Getting to the point: What women newly diagnosed with breast cancer want to know about treatment-focused genetic testing

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Abstract

Background. An increasing number of women newly diagnosed with breast cancer with features indicative of an increased risk of carrying a germline mutation in a breast/ovarian cancer gene (BRCA1 or BRCA2) are being offered genetic testing to guide their breast cancer management (treatment-focused genetic testing - TFGT). This qualitative study aimed to identify young women’s information preferences regarding TFGT and, in particular, explore whether it added an overwhelming volume and complexity of information at diagnosis. Based on these preferences, we developed and evaluated a novel educational resource.

Methods. Women (n=26) with breast cancer (<50 yrs) who either (1) previously had TFGT (n=14) or (2) had a recent diagnosis and were asked about their hypothetical views of TFGT (n=12) participated in semi-structured interviews. Following these interviews, a brief pamphlet on TFGT was developed and pilot-tested with 17 of the women.

Results: Most women wanted to be informed about TFGT at or around the time of their cancer diagnosis when treatment decisions are discussed. There was no clear majority preference for which type of health professional should provide information on TFGT. Most women preferred to receive the information in a face-to-face consultation, with brief written information about TFGT viewed as important supporting material. The educational resource developed was well received and modified based on the results obtained from pilot-testing.

Conclusion: The potential for more widespread TFGT in the future indicates a need for patient education materials to enable woman to make informed choices about TFGT. This pilot study has provided timely initial evidence on the efficacy of a brief written resource in preparing women for decision-making about TFGT.
Introduction

Traditionally, genetic testing for a germline mutation in a breast/ovarian cancer gene (such as BRCA1 or BRCA2 genes) in women with a breast cancer diagnosis has been confined to those with a family history of breast and/or ovarian cancer. Testing of such women usually takes place after completion of active cancer treatment, for the purpose of guiding their future risk management in relation to a new primary breast cancer and/or ovarian cancers. In addition, genetic risk information also has significant risk management implications for the woman’s unaffected genetic relatives, who may themselves consider predictive genetic testing to clarify their risk of breast and/or ovarian cancer.

However, there is growing evidence that mutation status may influence breast cancer management recommendations, and that there may be benefits in having genetic counselling and testing available shortly after a cancer diagnosis (Silva, 2008; Trainer et al., 2010; Tutt & Ashworth, 2008). This process, hereafter referred to as treatment-focused genetic testing (TFGT), may help guide a woman’s initial cancer treatment in addition to future risk management. In the first instance, TFGT may assist with the complex decision-making processes regarding a woman’s surgical options, including the selection of breast conservation or a therapeutic mastectomy, with or without a contralateral risk-reducing mastectomy. (Meiser et al., 2008)

Furthermore, evidence is accumulating that BRCA-associated tumours are more sensitive to platinum agents and potentially less resistant to taxanes (Quinn et al., 2007; Rottenberg et al., 2007; Trainer, et al., 2010) In addition, novel agents are currently being tested that target BRCA-tumours; specifically poly (ADP-ribose)
polymerase (PARP) inhibitors have been found to be active agents with high response rates in patients with recurrent disease (Fong et al., 2009; Fong et al., 2010). Thus, as more conclusive evidence on the efficacy of both conventional chemotherapy and novel agents becomes available, TFGT is likely to be increasingly used to tailor women’s cancer treatments.

Rapid advances in sequencing technology are likely to decrease the cost of genetic testing and the time frame within which results can be provided. In addition, the traditional approach of using family history as the major selection criteria for genetic testing is being challenged, as the proportion of BRCA1 and BRCA2 mutation carriers found in early onset breast cancers without a relevant family history ranged between 6% (Lalloo et al., 2003) and 78% (Choi, Lee, Bale, Carter, & Haffty, 2004) in a recent systematic review (Meiser, et al., 2008). As such, there is a pressing need for data on effective educational strategies regarding TFGT for women newly diagnosed with breast cancer, both with and without a relevant family history, in advance of this new technology being implemented widely into clinical practice.

Very little is currently known about the acceptability of TFGT among women with and without a relevant family history of breast and/or ovarian cancer and their associated information needs. Two prospective studies conducted in the US and the Netherlands have assessed the psychological and behavioural impact of TFGT. {Schlich-Bakker, 2006} {Schwartz, 2004} The Dutch study assessed the psychological impact of TFGT in women with breast cancer who were shortly to commence adjuvant radiotherapy. The authors found that distress levels did not increase after the offer of genetic counselling and testing. {Schlich-Bakker, 2006}
A small UK study, using a focus group methodology, explored whether women diagnosed with breast cancer under the age of 40 wanted information about genetic testing close to the time of diagnosis. All 13 participants had already been identified as BRCA carriers (Ardern-Jones, Kenen, & Eeles, 2005). The majority of women felt that an offer of genetic testing around the time of their cancer diagnosis would have been too stressful, although some women reported that this offer would be important if it had the potential to alter treatment decisions. All women agreed that there was that there was no one right time for everyone. An important limitation of these studies is that they included almost exclusively women with a relevant family history.

In a recent qualitative study, Vadaparampil et al. (2009) assessed the impact of a surgeon referral letter on recently diagnosed breast cancer patients’ uptake of BRCA testing. Many women who had been sent a letter by their surgeon for BRCA genetic counselling reported mixed reactions to the letter and some women were confused or concerned about why they had received a letter. About 20% of women did not recall receiving the letter. The authors concluded that a referral letter from the woman’s surgeon may not be the most effective means of informing patients about TFGT. The authors suggested that the letter may have been more effective if it had included more detailed information on the surgical implications of BRCA testing and on the process of TFGT; this suggestion was, however, not based on data regarding women’s preferences with regard to the content and format of the educational materials.

To our knowledge, there are currently no educational resources specifically for women newly diagnosed with breast cancer, that provide information about TFGT to
assist with informed decision-making. Detailed decision aids have been developed for people affected by cancer considering genetic testing after completion of their cancer treatment, and for unaffected people at increased risk of hereditary cancer considering predictive testing. (Gaff & Meiser, 2009; M.J. Green, Biesecker, McInerney, Mauger, & Fost, 2001; M. J. Green et al., 2004; M.J. Green et al., 2005; Mancini et al., 2006; Schwartz et al., 2001; van Roosmalen et al., 2004; Wakefield et al., 2008a; Wakefield et al., 2008b). Decision aids have been shown to be effective in meeting the information needs of these specific populations (Gaff & Meiser, 2009).

However, TFGT among young women with breast cancer is very different to genetic testing offered to patients following completion of their cancer treatment and to predictive testing, where knowledge of mutation status does not influence treatment. It is, therefore, critical to ascertain the specific information needs of patients newly diagnosed with breast cancer regarding TFGT in order to inform the development of appropriate educational materials that will not overwhelm these patients at a highly stressful time. Specifically, data regarding the content, format and mode of delivery of information about TFGT will inform the development of high quality educational materials.

This study was carried out in two stages. First, in-depth interviews were conducted to identify the information and communication needs regarding TFGT, of women newly diagnosed with breast cancer who were aged \leq 50 years. The age cut off was chosen as younger women may be more likely to use mastectomy for future prevention as they have less life-years at risk that would benefit from a mastectomy. The views of young women with and without a relevant family history of breast and/or ovarian cancer were included because both groups are likely to be targeted for TFGT in the
near future. On the basis of the findings from the qualitative study in Stage 1, we then developed and pilot-tested a psycho-educational resource in Stage 2 to enable women newly diagnosed with breast cancer to make informed decisions about genetic testing for germline BRCA mutations and to facilitate discussions with their health professionals. Because the development of the educational resource was guided by women’s preferences identified in indepth interviews about TFGT conducted in Stage 1, it was expected that the materials would be evaluated equally favourably by women who had already undergone TFGT and by those who had not. We predicted, therefore, no differences between the two groups of women in terms of their satisfaction with the resource; in the emotional impact of reading the materials, or in their perceived understanding of TFGT.

**Methods**

**Sample**

Two different groups of women with breast cancer (≤50 yrs) were recruited. Group A was comprised of women, ascertained through two family cancer clinics in Sydney and Melbourne, who had already undergone TFGT to facilitate surgical decisions (Group A denotes actual decision-making about TFGT); all had a strong family history of breast/ovarian cancer according to national guidelines (National Breast Cancer Centre, 2006). Group H comprised women who were unselected for family history and diagnosed with breast cancer within the previous 6-12 months at an oncology clinic in Sydney (Group H denotes hypothetical decision-making about TFGT). Exclusion criteria included having had a breast cancer recurrence to avoid undue participant burden; being under 18 years of age; having insufficient English language skills to complete the interview unaided, or having obvious intellectual or
mental impairment For both groups, a letter of invitation to participate in the study interview was sent by the treating clinician. The study recruitment process is presented in Figure 1. Forty-seven letters of invitation were mailed, with 34 women opting into the study (response rate of 72%). Of these women, five were ineligible and one woman could not be contacted for an interview. The data for two of the 28 women interviewed were excluded because they had genetic counselling after their definitive breast treatment. Approval was obtained from the relevant Human Research Ethics Committees.

