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Modes of Delivery of Genetic Testing Services and the Uptake of Cancer Risk Management Strategies in *BRCA1* and *BRCA2* Carriers

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Abstract

BRCA testing services are now offered by various healthcare providers, thus it is important to evaluate whether the implementation of cancer risk management (CRM) strategies varies by service provider. Using a registry-based sample of 795 female *BRCA* mutation carriers, we explored the association between uptake of CRM strategies with duration of genetic counseling (GC) sessions, provider type, and other demographic and clinical variables. All participants completed a baseline questionnaire. Information about uptake of CRM strategies was collected for a subset of 438 participants who completed additional questions. Summary statistics and Pearson Chi square tests were used to examine the associations between demographic and clinical variables with service delivery factors and with the uptake of various CRM strategies. Overall uptake of CRM strategies was high across all provider types. However, GC sessions were longer when provided by a genetics professional than by another provider ($p<0.001$). Furthermore, higher frequencies of uptake of most CRM strategies were associated with longer GC sessions and when testing was done with involvement of a genetics professional. Identification of factors to optimize delivery of these specialized GC services is important to maximize implementation of CRM strategies in *BRCA* carriers.

Keywords

BRCA; Genetic testing; Genetic counseling; Clinical practice delivery

INTRODUCTION

The discovery of the *BRCA1* and *BRCA2(BRCA)* genes (1, 2) offers an opportunity to identify women at elevated risk for breast and ovarian cancer prior to diagnosis and provide appropriate medical services(3, 4). Women with *BRCA* mutations are estimated to have

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CONFLICT OF INTEREST: None

lifetime risks of 70% for breast cancer and 20%–40% for ovarian cancer(4). Guidelines exist for women identified as carrying a *BRCA* mutation regarding cancer risk management (CRM) strategies, such as screening, chemoprevention or preventive surgery(5). Furthermore, there are national recommendations which outline the essential elements to be covered when delivering cancer genetic risk assessment (CGRA) services during conduct of *BRCA* testing(6, 7).

The decision to pursue a specific CRM strategy depends on several factors, including patient preferences, access to care and provider-related factors(4, 8–10). However, there remains a lack of data to determine whether the mode of delivery of genetic testing services (i.e., how and by whom) is associated with subsequent uptake of the various CRM strategies. In fact, especially as CGRA services (including genetic counseling (GC) and genetic testing) have gradually shifted from primarily an academic-based clinical activity (offered by genetics professionals) to a community-based setting (offered by a wide range of providers)(11), this information is needed to inform policy decisions aimed at implementing high-quality and cost-effective genetic testing services and follow-up.

The objective of the current study was to determine whether the time devoted to the delivery of CGRA services differs by provider type. Furthermore, we explored the association between uptake of CRM strategies in female *BRCA* mutation carriers with duration of genetic counseling (GC) sessions, provider type, and other demographic and clinical variables.

MATERIALS AND METHODS

Study Population

Study subjects were women drawn from the Inherited Cancer Registry (ICARE) database between age 21–80, with or without cancer, living in the United States who self-reported carrying a *BRCA* mutation. The ICARE initiative was launched in summer 2010 to assess factors associated with adherence to CRM strategies in *BRCA* carriers. Participants in ICARE are recruited through various clinical centers, directly online through the registry website (www.moffitt.org\ICARE), and through local and national outreach activities, in an effort to provide patients tested across diverse settings with a research link(4).

All women were tested in the only commercial laboratory that performs *BRCA* testing in the United States. Participants were residents from 46 states, tested at the discretion of treating healthcare providers prior to enrollment in ICARE between 2010–2012. The study was approved through the University of South Florida's Institutional Review Board.

Procedures

A cross-sectional survey at the time of study recruitment recorded demographic and clinical information. The method of GC service delivery was recorded through collecting information on the specialty of the provider who ordered testing, provided GC, and length of GC sessions. Genetics professionals were defined as masters-trained genetic counselor (working in conjunction with a physician, when listed as the individual who ordered the *BRCA* testing) or MD clinical geneticist. Among a subset of participants who agreed to

complete additional questions collected through a questionnaire (either paper-based or online), information to assess uptake of various NCCN-recommended CRM modalities (ever versus never) was available(5).

Statistical analysis

Participant characteristics were summarized using descriptive statistics, including means and proportions for continuous variables and frequencies and proportions for categorical variables. Contingency tables were tabulated and chi-square tests were conducted to examine the association of demographic and clinical variables with delivery of genetic testing services. The association of demographic, clinical, and service delivery factors with CRM strategies (categorized as 'ever' versus 'never') was done by calculating the proportions of *BRCA* carriers who reported the uptake of these strategies. Various subset analyses were performed, defined by demographic, clinical and service delivery factors. All statistical tests were performed using statistical software R version 2.15.1.

RESULTS

Of the 795 participants who completed the baseline survey, 438 had collection of additional information through a questionnaire about CRM practices. Clinical and demographic characteristics for the overall group and for those in the subset in whom CRM practices were available were similar (Table 1).

On average, uptake of CRM strategies was assessed four years after the date of genetic testing. The mean age of participants was 48 years, the majority was married/cohabitating and college graduates (59.1%) and approximately one-half had a personal history of cancer. We explored variables associated with delivery of genetic testing services (Table 2), which showed that duration of GC session, was longer when conducted by a genetics professional than when it was conducted by another specialist without involvement of a genetics professional ($p<0.001$).

In the 438 women in whom information on uptake of CRM strategies was available, we observed higher frequencies in the uptake of most CRM strategies with GC sessions ≥30 minutes compared to shorter sessions (Table 3). A similar trend was observed based on who ordered the *BRCA* test and who conducted the pre-test GC session, with higher frequencies in the uptake of most CRM strategies when a genetics professional was involved. In fact, the difference in uptake of MRI was significantly higher when *BRCA* testing was ordered by a genetics professional ($p=0.05$) and when the pre-test GC session was conducted by a genetics professional ($p=0.026$). Finally, there was a trend towards greater uptake of breast MRI, bilateral mastectomy and tamoxifen among women with a family history of breast cancer compared to those with no family history, but the difference did not reach statistical significance. Similarly, the uptake of ovarian screening was higher in women with a family history of ovarian cancer compared to no family history for both serum CA-125 (67.5% versus 53.0%, respectively, $p=0.007$) and trans-vaginal ultrasound (64.9% versus 47.0%, respectively, $p=0.001$).

DISCUSSION

We assessed the delivery of CGRA services and its impact on CRM practices in a sample female *BRCA* mutation carriers recruited across the United States. Results suggest that: 1) genetics professionals spend a longer amount of time to deliver genetics services compared to other providers ($p<0.001$); 2) high uptake of CRM strategies across all providers within our sample; and 3) higher frequencies in the uptake of most CRM strategies with longer GC sessions and with the involvement of a genetics professional.

Over the last several years, guidelines for the delivery of CGRA services (6, 7) and for the CRM of women with a *BRCA* mutations (5) have been published. CGRA conventionally begins with a 60–90 minute in-person pretest GC session for risk assessment(11). Patients who proceed with testing return for in-person disclosure of test results and detailed discussion of CRM strategies. If a woman is identified as a *BRCA* mutation carrier, the primary objective is to minimize her high lifetime risks of breast and ovarian cancer through uptake of available CRM strategies(3, 4) To that end, the National Comprehensive Cancer Network (NCCN) CRM guidelines for *BRCA* carriers include the following modalities: mammograms and breast magnetic resonance imaging (MRI) for breast cancer screening; consideration of serum CA-125 level and transvaginal ultrasounds for ovarian cancer screening; bilateral prophylactic mastectomy and salpingo-oophorectomy as surgical risk reduction strategies; and tamoxifen as a medical breast cancer risk reduction strategy(5).

Several professional organizations recommend involvement of genetics professionals when genetic testing services are delivered(6, 7). Components of CGRA services include generating a 3-generation pedigree, providing detailed risk assessment with differential diagnosis, discussing potential test results and CRM options for hereditary cancer, and obtaining written informed consent prior to testing, which require large amounts of provider time (6, 7). Thus our observed association of longer GC sessions when a genetics professional was involved it is not surprising. The implications of this observation warrants evaluation, given that CGRA services are increasingly delivered in the community setting by a diverse group of providers who have not had formal training in genetics(11).

