Introduction

Access to genetic testing has rapidly expanded as the cost of DNA sequencing decreases. As traditional delivery models of genetic testing are often time consuming for patients and providers, the use of alternative models to deliver genetic testing are being explored. Color has implemented one such model, integrating new elements of the genetic testing process such as online ordering, telephone genetic counseling sessions, and reports tailored for both providers and patients. All testing is provider ordered and can either be ordered by a patient’s own provider (traditional model) or by a third-party network of providers (non-traditional model).

Previous research on outcomes after genetic testing are scarce due to the new and unique nature of many genetic tests. Although past literature is limited, some research revealed the rate of colonoscopies decreased from 59% to 8% before and after genetic testing among negatives, while others found as many as 32% of BRCA1 and BRCA2 carriers are not compliant with annual mammography recommendations.

The purpose of this study is to explore and compare cancer screening behavior in individuals who received a genetic test for hereditary cancer through traditional and non-traditional models of testing. Specific aims of the study include: 1) Compare self-reported screening behavior to recommended guidelines, and 2) assess differences in behavior between models of testing.

Methods

In this study, a quantitative retrospective computer administered survey was used to analyze the outcomes of Color clients who have negative results (no mutations identified), increased risk (no mutations identified, increased risk of breast cancer according to the Gail or Claus model), and positive results (pathogenic mutation identified) in only the BRCA1, BRCA2, and Lynch syndrome genes MLH1, MSH2, MSH6, PMS2, and EPCAM. Individuals aged 18-90 who had a negative, increased risk, or pathogenic mutation report one year or more prior to the start of data collection were eligible for this study. An email invitation to participate in the study was sent to a total of 3,436 individuals and 249 individuals responded. Participants had the option to enter into a raffle to win one of ten $75 gift cards upon completion of the survey.

Survey responses were collected in an anonymized manner. Demographic information previously provided by the individual was attached to the survey responses including age, gender, and result type. However, no personal identifying information was collected. All individuals consented to have their de-identified information used in this anonymized study. All information was reported by the individuals who participated.

The survey data was analyzed in aggregate with difference in proportions hypothesis testing to explore differences between the traditional and non-traditional models. Cancer screening actions were compared to screening recommendations by the National Comprehensive Cancer Network for Genetic/Familial High-Risk Assessment Breast and Ovarian (BRCA1 and BRCA2) Genetic/Familial High-Risk Assessment Colorectal (Lynch syndrome genes), and Breast Cancer Screening and Diagnosis(1) (Negative and Negative with increased risk). Specific screening actions assessed by the survey and used for analysis are detailed in Table 1. Participants were asked “One year before you took this genetic test, which of the following had you done?” and “Since taking this genetic test, which of the following screening actions have you taken?” and given the option to select all answer choices that applied. The answer choices included but were not limited to: “Mammogram”, “Breast exam by a healthcare provider”, “Breast imaging using MRI”, and “Colonoscopy.” Timing of these actions post-testing was collected and analyzed.

<table>
<thead>
<tr>
<th>Table 1. Guidelines</th>
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<tr>
<td>Breast cancer screening actions derived from NCCN guidelines used to gauge client compliance levels. Breast cancer screening actions are recommended until age 75 unless otherwise indicated, upon which screening becomes individualized.</td>
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<table>
<thead>
<tr>
<th>Population</th>
<th>Screening Action</th>
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<tbody>
<tr>
<td></td>
<td>Mammogram</td>
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<tr>
<td>Negative Female</td>
<td>Every year from age 40</td>
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<tr>
<td>Negative Male</td>
<td>Every year from age 40</td>
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<td>Negative Female, increased risk (Claus) Negative Female, increased risk (Gail) BRCA1 positives Female BRCA1 positives Female BRCA2 positives Female BRCA2 positives Female BRCA2 positives Male BRCA2 positives Male Lynch positives Female Lynch positives Male</td>
<td>Every year from age 30</td>
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</table>

Conclusions

- Participants in the traditional model may have done slightly more screening actions before testing due to increased provider contact.
- However, genetic testing through this non-traditional model can educate about screening guidelines and spur knowledge and compliance to the same degree as a traditional model post-testing.
- Based on the data presented, the non-traditional model of genetic testing is not inferior to the model of traditional genetic testing with respect to compliance with gene-specific recommendations.

References