Data collection

Stage 1: Qualitative analyses of the information and communication needs of women diagnosed with breast cancer

Prior to the telephone interview, women were mailed a consent form, a purposively developed one-page information sheet regarding TFGT and a decision aid about another topic (as an example of one type of educational material) to elicit preferences for specific information presentation. The sample decision aid included a personal worksheet designed to elicit the perceived pros and cons of particular management options. (O'Connor et al., 1998)

A qualitative data collection method was used to identify the range of preferences about information provision (Denzin & Lincoln, 1994). The interviews were semi-structured with probes to elicit more information, as appropriate. Questions explored the following issues: women’s attitudes and preferences with regard to timing, mode of delivery and format of information regarding TFGT. Results from early interviews were used to suggest additional lines of questioning in subsequent interviews to
ensure that divergent points of view were explored (Miles & Huberman, 1994). All interviews were conducted by MG, who has extensive experience both as a cancer genetic counsellor and an oncology nurse. Sampling was discontinued at the point when data saturation was reached (Denzin & Lincoln, 1994).

Data analysis

The conceptual framework of Miles and Huberman (1994) was used to guide the analysis. MG and KW identified the initial themes and categories and coded two transcripts concurrently to further refine themes and categories; if discrepancies occurred with respect to specific categories, discussions took place until consensus was achieved. KW then coded the transcripts using the qualitative data analysis software QSR NVivo 8.0 to categorise the data and to facilitate systematic comparisons based on participant characteristics, including participant group (Group A versus Group H), and whether or not a woman had children. EZ conducted all data analyses, using QSR NVivo 8.0 and wrote the descriptive text on the findings. The use of multiple coders and analysts are strategies recommended by Miles and Huberman (1994) to reduce the potential for researcher bias and to increase the validity of the findings.

Stage 2: Development and pilot evaluation of the pamphlet

Procedure

The one-page bi-fold pamphlet developed was designed to provide basic information about TFGT, and to facilitate women’s discussions with their health professional(s) about TFGT. It provided information about TFGT including why women may wish to
consider it, what it involves, and the potential consequences and implications of TFGT results. The early prototypes were developed iteratively involving a multidisciplinary committee, including researchers and clinicians with expertise in clinical genetics, genetic counselling, genetics education, oncology, and psychology. Readability level of the pamphlet was adjusted to 9th grade (National Health & Medical Research Council: NHMRC, 1999). Women who participated in Stage 1 were invited to participate in Stage 2 interviews, which evaluated the acceptability and impact of the pamphlet. Women who wished to be involved in Stage 2 were interviewed by telephone by MP, who was not involved in the development of the materials.

Measures

The following items were included in the Stage 2 interview; they were based on similar items used in previous related studies (Peate et al., 2009; Wakefield et al., 2007)

Satisfaction with the pamphlet: Fourteen items (shown in Table 2) evaluated the amount of information provided, perceived usefulness, and satisfaction with the pamphlet in a combination of structured categorical and open-ended responses. Women were also asked to identify areas that required more or less detail, and to specify what they liked best and least about the pamphlet.

Perceived improvement in understanding: Nine items (shown in Table 3) assessed the perceived extent to which the pamphlet would have improved women’s understanding of TFGT.
Emotional impact of the pamphlet: Participants were asked to rank how much the pamphlet had made them feel worried or concerned, or upset or sad (2 items).

Importance of TFGT: Importance of TFGT was determined using two three-point rating scales, asking participants to indicate how important they felt TFGT would have been at the time of diagnosis, their perceptions of the importance attributed to TFGT by their clinician, and how helpful the pamphlet would have been in reaching their decision about whether or not to have TFGT.

Results

Stage 1: Qualitative analyses of the information and communication needs of women diagnosed with breast cancer

Women (n=26) with breast cancer (≤50 yrs) who either (1) previously had TFGT (Group A, n=14) or (2) had a recent diagnosis and were asked about their hypothetical views of TFGT (Group H, n=12) participated in semi-structured interviews. Twenty-six interviews were transcribed verbatim and analysed. Table 1 shows women’s sociodemographic, medical and family history characteristics. The mean age of participants was 42 years (SD = 5 years), with the mean age of cancer diagnosis being 41 years (SD = 5 years). Over 70% of participants each were married, or co-habiting, had a post-school qualification, and had a family history of breast and/or ovarian cancer. Just under half (46%) had no children, and just over half (54%) did not have a daughter. A Chi-square test for independence indicated that the proportion of women with a relevant family history of breast/ovarian cancer was significantly higher in Group A (100%) compared to Group H (42%), \( \chi^2 (1, n = 26) = 8.41, p = .004, \phi = - \)
.66. There were no differences between the groups in parity or in the presence or absence of daughters. Two participants reported a previous cancer diagnosis, and only two were known to carry a BRCA mutation. Hereafter, participants will be denoted by their group (A or H), and C will denote participants with children, and NC participants with no children, followed by their identification number.

When should TFGT be presented and by whom?

