Participant screening behaviors and information sharing after genetic testing

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Background

Advancements in genotyping arrays and next generation sequencing (NGS) technologies as well as decreasing costs have made genetic testing more affordable and accessible. Public interest in genetic testing has also grown in recent years, with more than 26 million Americans having had genetic testing for ancestry and/or health to date.[1] Clinical-grade genetic testing is most often offered through healthcare providers through an in-office consultation, however, this process can be time-consuming and cumbersome for both the healthcare provider and the patient.

Color offers an alternative delivery model for genetic testing performed in a CAP-accredited and CLIA-certified laboratory. Color's end-to-end delivery model includes online enrollment, provider education, personalized reports with screening guidelines, complimentary access to genetic counselors, and cascade screening. Importantly, genetic testing can be ordered by the individual's healthcare provider in-office (traditional model) or by an independent healthcare provider from an external network (non-traditional model).

However, data on how genetic testing offered through traditional and non-traditional models impact health outcomes is limited. Past studies have found that as many as 32% of *BRCA1* and *BRCA2* carriers are not compliant with annual mammography recommendations[2] and that the rate of colonoscopies decreased from 59% to 8% after genetic testing among individuals with negative results.[3] To our knowledge, no research has examined screening behavior and information sharing between non-traditional and traditional genetic testing models.

Objectives

To 1) compare screening behavior with recommended screening guidelines and 2) assess information sharing behavior in individuals who underwent genetic testing for hereditary cancer risk through traditional (ordered by a healthcare provider in-office) and non-traditional (ordered by an independent healthcare provider from an external network) models.

Methods

A quantitative, retrospective computer-administered survey was used to analyze outcomes of Color clients who received clinical-grade genetic testing for genes associated with hereditary breast and ovarian cancer (*BRCA1* and *BRCA2*) and Lynch syndrome (*MLH1, MSH2, MSH6, PMS2,* and *EPCAM*). Individuals who received a negative or positive report one year or more prior to the start of data collection were eligible for this study and invited to participate through email.

Individuals who agreed to participate gave informed consent for their de-identified information to be used in this study. All phenotypic information was self-reported by the individual through an online, interactive health history tool. Participants were entered into a raffle to win one of ten $75 gift cards upon completion of the survey.

The survey data was analyzed in aggregate to explore behavioral differences between traditional and non-traditional models of testing. Comparisons between non-traditional and traditional models were calculated using Fisher's exact test. Cancer screening actions were compared 1) to population screening recommendations as provided by the National Comprehensive Cancer Network (NCCN) for Breast Cancer Screening and Diagnosis[4] and for Colorectal Cancer Screening[5] and 2) to gene-specific screening recommendations as provided by NCCN for Genetic/Familial High-Risk Assessment Breast and Ovarian[6] and Genetic/Familial High-Risk Assessment Colorectal.[7]

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Disclosures

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Results

Table 1. Breast and colorectal cancer screening guidelines as provided by NCCN by result type.

Population screening actions are recommended for breast cancer until age 75 and for colon cancer from age 50-75. However, screening actions become personalized for individuals with confirmed pathogenic variants (gene-specific recommendations).

Screening Action	Negative Results	Positive Results					
	Female	*BRCA1* Female	*BRCA1* Male	*BRCA2* Female	*BRCA2* Male	Lynch Female	Lynch Male
Mammogram	Every year from age 40	Every year from age 30	--	Every year from age 30	--	--	--
Breast exam by a healthcare provider	Every 1-3 years from age 25-40; Every year from age 40-75	Every 6-12 months from age 25	Every year over age 35	Every 6-12 months from age 25	Every year from age 35	--	--
Breast imaging using MRI	--	Every year from age 25	--	Every year from age 25	--	--	--
Colonoscopy	--	--	--	--	--	Every 1-2 years from age 25	Ever 1-2 years from age 25

Table 2. Participant characteristics.

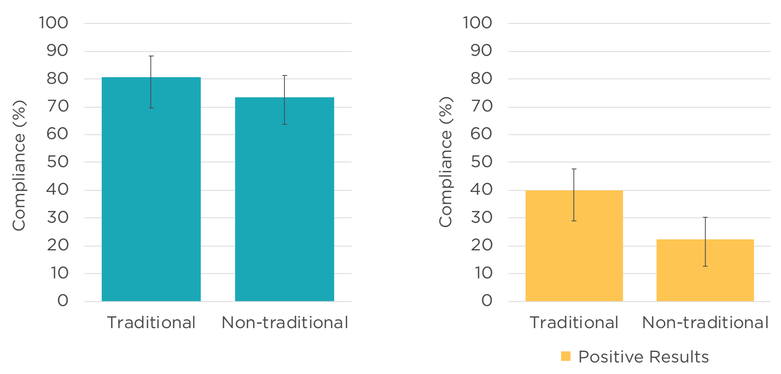
The cohort included 161 participants, the majority of which were women, over age 40 years, and Caucasian. Approximately half of participants had completed a bachelor's degree or higher. Participant characteristics were similar between traditional and non-traditional models of genetic testing.

