Background
Breast cancer is caused by an inherited predisposition in approximately 5-10% of women and 10% of men with breast cancer. Additionally, up to 25% of women with ovarian cancer can attribute their cancer to a hereditary condition. Germline mutations in breast and ovarian cancer susceptibility genes often cause cancer at a younger age than patients with wild-type status, and many of these germline mutations also increase the risk for other types of cancer, especially endometrial cancer. It is important to identify patients at risk for cancers associated with known genetic mutations, and to implement appropriate risk-reducing interventions and screening protocols as recommended by groups such as the National Comprehensive Cancer Network (NCCN) in order to reduce their cancer risk. Based on our review of literature, the majority of patients who carry such genetic mutations do not receive the appropriate genetic counseling and recommended interventions. Prior studies highlight the importance of identifying potential barriers to the proper recognition of these patients and the timely use of genetic counseling, recommended genetic testing and risk-reducing procedures.

Methods
We identified approximately 200 patients between 7/1/2013 and 6/30/2016 with breast or ovarian cancer at a large cancer treatment center who met NCCN criteria for genetic counseling and testing based solely on their personal cancer diagnosis.

Results
Approximately 70% of the included patients underwent appropriate genetic testing and counseling. Of those that did not undergo genetic counseling and/or testing, most were not referred to genetic counseling services by their oncologist. Older patients (>65 years old) were also less likely to be referred to genetic counselors than younger patients (<65 years old), and were more often counseled by a physician rather than by a certified genetic counselor. Once found to have a pathogenic hereditary mutation, most patients elected to undergo appropriate risk-reducing procedures, surveillance, and family discussions.

Conclusions
Our results indicate that receiving the initial referral to either genetic testing or genetic counseling may be the biggest hurdle to overcome for these at-risk patients at our institution.

Background
NCCN guidelines outline patient criteria for those diagnosed with breast and ovarian cancer that should be offered genetic counseling and testing based on their personal and/or family history. Approximately 5-10% of individuals with breast cancer and up to 25% of women with ovarian cancer can attribute their disease to an inherited gene mutation, thus representing thousands of patients each year who are also at potential risk for another primary cancer and have relatives that may also be at risk. Although the BRCA1/2 genes makeup the majority of causes of hereditary breast and ovarian cancers, mutations in other cancer genes also significantly contribute to hereditary cancer syndromes.

Genetic counseling is a key healthcare component for patients at risk for these heritable mutations. Unfortunately, genetic counseling is widely underutilized in the oncology setting, despite NCCN guidelines and recommendations. Insufficient use of these resources may negatively impact the level of suffering and cost in potentially preventable cancer diagnoses.

A retrospective chart review was conducted to determine the number of breast and ovarian cancer patients who met NCCN criteria for genetic testing, received genetic counseling, and testing, and NCCN guideline-directed risk-reduction management at a single large cancer center.

The purpose of this study was to identify the barriers to the use of genetic counseling and genetic testing in a high-risk population.

Methods
A retrospective chart review was conducted to evaluate patients who were diagnosed with breast or ovarian cancer at Baylor Scott & White Cancer Center between 7/1/2013 and 6/30/2016.

Study Participants
All patients (21 total) met one of the following NCCN criteria at the time of their diagnosis:
1. Women diagnosed with breast cancer (including DCIS) at or before age 65.
2. Women diagnosed with ductal carcinoma in situ (DCIS) at or before age 65.
4. Diagnosed with breast cancer at or before age 50 with a first-degree relative diagnosed with breast cancer at any age.
5. Breast or ovarian cancer at any age.
6. Breast or ovarian cancer at any age.

Data Analysis
Patients were identified who underwent genetic counseling with or without genetic testing.

If a patient was noted to have a pathogenic germline mutation, further medical review was performed to investigate whether they were appropriately counseled or pursued prophylactic surgery, as recommended by NCCN guidelines for "Genetic/Hereditary High-Risk Assessment Breast and Ovarian Cancer" (version 1.2015, from October 5, 2015).

Contact Information
Please feel free to contact the authors at CancerGenetics@texashealth.org

References

Discussion
This study indicates that appropriate use of genetic counseling and testing as recommended by NCCN guidelines was occurred for nearly 70% of the identified cohort. This rate of use far exceeds those previously reported in other publications.

Of the patients that did not receive genetic counseling, the overwhelming majority were not referred to the genetic counseling department. This suggests that most patients are interested in pursuing genetic counseling, as such, physician education on the appropriate referral process is critical for patients to receive comprehensive cancer care.

These results indicate that receiving the initial referral to either genetic testing or genetic counseling may be the biggest hurdle to overcome for these at-risk patients.

This study did not identify any notable disparities in use of genetic counseling/testing among the patient demographics when sorted by race.

This study did not find that unrepresented patients were less likely to receive genetic counseling/testing compared to patients, possibly due to patient financial assistance programs in place by the genetic counseling program and commercial laboratories for such patients.

Older patients (>65 years old) were less likely to be referred to genetic counseling if their primary care provider was (65 years old), and were more often counseled by a physician rather than by a certified genetic counselor.

The implementation of genetic counseling for heritable cancer risk in patients with pathogenic mutations who underwent genetic counseling were largely in keeping with recommended risk-reducing procedures, heightened surveillance, and familial risk assessment.

A notable difference was observed in whether family planning and risk reduction counseling were delivered by genetic counselors or by physicians who performed genetic counseling. More often, physician-directed counseling did not address these two important components of hereditary conditions. As such, this study suggests that every patient with indicators for genetic counseling be seen by a certified genetic counselor to ensure full assessment of risk at the patient and family.

Conclusion
Lack of timely physician referrals was identified as the single most important barrier to the use of certified genetic counselors and genetic testing at the study’s institution.

This finding has prompted the genetic counseling program to re-evaluate their efforts at physician education regarding NCCN guidelines for genetic testing for breast and ovarian cancer patients and the available resources at the institution.

Further plans for increased collaboration between physicians and certified genetic counseling are ongoing.

With better use of current NCCN guidelines, oncologists and other health care providers will be able to more effectively guide patients and their families to receive appropriate risk management.

The role of genetic counselors is becoming an increasingly important part of healthcare, and there remains a need to establish institutional barriers to their use.