Overall, many participants (n = 16) felt TFGT should be offered before decisions on cancer management, including surgery, were made, in order to get “everything over in one go.” (052). The majority of participants (n = 14), preferred TFGT to be discussed close to diagnosis and in the context of treatment decisions being discussed.

“I think not in the initial diagnosis, because that’s such a shock in itself. The best time for me was when it came to discussing my treatment…but when it was talking about the whole theme of what my treatment would look like, in amongst that...” (A,C,047)

A number of women (n = 8), however, preferred TFGT to be presented at the time of diagnosis, whilst acknowledging that this time was fraught and emotionally overwhelming. As expressed by one woman:
“I think it has to come at the start as part of being diagnosed and then everything explained. When you’re diagnosed you are learning all this new language so I suppose this then has to come into play as well. *Because you’re asking what caused it – how did I get it?*” (A,NC,045)

There was no clear majority preference for which health professional should introduce TFGT. Eight women expressed a preference for the surgeon as the best person to initiate a discussion about TFGT, as the person who would perform the surgery. Six participants preferred to have the discussion with their oncologist, and six with a genetics practitioner (clinical geneticist or genetic counsellor). Five preferred to involve the breast care nurse, as “someone who’s consistent through your care” (047), and one woman did not specify a preference for a particular type of health professional. Of the fourteen women who preferred to have TFGT raised by their oncologist or surgeon, eight wanted to be able to discuss the test result with a genetic health professional when it became available, and two wanted to discuss the implications with the breast care nurse.

“Because you’re working in a very small timeframe, it’s going to be someone like the surgeon who brings it up, but if you have a way of having either the breast care nurse or genetic counsellor on hand so that …if you’ve got more questions, these people can answer your questions, sit down, talk with you more and help with decision-making.” (H,NC,073)

*How should TFGT be presented?*

The vast majority of women (n = 22) preferred to receive information about the availability of TFGT during a face-to-face consultation. This enabled them to ask questions spontaneously, as one woman expressed:
“I found it valuable that I had the opportunity to ask questions. It was a fairly emotional time because I’d just been diagnosed but then having some literature to read to follow-up afterwards was also valuable.” (A,C,048)

A face-to-face consultation also provided the opportunity for a more personalised approach, including the option of having a support person present. It also allowed the patient to determine the level of information they felt they could assimilate.

“It’s funny because I was given all the DVDs and the website, and I have never gone to any one of them. Even though what I do is research, so usually I would do exactly that, I was actually afraid of all those things because I didn’t want to find information that I didn’t want to know. So by asking questions I had at least a feeling of control over it.” (A,C,042)

Additional written supporting information to accompany an offer of TFGT was seen as essential by most participants (n = 16), allowing them to take information home, absorb it, and formulate questions at their own pace.

“I found the whole process was overwhelming and it was useful to take things away, get home, have a cup of tea, read over it, read over it again, and have it in writing in front of me. (A,C,072)
Nine women felt a web-site would be a useful supplementary tool, and eight felt they would have benefited from watching a DVD or other audio-visual material.

What information should be presented?

Whilst all participants agreed that they would like all the implications of TFGT relating to themselves covered, several participants expressed feeling overwhelmed by information around the time of their diagnosis, and the majority (n = 21) preferred information to be brief and “to the point” (A,C,054). Eleven women preferred a leaflet, and another 11 felt a decision aid could be helpful; however only five participants reported that they would use a personal worksheet to help them arrive at a decision on whether or not to have TFGT.

The example information sheet provided to participants was found to provide an acceptable level of information for 15 women. Those that did not find it acceptable (n = 5) expressed a desire to discuss the information face-to-face with a health professional instead of receiving educational materials. The remaining four participants did not express a viewpoint on the example provided, and six further women did not provide a response for this question.

Several participants listed specific topic areas they would like covered in the educational materials, including: the purpose of TFGT (H,C,070), the chance of carrying a gene mutation (H,NC,034; A,C,053), other factors that may elevate a woman’s breast cancer risk above that of population risk (A,C,053), the impact of the test result (A,C,061), particularly on treatment (H,NC,034), what the blood test involves (H,C,070), how long it takes for the results to be returned (A,C,061), that
family members would need to make up their own minds about having predictive genetic testing (H,NC,015) and that family members may have unexpected emotional reactions to the news (A,C,017), such as anger and hostility.

**Implications for themselves**

Women (n = 9) emphasised their desire for the information provided to focus on the impact of TFGT on themselves, the meaning of the test result, and, in particular, the impact of TFGT on their treatment options. Many participants (n = 16) expressed a desire for all available information up front, including information on the chances of developing a second breast cancer (n = 6) and/or ovarian cancer (n = 13).

