Are we being overly cautious? A qualitative inquiry into the experiences and perceptions of treatment-focused germline *BRCA* genetic testing among women recently diagnosed with breast cancer.

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ABSTRACT

Purpose: Women with breast cancer, who are found to be *BRCA*1/2 mutation carriers, have a high risk of ovarian cancer and metachronous breast cancer. Treatment-focused genetic testing (TFGT), offered around the time of diagnosis, allows genetic test results to inform surgical treatment decisions. However, concern has been raised that offering TFGT at this time may overly increase psychological burden. This study aimed to qualitatively explore women's attitudes and experiences of TFGT.

Methods: Women who had been diagnosed with breast cancer at age 50 years or less, undertook a semi-structured telephone interview (n=26). The sample included women who had been offered TFGT, based on family history and/or other risk criteria (*n*=14), and women who had been diagnosed within the past 6-12 months, and had not been offered TFGT (*n*=12). Interviews explored women's attitudes towards TFGT, perceived benefits and disadvantages, implications of TFGT, and impact on surgical decision-making. Interviews were transcribed verbatim and thematically analyzed.

Results: Women expressed positive attitudes towards TFGT and felt it was highly relevant to their surgical decision-making. They did not feel that an offer of TFGT shortly after, or at the time of diagnosis, added undue psychological burden. The majority of women interviewed felt that TFGT should be incorporated into standard clinical care.

Conclusions: TFGT is viewed favorably by women newly diagnosed with breast cancer. Future randomized controlled trials are needed to examine the long term impact of TFGT. We conclude that an offer of TFGT is not perceived as 'too much, too soon' by relevant patients.

INTRODUCTION

Traditionally genetic testing for genes associated with the hereditary breast/ovarian cancer (HBOC) syndrome has been offered to those with a moderate to high-risk family history of these cancers. However, from 6% [1] to 78% [2] of germline *BRCA*1/2 mutation carriers have no striking family history and several studies have shown that women with early onset breast cancer and only a small number of first and/or second degree relatives with breast or ovarian cancer are at higher risk of carrying a *BRCA*1/2 mutation than the general population [1, 3, 4]and therefore could be suitable for genetic testing.

Treatment-focused genetic testing (TFGT) for germline *BRCA*1 or *BRCA*2 gene mutations describes the process of genetic risk assessment and testing initiated at a time point to aid selection of cancer treatment and/or prevention. It has been used to provide women who are recently diagnosed with breast cancer with important information on which to base their treatment decisions [5-7]. Several factors point to the utility of TFGT at this time. Women that are found to be carriers of a *BRCA*1/2 mutation have a 43% and 35% risk of contralateral breast cancer with *BRCA*1 and *BRCA*2 mutations respectively [8], and a 42-48% risk of ipsilateral breast cancer recurrence by 13 years [9]. Bilateral prophylactic mastectomy is known to reduce the risk of metachronous breast cancer in *BRCA* mutation carriers by 96% [10, 11]. Bilateral risk-reducing salpingo-oophorectomy (RRSO) before the age of 50 also reduces the risk of breast cancer in these women by 53% and the risk of ovarian cancer by 90%, which offers a significant advantage given the limitations of surveillance for ovarian cancer, and poor prognosis [12, 13]. During preoperative chemotherapy, women who have a known *BRCA*1/2 mutation can be counseled regarding whether to undertake contralateral prophylactic mastectomy, breast

reconstruction and RRSO. Given that radiation therapy can interfere with plastic surgical reconstruction options, decisions regarding prophylactic management are best made when *BRCA* status is known [6, 14]. In the future TFGT may also guide adjuvant chemotherapy, if agents that target *BRCA* mutations, such as poly (ADP-ribose) polymerase (PARP) inhibitors [15] progress into the adjuvant setting. The concern, however, is that TFGT may also increase the burden of decision making at a vulnerable time for newly diagnosed women, especially if the genetic test result is inconclusive, or if surgical treatment is delayed while waiting for the test result [14]. An inconclusive test result can occur when a deleterious *BRCA1 or BRCA2* gene mutation is not detected in a woman with a strong family history of breast and/or ovarian cancer. As not all deleterious mutations have been discovered, it is still possible that the woman carries an unknown mutation, and may still be at increased risk.

In the past, *BRCA* genetic test results have taken several months to be returned to patients. With the improvements in genetic testing technology it is now possible for test results to be available within one to two weeks, allowing treatment decisions to be made in a timely manner. Palomares et al. [16] recommended that the optimum time for offering TFGT was during adjuvant chemotherapy, after excision of the primary tumor, to enable final surgical decisions regarding breast conservation or mastectomy to be made before radiotherapy would otherwise commence. They found that it was possible to obtain genetic testing results within this time frame (3-4 weeks).

