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Video-assisted genetic counseling in patients with ovarian, fallopian and peritoneal carcinoma

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Abstract

Objectives.—To compare the proportion of patients with ovarian, fallopian or peritoneal carcinoma who receive genetic testing after observing a genetic counseling video versus after traditional referral for genetic counseling and testing at physician discretion.

Methods.—A retrospective chart review was performed of all patients seen at the West Cancer Center for evaluation of ovarian, fallopian or peritoneal carcinoma from 7/2014 to 8/2015. Patients seen between 7/2014 and 12/2014 were offered standard genetic counseling. We adopted a new standard of care from 3/2015 to 8/2015 involving the use of a genetic counseling video on a digital tablet. The video was shown to patients with ovarian, fallopian or peritoneal cancer, who were then given the option to undergo genetic testing at the end of the viewing. We compared the number and proportion of patients who received genetic testing in both groups.

Results.—The initial group of 267 patients received referral and te\sting at the physician's discretion between 8/2014 and 12/2014. 77/267 (29%) of these patients underwent genetic testing. 295 patients viewed the condensed genetic counseling video with the option to receive testing the same day between 3/2015 and 8/2015. 162/295 (55%) of these patients received testing. The transition from a referral method to the video counseling method resulted in a significant increase of patients tested ($p < 0.001$).

Conclusion.—Using a genetic counseling video and providing an immediate option for testing significantly increased the proportion of patients with ovarian, fallopian or peritoneal carcinoma who received genetic testing.

Keywords

Ovarian cancer; Genetic testing; Hereditary cancer syndromes

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Conflict of interest statement

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1. Introduction

The process of genetic counseling and testing in ovarian cancer patients is becoming an increasingly crucial aspect of patient care in this population [1,2]. Ovarian cancer claims the highest mortality rate of all gynecologic cancers and has the strongest connection to hereditary syndromes, particularly BRCA1/2 [2]. The finding of a pathogenic variant of BRCA1/2 in a patient is important, not just for the benefit of family members and the prevention of future disease, but for the approach to the patient's treatment: The advent of poly(ADP-ribose) polymerase (PARP) inhibitors marked the beginning of the likely exponential growth of molecular directed therapies in cancer treatment. [3].

NCCN, ASCO and SGO guidelines now recommend BRCA1/2 testing in all patients with a diagnosis of epithelial ovarian, peritoneal or fallopian carcinoma [4–6]. National practice falls significantly short of this, with physician referral often depending on family history and age at diagnosis; cited national testing rates range from 14 to 25% [2,7]. Perhaps more importantly, as many as 16% of mutation carriers would not have been identified by family history [3].

Although universal testing is now the standard of care, the process of counseling and testing needs to be improved upon, especially in areas of high patient volume or with limited access to genetic counselors [2,7]. While attempts have been made at new service delivery models, there is still no definitive or widely used algorithm for genetic counseling education [8]. We sought to establish an integrated model of concise education with immediate access to testing by utilizing a condensed genetic counseling video. We then compared the proportion of patients with ovarian, fallopian or peritoneal carcinoma who received testing using this method versus using the traditional referral method.

2. Methods

A retrospective chart review was performed of all patients seen at the West Cancer Center in Memphis, TN for evaluation of ovarian, fallopian or peritoneal carcinoma from 7/2014 to 8/2015. Patients seen between 7/2014 and 12/2014 were offered standard genetic counseling and testing at physician discretion during their initial appointment. The traditional counseling method involved the referral of patients at physician discretion to a certified genetic counselor. This appointment took place at a later time, involving approximately 30–45 min of face-to-face conversation and education, followed by the option of BRCA1/2 testing with reflex to a multi-gene panel.

Physicians had access to a new method of patient education and testing that was adopted from 3/2015 to 8/2015. Patient with ovarian, fallopian or peritoneal cancer seen during this period were instead shown a condensed, standardized counseling video on a tablet during their appointment at the physician's discretion. Those patients whose insurance required traditional genetic counseling prior to testing were not able to receive video counseling. The video was created by a certified genetic counselor at West Cancer Center. It is 7 min long and consists of a discussion of genes and mutations, red flags for hereditary cancer, a review

of BRCA1/2, the possibility of finding other mutations, potential impact on family members, the legal protection of genetic information, and the interpretation of test results.

