ANALYSIS OF VARIABLES AFFECTING CASCADE TESTING FOR HEREDITARY GENETIC CANCER RISK

Atrium Health

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INTRODUCTION

- Studies across ethnic backgrounds, education levels, and socioeconomic statuses have shown that barriers to cascade testing include health insurance, cultural differences, and the patient's understanding of the result
- White populations have undergone the most testing for *BRCA1*/*BRCA2* when compared to Black and Hispanic populations, while Black women have had lowest testing rates
- Cost and lower exposure to genetic counseling are barriers to minority populations receiving genetic testing
- Additional studies have suggested that cascade testing rates across all populations could be improved
- Bednar et al cited a 30.5% uptake in cascade testing, despite 70.3% of first-degree relatives indicated knowing they were at a high hereditary risk for cancer

OBJECTIVE

Determine whether a patient's demographics or the gene in which a pathogenic variant was identified is correlated with the likelihood of cascade testing in families. It is important to understand how demographic trends and genetic testing results are correlated with cascade testing rates to allow further customization of cascade testing resources for patients and families.

METHODS

This retrospective study evaluated the prognostic value of baseline demographic factors on cascade testing rates. The odds of each group receiving cascade testing (defined as at least one relative having pursued cascade testing and genetic counseling) were determined by comparing to a reference group, usually the group with the highest number of participants.

- This study included adult patients who received genetic counseling and had a pathogenic variant in a hereditary cancer gene identified by genetic testing between 2010-2019.
- Logistic regression analysis was used to evaluate the prognostic value of baseline demographic factors on the rate of cascade testing.
- Univariate models were used to identify individual prognostic factors. Multivariable models, including backward elimination, were used to identify independent prognostic factors. The same analyses were utilized for gene-specific assessments.
- Genes were placed into disease groups based upon the highest correlation with a specific cancer type. For instance, *ATM, BRCA1, BRCA2,* and others most associated with breast cancer were grouped in the "Breast" disease group.

DEMOGRAPHICS

Of 1,709 patients:

- 79% were female with 81% White and 13% Black
- Age groups were 18-34 years (18%), 35-50 years (34%), 51-69 years (38%), and ≥70 years (10%)
- Median household income was $60,000
- Insurance type was 60% private, 17% Medicare, 13% unknown, 5% self-pay, and 0.5% no charge

RESULTS

By univariate analysis, race, sex, and income were not associated with cascade testing uptake (p=0.86, p=0.98, p=0.05, respectively).

UNIVARIATE ANALYSIS	
Carrier status and the gynecologic disease group were associated with **decreased odds of a relative pursuing cascade testing** as compared to the breast disease group	**P=.01 AND P=.047**
Older age was associated with **decreased odds** of cascade testing	**P=.01**
Patients with an **unknown insurance type** or testing that was **performed at no charge** had increased odds of a relative pursuing cascade testing as compared to those with private insurance.	**P=.001 and P=.01**

MULTIVARIABLE ANALYSIS	
Patients with **carrier status had lower odds of cascade testing** compared to patients with gene variants associated with breast cancer	**Odds Ratio = 0.27 95% CI: 0.17-0.41, P<.001**
Older age was associated with **decreased odds** of cascade testing	**Odds Ratio = 0.91 95% CI: 0.83-0.99, P=0.03**
Compared to patients with private insurance, patients who were **not charged or had unknown insurance were more likely** to have a relative who underwent cascade testing	**Odds Ratio = 10.49 95% CI: 1.98-55.61, P=0.01** **Odds Ratio = 2.09 95% CI: 1.52-2.87, P<.001**

DISCUSSION

1. Our study indicates the need for educational and supportive resources for families who may benefit from cascade testing.
2. Further studies of large, diverse populations should investigate how a genetic counselor can assist in coordinating cascade testing, and the potential impact a family letter can have on uptake.
3. In our population, older age was associated with a decreased odds of cascade testing. This suggests that additional support can be offered to older patients. Further study could evaluate which barriers older patients are facing.
4. Carrier status had a lower odds of cascade testing. Resources could be developed specifically for this population.
5. For patients with private insurance, it could be a useful tool to have resources for them compared to those with testing at no charge to help them understand familial testing options that may also be at no cost to them.

FUTURE DIRECTIONS

Future research could be conducted to better understand the motivations behind some of the quantitative findings found in this study.

- Specifically, understanding motivations for cascade testing with specific gene types in a qualitative form could be better described in the future
- Motivations for cascade testing as well as resources/barriers for patients who are of older age may be an area that can provide further assessment.
- Qualitative research focused on motivations for cascade testing in the populations of individuals who are of older age, or have a carrier status, could provide rich data to the understanding of this area. Carrier status in a prenatal versus cancer setting may be of interest to understand.
- We acknowledge that most of our patients were White individuals; there is ample need to promote future studies that have a more diverse background of individuals to further understand cascade testing in all populations.

Overall, future research is needed to determine the best way for genetic counselors to facilitate cascade testing for hereditary cancer at their respective institutions.

Scan here for references: