PMS2	3
Rad51C	2
NBN	5
BARD1	2
BRIP1	2
MUTYH	21
MLH1	2
MSH2	1
CDKN2A	1

Breast / HBOC
Colon/Lynch
Ovarian
Mel/Panc

RESULTS

	2016	2017	2018
Total patients seen in breast center	11196	11084	10985
total patients screened for family history	9726	9702	10120
Total patients with family history of cancer	5008	6449	7183
total patients meeting NCCN criteria	1184	1263	1179
Total patients with >20% TC	705	643	679
Attempted tests	317	360	336
Overall results	292	336	314
Denied cases	25	24	22
Positive cases	18	34	26
Percent of Positive cases	6.16%	10.12%	8.28%

CONCLUSIONS

During the past few years our program has evolved from bringing genetic evaluation to a genetically underserved area, to developing a program which includes cancer risk assessment, genetic cancer screening, genetic cancer evaluation and testing, and developing a treatment plan all in one location. In the population surveyed, we have found as high as 6.7% high risk for breast cancer. Most general surgery offices treat breast cancer and in some cases treat women at increased risk for breast cancer. General surgery needs to spearhead genetic testing in the breast cancer population. As seen here, over one-third of our positive mutations were found in a patient with an HBOC classified cancer. It is imperative to bring awareness for a genetics risk assessment to those who treat breast cancer, the surgical office. This program was developed to reduce the risk of breast cancer in our community. A program using a multidisciplinary approach should be utilized in General Surgery and Breast Care Clinics to perform genetic risk assessments.

REFERENCES

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Abstract 582132