All patients were electronically given the option to undergo BRCA1/2 testing at the end of the video per NCCN guidelines; they were also given the option to reflex to a comprehensive multi-gene panel if BRCA testing was negative. Final results were discussed with all patients. Patients with positive results underwent formal consultation with a genetic counselor. We compared the number and frequency of patients who received genetic testing in both groups. Demographic characteristics, including age at diagnosis, BMI (body mass index), race, disease stage and disease grade, were compared. Statistical package for the social sciences was used to analyze the data; a chi square test was used to compare the discrete variables and a *t*-test was used for continuous variables.

3. Results

A total of 562 patients were included in the study. Demographic details of these patients are provided in Table 1. No demographic characteristics were significantly different between the two groups, including age, race, BMI, disease stage or disease grade. 267 patients received referral and testing at the physician's discretion between 7/2014 and 12/2014. 77/267 (29%) of patients in the traditional referral group ultimately underwent genetic testing (Table 2). 295 patients viewed the condensed genetic counseling video with the option to receive testing the same day between 3/2015 and 8/2015. Among patients who viewed the counseling video and were offered testing the same day as their initial visit, 162/295 (55%) received testing (Table 3). The transition from a referral method to the video counseling method resulted in a significant increase of patients tested ($p \leq 0.001$) with a 95% confidence interval 0.26 ± 0.08 (0.18–0.34). There was not a significant difference in mutation detection rate between the two groups: 21/267 or 7.9% had detected BRCA mutations in the traditional group; 24/295 or 8.1% of patients had detected BRCA mutations in the video counseling group ($p = 0.91$). 5/267 (1.9%) in the traditional group had other detected mutations; 9/295 (3%) had other detected mutations in the video counseling group ($p = 0.37$).

4. Discussion

Our study demonstrates a significant increase in the proportion of patients tested when using the video counseling method versus the traditional referral method. This alternative education and testing algorithm could be a potential tool to adhere to NCCN guidelines concerning genetic testing in this patient population.

Approximately 13–17% of ovarian cancer patients have a germline BRCA mutation [9]. The diagnosis of a BRCA1 mutation carries a 40% lifetime risk of ovarian cancer; that of BRCA2 a 20% lifetime risk [10,11]. Within the last two years, SGO, ASCO and NCCN have all concluded that universal testing of all patients with epithelial ovarian, fallopian or peritoneal carcinoma should be standard of care. This standard, however, has proven difficult to implement. A large percentage of patients are unaware BRCA genetic testing or its applicability to their diagnosis, treatment or family members [12]. A recent article

that reviewed the counseling and testing practices of oncology centers from 22 collaborative groups in 19 different countries found that only 55% routinely offer BRCA testing to all women with ovarian cancer [2]. This disparity is likely secondary to remaining dependence on high-risk paradigms for referral determination [2].

Several models exist for stratifying risk, including the Myriad and Manchester scoring system, but these systems have been found to consistently underestimate the probability of mutation detection [13]. Multiple studies demonstrate that family history alone cannot accurately detect all population mutations [10,14]. Moller et al. found, after testing all women who presented to their clinic with ovarian carcinoma, that only one third of the 23% with a BRCA1/2 mutation would have met testing requirements by their family history [14].

Conversely, the DNA-BONus trial found that when an adequate pedigree was obtained, current testing criteria based on age and family history were actually sufficient in identifying all mutations. Despite this finding, there was a higher than average percent of pathogenic variants in this ovarian cancer patient population (22.3%) leading the authors to conclude that testing should be offered to all patients with epithelial ovarian cancer [15].

The increasing demand for an expansion of BRCA1/2 testing in patients with ovarian, fallopian or peritoneal carcinoma has created a need for a streamlined education and referral process. Lheureux et al., after surveying current BRCA1/2 testing practices across multiple institutions, concludes that “the current BRCA1/2 testing/counseling paradigm may be suboptimal,” and recommends seeking “alternative strategies to provide real time information” [2].

Various alternative strategies have been suggested, including telephone counseling, computer-based decision modules and group counseling. Telephone counseling was found to be equally efficacious in regards to patient education; however, testing rate was lower in the telephone counseled patients, likely because patients who opted for telephone counseling lived in remote areas [16]. Green et al. compared standard counseling to a computer based decision aid for breast cancer patients [17]. Although the computer-based model was more effective at educating patients, testing decisions did not differ significantly between the computer and counselor group. In this study, testing was not offered immediately after use of the program. Group counseling was found to be more efficient in Ridge et al., however, a significant number of patients decline, citing the need for a more private and personalized approach [18].