“For me, I wanted to know everything. I did not know the increased risk of ovarian cancer associated with BRCA1 and 2. ….that was very valuable to know that, because again it would have made me make decisions about prophylactic oophorectomy.” (A,C,048)

There was concern, however, that listing the increased risks of other cancers may further frighten women at an already vulnerable time, and in some instances, may dissuade them from participating in TFGT. As a result, some participants believed that the positive outcomes listed in the description of such a test should be more clearly expressed.

While the majority (n = 16) preferred all clinically relevant information to be presented briefly, some (n = 7) preferred detailed information on surgical options to
be discussed upfront. Conversely, a few (n = 3) preferred the details about surgical options to be discussed only if a positive test result was received.

“It’s unnecessary worry to think, you know, if you read all that and you think. ‘Oh my god is that what I’m going to have to go through?’”

(A,C,054)

Implications for the family

Many women (n = 18) felt that the brochure should “remain focused on the woman” (A,NC,052), and family implications should only be mentioned briefly, with more detail provided in face-to-face genetic counselling should they receive a positive genetic test result.

“I think it’s probably worth mentioning in there. If you have this gene change other members of your family may also have inherited this. You don’t need to go into full scale statistics.” (H,NC,073)

Stage 2: Development and pilot evaluation of the pamphlet

Letters of invitation were sent to 23 of the 26 of the women who participated in Stage 1. Three participants from Stage 1 were not approached (two women declined approach for pilot-testing at Stage 1 and another woman was diagnosed with metastatic breast cancer after her Stage 1 interview). Eighteen women opted into Stage 2 (response rate of 78%), with one women unable to be scheduled for interview. Seventeen women, 10 from Group A and 7 from Group H, participated in Stage 2 interviews.
Chi-square tests for independence were conducted and there were no differences between women in Group A and in Group H regarding their satisfaction with the information in the pamphlet $\chi^2 (1, n = 17)$, perceived improvement in understanding, emotional impact of the pamphlet, or in perceived importance of TFGT. The findings, therefore, are reported for the sample as a whole rather than for each group.

**Satisfaction with the pamphlet**

Almost all women ($n = 16$) reported being ‘satisfied’ or ‘very satisfied’ with the information provided or reported that the amount of information in the pamphlet was ‘about right’ ($n = 15$). Most women ($n = 14$) reported that the pamphlet would have been ‘very useful’ around the time of diagnosis, with three stating that it would have been ‘somewhat useful’.

More than half of participants ($n = 10$) mentioned one or more parts of the pamphlet where more detail was needed: the implications of TFGT ($n = 6$), surgical options ($n = 3$) and more specific information about timing of TFGT ($n = 3$). Women reported that what they liked best about the pamphlet was the format, including the question and answer style ($n = 3$); the layout and order of topics ($n = 2$), and highlighting of important points ($n = 1$). Aspects of the pamphlet that women liked least also related to presentation, and included the cover photograph ($n = 4$), the dated format and the photograph on page 3 ($n = 3$). The section of the pamphlet that was identified most frequently as confusing was ‘what if a gene fault is not found/is found in me?’ ($n = 3$). Suggestions for improving the pamphlet included clarifying the timing of genetic testing ($n = 2$); providing a contact number or the location of a genetic counsellor ($n = 18$).
2); clarifying the section ‘NOT finding a faulty gene’ (n = 1), removing the ‘TFGT’ acronym (n = 1), and modernising the layout (n = 1).

Perceived improvement in understanding

Most participants (n=15) reported that the booklet had improved their understanding. The mean improvement in understanding for each item is shown in Table 3. The largest perceived improvement in understanding was in relation to the purpose of TFGT. The smallest perceived improvement in understanding was in relation to the disadvantages of TFGT in the woman’s situation.

Emotional impact of the pamphlet

Most participants (n = 13) reported feeling ‘not at all’ worried or concerned by reading the pamphlet, while some (n = 4) reported feeling ‘a little’ or ‘somewhat’ worried or concerned. Of the four who were worried after reading the pamphlet, three reported in the open-ended question that it reminded them about the time of their breast cancer diagnosis (n=4) and one woman raised concerns about her relatives (n = 1). Almost all participants (n = 15) reported that reading the pamphlet did not make them feel at all ‘sad or upset’.

Perceived importance of information provision about TFGT

Fifteen participants reported that TFGT was ‘very important’ for women in their situation. Eleven women perceived that their clinicians believed that TFGT was ‘very
important’, and five women that it was ‘somewhat’ or ‘not at all important’. Seven participants reported that the pamphlet would have been enough to make a decision about TFGT soon after diagnosis. The participants who reported that the pamphlet would not have been enough (n=10), said that they would in addition have needed to speak to someone about TFGT (n=10), with six indicating they would want to speak to a health professional or search online for more information, and four not indicating a preference.