Despite these findings, one of the main questions that still remains is whether the longer GC session translates to higher uptake of CRM strategies, used as a surrogate to measure patient benefit. In fact, we observed higher frequencies of uptake of most CRM strategies when genetics professionals were involved in ordering *BRCA* testing and conducting pre-test GC sessions. Furthermore, we observed higher frequencies of uptake of most CRM strategies in those with longer GC sessions, suggesting that the time intensity may potentially translate into patient benefit itself. Alternatively, it is also possible that highly motivated patients seek out medical experts (such as genetics providers) and naturally spend more time asking questions. Thus, there remains a need for confirmatory studies to definitively address this question.

Determining whether the traditional model for delivering CGRA services results in maximizing benefit to *BRCA* mutation carriers is important to inform policy level decisions. Furthermore, other measures of patient benefit with the involvement of genetics

professionals include psychological and family-centered benefits which would also be important to evaluate through future efforts. If current standards for delivering CGRA services do not translate to patient benefit, the existing guidelines for GC should be re-addressed. In fact, this issue is especially important to address as more community-based providers identify, test, and manage high risk individuals within the United States(11).

In the end, there has been a lack of prior patient-based efforts to evaluate how and by whom they receive *BRCA* testing services, and its impact on patient benefit. Furthermore, most studies of *BRCA* carrier populations in the United States have primarily included patients who received GC through genetics professionals, mainly at academic centers. This is largely due to the fact that most data collection efforts occur at academic centers as part of research initiatives, thus many *BRCA* carriers tested in the community setting may not be aware of or provided with information about research opportunities. Thus it is necessary to recruit *BRCA* carriers from diverse settings to better understand how the delivery of genetic testing services may impact the uptake of CRM strategies, as was done through the current study.

As for provider-based efforts, a few surveys in diverse groups of physicians who offer *BRCA* testing have assessed CRM strategies made to *BRCA* carriers(9, 10). In fact, results suggest general adherence to NCCN surveillance guidelines despite lack proficiency in genetics knowledge (9, 10, 12). In contrast, negative outcomes reported through case reports when genetic testing occurs without adequate GC include misinterpretation of test results and inappropriate cancer screening/prevention recommendations(13). These observations require further systematic study in large unselected samples of *BRCA* carriers from diverse settings to enhance their generalizability. To that end, through peer-reviewed funding secured for ICARE in 2010, we developed an academic-community partnership through which we provide educational resources focused on CGRA to practitioners from diverse settings, as well a research link for their patients through enrollment in our registry.

Our study has several strengths, including the large sample size and the recruitment of participants diagnosed and treated by a diverse group of providers across the United States. Despite these strengths, we had limited power to detect differences in delivery of CGRA services by provider type due to the large proportion study participants who were counseled through genetics professionals, thus could not run multivariable models.. Additionally, our study population encompassed a well educated and affluent population, which limits the generalizability of our results. Furthermore, uptake was self-reported, without medical record confirmation. Moreover, our cross-sectional survey only measured adherence to CRM modality without assessment of recommended intervals and trends over time. Finally, length of GC sessions were collected, without assessment of the components collected and discussed during the session, which is required to evaluate quality of GC services.

Ultimately, findings from our study indicated longer GC sessions when provided by genetics professionals, but overall high uptake of NCCN-recommended CRM strategies across all providers. Furthermore, there were higher frequencies of uptake of most CRM strategies for those with longer pre- and post-test GC sessions and in those who indicated that a genetics professional was involved in their *BRCA* GC and testing. Future efforts are needed to assess conduct of CGRA services in diverse settings by various providers.