Perhaps it is the combined intervention of both standardized and efficient initial counseling and the opportunity for immediate testing that makes testing more approachable to patients. Sie et al. found that a significant majority of breast cancer patients preferred having indirect education followed by immediate testing rather than initial face-to-face consultation. Furthermore, this DNA-direct method reduced processing time by one month [19].

The option of reflex to a multi-gene panel was an important aspect of the study. In an article by Minion et al., 6.4% of ovarian cancer patients with negative BRCA1/2 testing had a pathogenic mutation on a multi-gene panel; the most common of these were BRIP1 and MSH6 [1]. Multi-gene panels thus offer a more comprehensive view of a

patient's genetic profile. While this is helpful if the testing reveals actionable mutations, the information may overwhelm or confuse the patient, particularly if it results in a variant of undetermined significance (VUS) [1]. Further studies are needed to better illuminate appropriate recommendations for specific mutations. It is of upmost importance that patients are educated on the meaning of other mutations as well as variations of undetermined significance and that they are given a clear choice between the BRCA1/2 testing alone and with reflex to the multi-gene panel; these topics are all addressed in the video.

Our study examines a novel combination of standardized counseling, immediate testing, and more traditional face-to-face counseling when applicable.

Strengths of our study include a large patient population and simplicity of design. Weaknesses include the retrospective design. Furthermore, the implementation of this new standard of care was not adhered to with 100% compliance, implying that the actual testing rate of patients who viewed the video could be higher than the number cited. The cited testing rate in the traditional counseling portion could also beattributed to multiple factors whichwere not recorded in the medical record, including patients' declination of counseling, failure of the physician to universally refer, failure of the patient to follow-up with the genetic counselor, and declination of testing after counseling. We were not able to retrospectively gather these details. Patients able to receive video counseling only prior to testing were also limited by insurance, as some insurance providers require formal genetic counseling. The use of the video is also restricted to English-speaking patients only. Those patients whose insurance restricted them to standard genetic counseling were thus not able to view the video; they will also have to be excluded from any prospective trial. However, as further evidence regarding the efficacy of alternative counseling strategies continues to emerge, perhaps these policies will also evolve, allowing for other cost and time effective pathways to universal testing.

There was not a significant difference in mutation detection between the two groups. BRCA incidence rates in both groups were lower than cited incidences in the literature, suggesting that there were mutations in patients who were not tested [9]. This finding further emphasizes the importance of universal testing.

We plan to further evaluate this model of service delivery in a randomized prospective trial. Primary outcomes will include time to testing, patient understanding and adherence to national guidelines. Secondary outcomes will include quality of life, cost, and a demographic description of patients who prefer this immediate testing method to initial face-to-face counseling.

The ideal combination of efficient, affordable education with easy access to testing and appropriate formal follow-up has yet to be determined; however, the burgeoning reality of pathogenic genetic variant identification and potential tailored therapeutics makes the establishment of such a method a matter of great importance. The use of a condensed genetic counseling video could bridge the gap between patient and genetic counselor, allowing widespread testing to informed patients and providing, not a replacement of the genetic counseling role, but improved access to appropriate candidates. It is in essence

an integrated model of concise education, immediate access to testing, and appropriate guidance by a genetic counselor to those who need it.

References

[1]. Minion L, Dolinsky J, Chase D, et al. , Hereditary predisposition to ovarian cancer, looking beyond BRCA1/BRCA2, Gynecol. Oncol 137 (2015) 86–92. [PubMed: 25622547]

[2]. Lheureux S, Karakasis K, Harter P, et al. , Germline BRCA1/2 testing practices in ovarian cancer: current state and opportunities for new directions, Gynecol. Oncol 140 (2016) 90–94. [PubMed: 26475959]

[3]. Demsky R, McCuaig J, Maganti M, Murphy KJ, Rosen B, Armel SR, Keeping it simple: genetics referrals for all invasive serous ovarian cancers, Gynecol. Oncol 130 (2012) 329–333.

[4]. National Comprehensive Cancer Network, NCCN Clinical Practice Guidelines in Oncology: Genetic/Familial High-risk Assessment: Breast and Ovarian. V2, 2015.

[5]. Society of Gynecologic Oncology, SGO Clinical Practice Statement; Genetic Testing for Ovarian Cancer, October 2014.

[6]. Lu KH, et al. , "American Society of Clinical Oncology." American Society of Clinical Oncology Expert statement, collection and use of a cancer family history for oncology providers, J. Clin. Oncol 32 (8) (2014) 833–840. [PubMed: 24493721]

[7]. Febbraro T, Robison K, Wilbur J, et al. , Adherence patterns to National Comprehensive Cancer Network (NCCN) guidelines for referral to cancer genetic professionals, Gynecol. Oncol. 138 (2015) 109–114.

[8]. Trepanier A, Allain D, Models of service delivery for cancer genetic risk assessment and counseling, J. Genet. Couns 23 (2014) 239–253. [PubMed: 24158360]

[9]. Antoniou, et al. , Average risks of breast and ovarian cancer associated with BRCA1 and BRCA2 mutations detected in a case series unselected for family history: a combined analysis of 22 studies, Am. J. Hum. Genet 72 (2003) 1117–1130. [PubMed: 12677558]

[10]. Alsop K, Fereday S, Meldrum C, et al. , BRCA mutation frequency and patterns of treatment response in BRCA mutation-positive women with ovarian cancer: a report from the Australian Ovarian Cancer Study Group, J. Clin. Oncol 30 (2011) 2654–2663.

[11]. Weissman S, Weiss S, Newlin A, Genetic testing by cancer site, The Cancer Journal 18 (2012) 320–327. [PubMed: 22846732]

[12]. Lacour, et al. , What women with ovarian cancer think and know about genetic testing, Gynecol. Oncol 111 (2008) 132–136. [PubMed: 18684498]

[13]. George A, UK BRCA mutation testing in patients with ovarian cancer, Br. J. Cancer 113 (2015) S17–S21. [PubMed: 26669451]

[14]. Moller P, Hagen AI, Apold J, Maehle L, Clark N, et al. , Genetic epidemiology of BRCA mutations – family history detects less than 50% of the mutation carriers, Eur. J. Cancer 43 (2007) 1713–1717. [PubMed: 17574839]

[15]. HIldegunn H, Bjorvatn C, Fiane B, Aas T, Woie K, Helge E, et al. , BRCA1/2 testing in newly diagnosed breast and ovarian cancer patients without prior genetic counseling: the DNA-BONus study, Eur. J. Hum. Genet 1–8 (2015).

[16]. Kinney AY, Butler KM, Pollet A, et al. , Expanding access to BRCA ½ genetic counseling with telephone delivery: a cluster, randomized trial, J. Natl. Cancer Inst 106 (12) (2014).

[17]. Green M, Peterson S, Baker M, Harper G, et al. , Effect of a computer-based decision on knowledge, perceptions and intentions about genetic testing for breast cancer susceptibility, JAMA 292 (2014) 442–452.

[18]. Ridge, et al. , Evaluation of group genetic counseling for hereditary breast and ovarian cancer, J. Genet. Couns 18 (2009) 87–100. [PubMed: 19127417]

[19]. Sie AS, van Zelst-Stams WA, Spruijt L, et al. , More breast cancer patients prefer BRCA mutation testing without prior face-to-face genetic counseling, Familial Cancer 13 (2014) 143–151. [PubMed: 24068317]

HIGHLIGHTS

- A new method of genetic counseling for ovarian cancer patients is proposed.
- Patients were shown a genetic counseling video and offered immediate testing.
- A significantly larger proportion of patients were tested using this new algorithm.

Table 1

Demographics.

	Traditional (n = 267)	Integrated (n = 297)
Age	58.9	59.5
Race (%)		
Black	17.8	19
White	79.5	78.8
Other	2.7	1.8
BMI	30.4	29.3
Late stage (%)	62.7	65.1
High grade (%)	75.6	73.1

Table 2

Traditional counseling group.

	Patients seen	Patients tested	Percentage tested
Provider A	97	20	21
Provider B	98	39	40
Provider C	71	18	25
Total	267	77	29

Table 3

Video-counseling group.

	Patients seen	Patients tested	Percentage tested
Provider A	98	47	48
Provider B	115	73	63
Provider C	82	42	51
Total	295	162	55