Discussion

All women who participated in this study agreed that TFGT should be offered before final decisions on cancer treatment options were made. Just over half of the women preferred TFGT to be discussed after diagnosis, at a time when treatment decisions are being discussed; and another third preferred TFGT to be presented at the time of diagnosis. These findings contrast with Arden-Jones et al.’s interviews of 13 women diagnosed with breast cancer under the age of 40, ascertained through a familial cancer clinic and subsequently identified as BRCA carriers (Ardern-Jones, et al., 2005). While a wide range of views regarding the preferred timing of an offer of TFGT was identified, the majority of women in Arden-Jones et al.’s study expressed the view that an offer of genetic testing around the time of diagnosis might add too much stress at an already stressful time.

There are several factors that may account for the differences in findings. First, the women in the study by Ardern-Jones et al. had undergone genetic testing between two months and 10 years after their diagnosis, and thus most women had completed their cancer treatment at the time of genetic testing. Consequently, all of these women were
providing their views on hypothetical TFGT. By contrast, more than half of the women in the current study had actually undergone TFGT in order to inform their surgical and/or radiotherapy decisions. It is possible that women who are reflecting on their actual rather than hypothetical experience of TFGT perceive tangible benefits from that process, and this may make it more likely for them to express a preference for TFGT at diagnosis or shortly diagnosis, when treatment options are being discussed. Second, the women in Ardern-Jones et al.’s study were interviewed between one and seven years following their diagnosis, and it is possible that women’s attitudes to TFGT may change over time, compared to our study where about half of all women had been diagnosed within the previous year and were thus closer to the time of decision-making. Finally, all the women included in Ardern-Jones et al.’s study had a relevant family history that suggested presence of a hereditary breast/ovarian cancer. The present study, however, included a substantial proportion of women with no relevant family history, who may be less likely to anticipate being a carrier of a BRCA mutation and the associated emotional impact, which in turn may account for preference to have TFGT at the time of diagnosis.

While additional written information was seen as essential by most participants, the vast majority of women preferred to receive information about TFGT during a face-to-face consultation with a health professional. These findings concur with those of Vadaparampil et al. (2009), who concluded that a referral letter alone from the woman’s surgeon may not be the most effective means of informing patients about TFGT. Our finding is not surprising, however, given that research into patients’ preferred communication strategies consistently show that patients prefer to receive
health-related information as part of an individual consultation with an expert (Andrews et al., 2006; Meiser, Mitchell, McGirr, Van Herten, & Schofield, 2005; Thewes et al., 2005). Very little is currently known about whether an individual consultation with an expert is always needed to achieve informed patient choices. For example, a previous randomised controlled trial involving women at low risk of being BRCA mutation carriers found that a computer program was more effective in improving knowledge of breast cancer and genetic testing, facilitating more accurate risk perceptions, and lowering anxiety compared to standard counselling (M. J. Green, et al., 2004). Given the likely increasing burden on familial cancer services and rising costs of health care, future prospective studies are required to determine the most effective ways of offering information about TFGT.

There was no clear majority preference for which type of health professional should provide information on TFGT. Two previous studies have produced divergent results, with patients interviewed in Ardern-Jones’ (2005) study reporting a preference for receiving information about genetic testing to come from a genetics practitioner. These patients had all attended a familial cancer clinic where they saw a genetics practitioner. In a prospective study, the majority of breast cancer patients who were offered genetic testing at the start of their radiotherapy preferred their surgeon to present the genetic information (K. Schlich-Bakker, ten Kroode, Warlam-Rodenhuis, van den Bout, & Ausems, 2007). In the current study, the majority also preferred all clinically relevant information to be presented briefly, with some preferring details on surgical options to be discussed upfront. Taken together, these findings suggests that the type of health professional is not critical as long as he or she is in a position to
present the clinically relevant information to the woman, answer her questions and
gauge the level of detail she feels she can assimilate.

This study also provided detailed information about the preferred content and level of
detail of any educational materials women wished to receive. In particular, the
majority of women preferred the information to be brief. Women emphasised their
need for information on the impact of TFGT for themselves, in particular in regards to
their treatment options, while family implications should be addressed later in face-to-
face genetic counselling should the woman prove to be a gene mutation carrier.
When asked to review the educational materials, women reported that they would
have been useful when making TFGT choices; it was perceived that the materials
would have improved their understanding of TFGT, the vast majority did not think the
pamphlet had a negative emotional impact, and it was generally well received. There
were no differences between the two groups in their satisfaction with the materials,
nor in their perceived understanding of TFGT. This is not surprising given that all
women had participated in in-depth qualitative interviews regarding TFGT in Stage 1;
they were required to read a one-page information sheet about TFGT prior to
interview, and women’s information preferences closely guided the development of
the materials.