It has been shown that newly diagnosed breast cancer patients are willing to undergo TFGT when available [14], and that it can be integrated into clinical practice [14, 17, 18]. Additionally, a high percentage of women opting for genetic testing (48%), elected to have bilateral mastectomy if they

were found to carry a *BRCA* mutation [19]. Whilst the feasibility of TFGT has been demonstrated, less is known about the acceptability and the psychosocial impact of offering this form of testing to women newly diagnosed with breast cancer, particularly among those without a strong family history of breast or related cancers. Schlich-Bakker et al. [20, 21] evaluated the impact of actively offering pedigree analysis and subsequent referral for genetic counseling to a group of women recently diagnosed with breast cancer and found that psychological distress did not increase. Looking more specifically at TFGT, a focus group study, assessing the perspectives of 13 mutation carriers from high-risk families, reported that women felt that the offer of genetic testing around the time of a breast cancer diagnosis may be too stressful, with women already having multiple demands on their coping abilities [21]. The focus group, however, did not include women who had actually undergone TFGT, and was limited only to women with a strong family history of breast or related cancers. As can be seen, the impact of TFGT is yet to be fully explored. What is known is based on pilot data, or extrapolated from the perspective of other target groups, such as those being offered genetic counselling only, without TFGT, or those considering a hypothetical scenario. Research, to date, has also been limited to those with an existing family history of breast or ovarian cancer.

The aim of this study was to qualitatively explore the actual experience of women who were eligible for, and had TFGT during their treatment process, as well as the hypothetical views toward TFGT of women who had been diagnosed with breast cancer, but had not had TFGT. The views of women with and without a strong family history of breast and/or ovarian cancer were included because both groups are likely to be targeted for TFGT in the near future as risk factors, other than family history, are incorporated into genetic testing criteria.

METHOD

Sample

Women diagnosed with breast cancer at 50 years of age or younger were recruited to the study. This age criterion was selected as younger women may be more likely to use mastectomy as a preventive measure because they have less life-years at risk that would benefit from a mastectomy. Group 1 comprised of women who had been offered TFGT to facilitate surgical decisions. Women in Group 1 had been offered TFGT for a range of indications such as being at high risk based on their family history [22]; or had been diagnosed with cancer before the age of 40, and/or had another risk factor i.e. another affected family member, or Ashkenazi Jewish ancestry. In Australia, the cost of the consultation with a genetics service is covered by a government subsidy through the national health system (called Medicare [23]) and there is no cost to the patient for genetic testing. Group 2 included women who had been diagnosed with breast cancer within the last 6-12 months, unselected for family history, and who had not had TFGT. To avoid undue participant burden, only women who had not relapsed were included in either group. Women were excluded if they were under 18 years of age, had insufficient English proficiency to undertake the interview unaided, or if they had a mental or intellectual disability.

At the time of the study, Group 1 received genetic counseling through their local familial cancer service, and all were referred for TFGT. Group 1 were seen on an urgent basis, and received genetic testing results within ten working days. Group 2 were identified through their oncology clinic and were not referred for genetic counseling or TFGT.

Recruitment

Group 1 was ascertained through two major familial cancer clinics in Sydney and Melbourne, Australia, and Group 2 through an oncology clinic at a major teaching hospital in Sydney. All potential participants received a letter of invitation from the treating clinician. Ethics approval was obtained from the relevant Human Research Ethics Committees.

Procedure

Prior to participation, women were mailed a consent form and a one-page information sheet regarding TFGT specifically designed for the purpose of this study. Women were asked to return the signed consent form and to read the information sheet prior to the interview. The study coordinator (MG) contacted each woman who provided informed consent to schedule a telephone interview at a time of their convenience.

Semi-structured telephone interviews were conducted by MG using an interview guide, while leaving wording and sequencing of discussion topics open, with probes to elicit more information, as appropriate. The interviews explored women's attitudes towards TFGT, the perceived benefits and disadvantages, perceived impacts on both the patient and family, and impact on surgical decision making. Interviews also explored attitudes towards the educational materials posted to participants, and preferences regarding the timing of the offer of TFGT, the mode and format of information delivery, and format of information delivered, which is reported elsewhere [24]. Emergent themes from early

interviews were used to guide lines of questioning in subsequent interviews, to ensure that divergent points of view were explored [25]. Sampling was discontinued when data saturation was reached [26].

Data analysis

The conceptual framework of Miles and Huberman [25] was used to guide the analysis. Initial themes were identified and MG and KW coded two transcripts concurrently, to confirm reliability of the coding scheme and further refine and expand on emergent themes. If discrepancies occurred with respect to specific themes, discussions took place until consensus was achieved. The remaining transcripts were then coded by EZ, using the qualitative data analysis software QSR NVivo 8.0. EZ also cross-tabulated emergent themes by participant characteristics [25]. The use of multiple coders and analysts is a strategy suggested by Miles and Huberman to reduce the potential for researcher bias and to increase the validity of the findings [25].