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Table 1

Demographic and Clinical Variables of Study Population

	Completion of baseline demographic and service delivery questions (n=795)	Completion of additional questions about medical management (n=438)
Age		
Mean (SD)	47.7 (11.0)	48.3 (11.4)
<50	419 (52.7)	238 (54.3)
≥50	328 (41.3)	200 (45.7)
Missing	48 (6)	0 (0.0)
Marital Status		
Married/Cohabitating	600 (75.7)	327 (74.7)
Other	189 (23.8)	110 (25.1)
Prefer not to answer	4 (0.5)	1 (0.2)
Missing	2 (0.3)	0 (0.0)
Education		
College Graduate	602 (75.7)	323 (73.7)
<College Graduate	187 (23.5)	112 (25.6)
Missing	6 (0.8)	3 (0.7)
Annual Income		
≥$50,000	537 (67.5)	297 (67.8)
<$50,000	140 (17.6)	91 (20.8)
Prefer not to answer	99 (12.5)	41 (9.4)
Missing	19 (2.4)	9 (2.1)
Personal Cancer History		
Breast Cancer	320 (40.3)	189 (43.2)
Ovarian Cancer	64 (8.1)	38 (8.7)
Both Breast and Ovarian Cancer	19 (2.4)	11 (2.5)
No Breast or Ovarian Cancer	430 (54.1)	222 (50.7)
Family history (#br/ov cancer)		
≤2	347 (43.6)	186 (42.5)
3	156 (19.6)	90 (20.5)
≥4	261 (32.8)	153 (34.9)
Missing	31 (3.9)	9 (2.1)

Table 2

Association of Demographic and Clinical Variables in the delivery of genetic testing services

	Pre-test discussion (n(%))			Post-test discussion(n(%))			Results notification(n(%))		
	<30 minutes	**≥30 minutes**	**P-value**	**<30 minutes**	**≥30 minutes**	**P-value**	**In person**	**Other**	**P-value**
Age									
<30	16 (42.1)	22 (57.9)		22 (64.7)	12 (35.3)		20 (58.8)	14 (41.2)	
30–49	180 (47.2)	201 (52.8)	0.087	199 (59.6)	135 (40.4)	0.112	207 (61.6)	129 (38.4)	0.940
≥50	128 (39.0)	200 (61.0)		158 (52.3)	144 (47.7)		195 (61.9)	120 (38.1)	
Marital Status									
Married/Cohabitating	258 (43.0)	342 (57.0)	0.993	304 (55.9)	240 (44.1)	0.544	337 (60.7)	218 (39.3)	0.344
Other	82 (43.4)	107 (56.6)		99 (58.9)	69 (41.1)		112 (65.1)	60 (34.9)	
Education									
College Graduate	254 (42.2)	348 (57.8)	0.473	315 (57.3)	235 (42.7)	0.753	336 (60.1)	223 (39.9)	0.299
<College Graduate	86 (45.5)	103 (54.5)		91 (55.5)	73 (44.5)		111 (64.9)	60 (35.1)	
Annual Income									
≥$50,000	229 (42.6)	308 (57.4)	0.235	268 (56.4)	207 (43.6)	0.545	299 (61.5)	187 (38.5)	0.531
<$50,000	70 (48.6)	74 (51.4)		79 (59.8)	53 (40.2)		79 (58.1)	57 (41.9)	
Family cancer history[1]									
≤2	117 (44.3)	147 (55.7)	0.531	134 (56.8)	102 (43.2)	>.999	140 (58.6)	99 (41.4)	0.336
>2	213 (41.7)	298 (58.3)		262 (56.6)	201 (43.4)		298 (62.6)	178 (37.4)	
Children									
No	38 (35.5)	69 (64.5)	0.251	50 (52.6)	45 (47.4)	0.525	67 (67.0)	33 (33.0)	0.809
Yes	120 (42.6)	162 (57.4)		157 (57.1)	118 (42.9)		180 (65.0)	97 (35.0)	
Genetics professional involved[2]									
no	153 (87.9)	21 (12.1)	<0.001	133 (83.1)	27 (16.9)	<0.001	75 (45.7)	89 (54.3)	<0.001
yes	133 (23.5)	432 (76.5)		274 (49.4)	281 (50.6)		375 (65.9)	194 (34.1)	

[1] includes breast or ovarian cancer in participant;

[2] defined as genetic counselor or geneticist