While this pilot study has provided timely preliminary evidence on the value of a brief
written resource in preparing women for decision-making about TFGT, it is not
possible to infer whether the educational materials alone would be as effective as
standard pre-test genetic counselling. Given the likely increasing burden on familial
cancer services and rising costs of health care, future prospective studies among larger
samples are required to determine the most effective ways of offering information about genetic testing to patients around the time of their breast cancer diagnosis. Suggested improvements to the pamphlet were incorporated after pilot-testing, including modernising the presentation and photographs, removing the ‘TFGT’ acronym, and clarifying confusing content. The new resource is currently being utilised in a randomised controlled trial assessing the efficacy of brief educational materials (intervention) compared to standard pre-test genetic counselling (control) among women newly diagnosed with breast cancer considering genetic testing on a variety of psychological, behavioural and decision-related outcomes.

There are limitations of this study that need to be acknowledged. First, this was a retrospective study in which women were asked to reflect upon their actual or hypothetical attitudes toward TFGT. Second, the current sample of women was highly motivated and well-educated. Third, the sample size was relatively small although acceptable for a qualitative inquiry. On the other hand, a key strength of this study was that the views of women with and without a relevant family history of breast and/or ovarian cancer were included. The current sample is likely to represent more fully the range of patients at elevated risk of carrying a BRCA mutation who will be targeted by TFGT in the near future, as high-risk features (e.g. breast cancer pathology) in addition to family history are incorporated into genetic testing criteria.

Conclusions and Implications for Nursing

In the near future, optimal management of young women with breast cancer with high risk features will require that genetic risk information is available upfront at diagnosis and prior to treatment so that it may be used to inform surgical and other treatment decisions. Health care providers including oncology nurses, surgeons and oncologists,
need to be appropriately equipped to educate women at a highly stressful time in their lives when they are already grappling with the emotional impact of their breast cancer diagnosis. This novel study represents a step toward that goal as it has produced a brief educational resource which will assist health care providers, including oncology nurses, to provide appropriate decisional support to women regarding TFGT during the stressful diagnostic period.
Acknowledgements

We are very grateful to the women who participated in this study and so generously shared their views. We would also like to thank Dr Kathy Tucker and Associate Professor Elizabeth Lobb for their valuable input into this study and assistance with ascertaining participants. This research was supported by a Fellowship Enhancement Fund to Bettina Meiser from the University of New South Wales. Associate Professor Bettina Meiser is supported by a Career Development Award from the National Health and Medical Research Council of Australia and a Cancer Institute New South Wales Career Development Fellowship.
Table 1: Participant demographics by group (N=26)

<table>
<thead>
<tr>
<th>Variable</th>
<th>Group A (N=14)*</th>
<th>Group H (N=12)</th>
<th>Total sample (N=26)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N</td>
<td>N</td>
<td>N</td>
</tr>
</tbody>
</table>
| Mean age (years, SD)                   | 42 (5.0)       | 43 (5.0)       | 42 (5.0)            |%
| Mean age at diagnosis (years, SD)      | 40 (5.0)       | 42 (5.0)       | 41 (5.0)            |
| Highest level of education             |                |                |                     |
| No post-school qualification           | 4              | 3              | 7                   | 27%
| Post school qualification              | 10             | 9              | 19                  | 73%
| Marital Status                         |                |                |                     |
| Married or cohabiting                  | 11             | 9              | 20                  | 77%
| Not married                            | 3              | 3              | 6                   | 23%
| Biological children                    |                |                |                     |
| Yes                                    | 9              | 5              | 14                  | 54%
| No                                     | 5              | 7              | 12                  | 46%
| Daughter(s)                            |                |                |                     |
| Yes                                    | 8              | 4              | 12                  | 46%
| No                                     | 6              | 8              | 14                  | 54%
| Previous cancer*                       | 2              | 0              | 2                   | 8%
| Family history breast or ovarian cancer|                |                |                     |
| Yes§                                   | 14             | 5              | 19                  | 73%
| No                                     | 0              | 7              | 7                   | 27%
| Mutation status                        |                |                |                     |
| BRCA carrier                           | 2              | 0              | 2                   | 8%
| Inconclusive result**                  | 12             | 0              | 12                  | 46%

*Group A denotes women who had TFGT, actual decision-making; **Group H denotes women who had not had TFGT – hypothetical decision-making; *Hodgkins lymphoma; Melanoma; **If a deleterious gene mutation is not detected in BRCA1 or BRCA2, and the participant does not have a family history of breast and/or ovarian cancer, it is unlikely that her breast cancer is due to an inherited mutation in a breast cancer protection gene. However, as not all breast cancer protection genes have been discovered, if a participant does have a strong family history of breast and/or ovarian cancer, it is still possible that she carries a mutation in an as yet undiscovered predisposition gene. For this reason, the result is termed, ‘inconclusive’, and the participant and her family may still be at increased risk of breast and ovarian cancer.