		Participants n	Population (%)	Traditional n (%)	Non-traditional n (%)
Total		161	100	67 (100)	94 (100)
Gender	Male	30	18.63	13 (19.40)	17(18.09)
	Female	131	81.37	54 (80.60)	77(81.91)
Age (Years)	18-25	4	2.48	0 (0.00)	4 (4.26)
	26-40	49	30.43	17 (25.37)	32 (34.04)
	41-75	99	61.49	45 (67.16)	54 (57.45)
	65+	9	5.59	5 (7.46)	4 (4.26)
Ethnicity	Asian	14	8.70	5 (7.46)	9 (9.57)
	Caucasian	129	80.12	53 (79.10)	76 (80.85)
	Hispanic	6	3.73	2 (2.99)	4 (4.26)
	Native American	1	0.62	0 (0.00)	1 (1.06)
	Multiple Ethnicities	5	3.11	2 (2.99)	3 (3.19)
	Prefer not to answer	6	3.73	5 (7.46)	1 (1.06)
Education	High school graduate, diploma or equivalent	7	4.35	2 (2.99)	5 (5.32)
	Trade/Technical/ Vocational training	6	3.73	4 (5.97)	2 (2.13)
	Some college, no degree	24	14.91	11 (16.42)	13 (13.83)
	Associate degree or equivalent	14	8.70	7 (10.45)	7 (7.45)
	Bachelor's degree	57	35.40	20 (20.85)	37 (39.36)
	Graduate degree	52	32.30	22 (32.84)	30 (31.91)
	Prefer not to answer	1	0.62	1 (1.49)	0 (0.00)

Figure 2. Compliance with screening guidelines prior to genetic testing.

(A) Compliance with population screening guidelines was similar between the traditional (80.6%) and non-traditional (73.4%) models (p = 0.348).

(B) Among those with positive results, compliance with gene-specific screening guidelines was similar between the traditional (40.0%) and non-traditional (22.4%) models (p = 0.096).

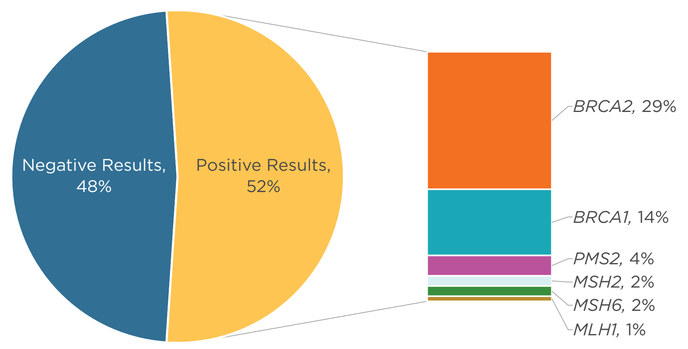


Conclusions

- Based on the results of this study, a non-traditional model of genetic testing is as effective as the traditional model of genetic testing with respect to compliance with recommended screening guidelines.
- Compliance with screening guidelines was similar between traditional and non-traditional models, regardless of test results. Participants in the traditional model may have been slightly more compliant before genetic testing due to contact with their healthcare provider.
- Sharing with family members was similar between traditional and non-traditional models, regardless of test results. Sharing results with family members can help them understand their own cancer risks.
- Approximately 80% of participants in this cohort were Caucasian and female so further research is needed to understand screening behaviors and sharing with family members in a more diverse population.

Figure 1. Test outcome by type.

(A) Approximately half of participants in the traditional model had a positive result (n = 35). Pathogenic variants were most frequently found in *BRCA2* (n = 19) and *BRCA1* (n = 7).



(B) Approximately half of participants in the non-traditional model had a positive result (n = 49). Pathogenic variants were most frequently found in *BRCA2* (n = 27) and *BRCA1* (n = 13).

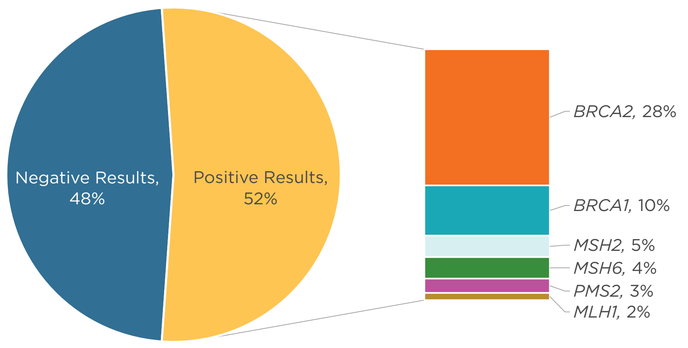


Figure 3. Compliance with screening guidelines after genetic testing.

Among those with negative results, compliance with population screening guidelines was similar between the traditional (68.8%) and non-traditional (64.4%) models (p = 0.808). Among those with positive results, compliance with gene-specific screening guidelines was similar between the traditional (78.1%) and non-traditional (77.8%) models (p = 0.356).

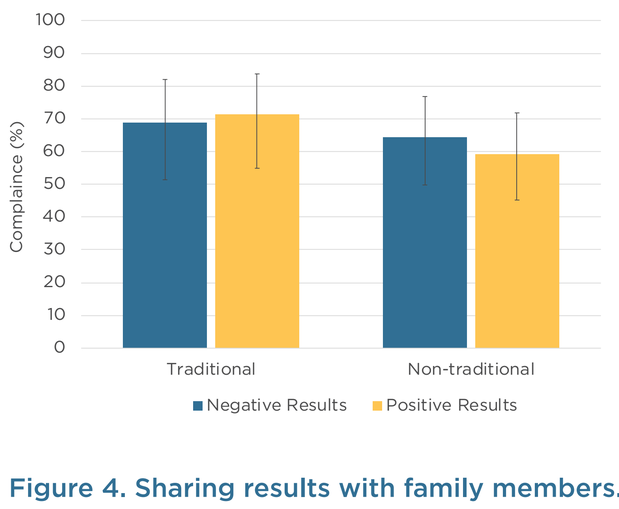


Figure 4. Sharing results with family members.

Among those with negative results, sharing with family members was similar between the traditional (93.8%) and non-traditional (86.7%) models (p = 0.457). Among those with positive results, 100% of individuals shared with family members in both models.