§ p < .01
### Table 2: Satisfaction with the educational materials by section

<table>
<thead>
<tr>
<th>Item</th>
<th>Description</th>
<th>Response Option</th>
<th>Yes n (%)</th>
<th>Non (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>10</td>
<td>Any parts of the pamphlet that you thought should have been explained in more detail?</td>
<td></td>
<td>10 (59%)</td>
<td>7 (41%)</td>
</tr>
<tr>
<td>11</td>
<td>Any information not covered in the pamphlet that you think should be included?</td>
<td></td>
<td>7 (41%)</td>
<td>10 (59%)</td>
</tr>
<tr>
<td>12</td>
<td>Any parts of the pamphlet you think could be left out?</td>
<td></td>
<td>4 (23%)</td>
<td>13 (77%)</td>
</tr>
<tr>
<td>15</td>
<td>In your opinion, was there anything in the pamphlet that was confusing?</td>
<td></td>
<td>7 (41%)</td>
<td>10 (59%)</td>
</tr>
<tr>
<td>17</td>
<td>Was the tone of the pamphlet positive enough for you?</td>
<td></td>
<td>15 (88%)</td>
<td>2 (12%)</td>
</tr>
<tr>
<td>13</td>
<td>How would you describe the amount of information in the pamphlet?</td>
<td></td>
<td>1 (6%)</td>
<td>15 (88%)</td>
</tr>
<tr>
<td>14a</td>
<td>Was the pamphlet clearly laid out?</td>
<td></td>
<td>14 (82%)</td>
<td>2 (12%)</td>
</tr>
<tr>
<td>14b</td>
<td>Was the pamphlet written in language that is easy to understand?</td>
<td></td>
<td>14 (82%)</td>
<td>2 (12%)</td>
</tr>
<tr>
<td>14c</td>
<td>Was the pamphlet useful?</td>
<td></td>
<td>14 (82%)</td>
<td>3 (18%)</td>
</tr>
<tr>
<td>14d</td>
<td>Was the pamphlet appealing to look at?</td>
<td></td>
<td>9 (53%)</td>
<td>5 (29%)</td>
</tr>
<tr>
<td>16</td>
<td>How satisfied were you with the information in the pamphlet?</td>
<td></td>
<td>10 (59%)</td>
<td>6 (35%)</td>
</tr>
</tbody>
</table>

Far too much/ too much | About right | Too little/ far too little

Very | Somewhat | Not very/ Not at all

Very satisfied | Satisfied | Dissatisfied/Very Dissatisfied
Table 3: Mean ratings of perceived improvement in understanding of treatment focused genetic testing (N=17)

<table>
<thead>
<tr>
<th>Item</th>
<th>To what extent do you think the pamphlet would have improved your understanding of …</th>
<th>Mean score (SD)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. What treatment focused genetic testing is</td>
<td></td>
<td>4.3 (0.9)</td>
</tr>
<tr>
<td>2. The purpose of treatment focused genetic testing</td>
<td></td>
<td>4.5 (0.7)</td>
</tr>
<tr>
<td>3. The relevance of treatment focused genetic testing in your situation</td>
<td></td>
<td>4.0 (0.9)</td>
</tr>
<tr>
<td>4. How treatment focused genetic testing is done</td>
<td></td>
<td>4.0 (0.9)</td>
</tr>
<tr>
<td>5. The benefits of treatment focused genetic testing in your situation</td>
<td></td>
<td>4.2 (0.9)</td>
</tr>
<tr>
<td>6. The disadvantages of treatment focused genetic testing in your situation</td>
<td></td>
<td>2.8 (1.4)</td>
</tr>
<tr>
<td>7. What it would mean if a faulty breast cancer gene was NOT found in me</td>
<td></td>
<td>3.8 (0.8)</td>
</tr>
<tr>
<td>8. What it would mean if a faulty breast cancer gene WAS found in me</td>
<td></td>
<td>4.2 (0.8)</td>
</tr>
<tr>
<td>9. What could be done is you were found to have inherited a faulty breast cancer gene</td>
<td></td>
<td>4.3 (0.7)</td>
</tr>
</tbody>
</table>

§ Higher mean ratings indicate greater improvement. Scores ranged from 1= ‘not at all’, 2= ‘a little’, 3= ‘somewhat’, 4= ‘quite a bit’ through to 5= ‘a lot’.
References


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offs and individualized treatment information for BRCA1/2 mutation carriers.

*Journal of Clinical Oncology, 22*(16), 3293-3301. doi: 10.1200/JCO.2004.05.066