RESULTS

A total 56 letters of invitation were mailed out by the treating centres (see Figure 1). Eleven women did not respond. Of the 39 women who responded, two women declined participation; five were ineligible to participate, and one woman could not be contacted for interview. The data for 2 of the 28 women interviewed were excluded because it was established during their interview that they had genetic counselling and testing *after* their definitive breast cancer treatment. A total of 26 interviews were transcribed and analysed. Participants (N = 26) included women with breast cancer who had either previously had TFGT (Group 1, n = 14), or women, recently diagnosed with breast cancer, who had not had TFGT (Group 2, n = 12) (participation rate across both groups 55%). Table 1 shows women's

sociodemographic, medical and family history characteristics. The mean age of participants at interview was 42 years, with a mean age at diagnosis of 41 years. The majority of participants was married or partnered, had post-school qualifications, and had a family history of breast and/or ovarian cancer. Fifty-four percent had children. Hereafter, participants will be denoted by their group (1 or 2); C will denote participants with children, and NC participants with no children, followed by their identifier.

[Insert Figure 1 about here]

[Insert Table 1 about here]

ACCEPTABILITY OF TFGT

Treatment focused genetic testing was equally acceptable to women across both groups, with the majority wanting the offer of genetic testing at or soon after their breast cancer diagnosis (*n*=17). Three women felt that the acceptability of TFGT was conditional on it being offered to women most likely to be found to be mutation carriers, or as long as it did not delay surgical options. For women who had been offered TFGT, the majority (*n* = 10) reported deciding to undertake genetic testing as soon as it was first offered, with most of these (*n* = 7) expressing the feeling that there was no decision to be made. One participant from Group 1 expressed the view that she needed time while waiting for her test result, to process the implications of TFGT because she felt overwhelmed by her recent cancer diagnosis. Quotes to illustrate the actual and hypothetical acceptability of TFGT are shown in Table 2.

[Insert Table 2 about here]

IS TFGT A SPECIAL TEST?

Women were asked to report whether they perceived TFGT to be different from other tests that they were required to undergo at diagnosis, and whether they felt it should be incorporated into their standard medical care. Seven participants perceived that TFGT was different to other tests due to its family implications. Nine women, however, felt that TFGT was no different to other tests they were subjected to shortly after their diagnosis. Twelve participants felt that TFGT should be part of the standard treatment process, offered to all women who met the inclusion criteria. An additional four women felt that it could be included in the standard testing regime, conditional on the test and its implications being adequately explained to patients. A recurring theme expressed by participants was 'it's just another test'. Table 3 presents quotes that illustrate participants' views on TFGT as a test.

[Insert Table 3 about here]

ADVANTAGES OF TFGT

The main advantage of TFGT expressed by participants (*n* = 13), was the opportunity to receive and emotionally deal with all bad news at one time. Not only was it about dealing with emotional responses 'all in one go', but TFGT also provided the opportunity to receive information to help guide decision-making about surgical options. TFGT also offered patients a sense of certainty (*n* = 6), and potentially opened up a range of treatment options (*n* = 10).

A significant motivator for TFGT reported by most participants (*n* = 19) was to obtain information about family risk, and to provide screening and testing choices, particularly to first-degree relatives, with a view to preventing the disease in others.

DISADVANTAGES OF TFGT

Increased anxiety, particularly in relation to waiting for the test result, at an otherwise already overwhelming time was reported as a potential disadvantage by ten participants. However, only two of these participants were from the group that had actually had TFGT. The perceived anxiety associated with waiting for the test result was sufficient for one participant to feel she would decline TFGT, had it been offered to her. Only one participant reported the impact of genetic testing on life insurance as a potential disadvantage of TFGT. While women acknowledged that the offer of TFGT occurs at a highly stressful time, the overriding theme was that the potential benefits of TFGT outweigh the disadvantages.

Four women reported that any cost associated with TFGT would have excluded them from testing, as they would not have been able to afford the additional expense. Almost half of the women interviewed (*n* = 11), however, reported that the benefits of TFGT were sufficient to warrant them paying the full cost of testing themselves (estimated to be over AUD2000), and an additional nine participants felt they would pay part of the cost. Table 4 presents quotations illustrating participants' views regarding the advantages and disadvantages of TFGT.

[Insert Table 4 about here]

IMPLICATIONS OF TFGT

Implications for the patient

A number of women (*n* = 8) felt the focus of testing shortly after diagnosis was to provide information for themselves, on which to base treatment decisions. The most frequently reported actual or hypothetical implication of a mutation positive result through TFGT on the woman herself was on surgical decisions. With the increased risk of contralateral breast cancer, the majority of women (*n* = 12) felt they would opt for a bilateral mastectomy. Nine participants expressed concern regarding the increased risk of ovarian cancer, and four would have a bilateral RRSO if they were found to be carriers.

The majority of women in Group 1 described the genetic test result as a significant element in their decision-making process regarding surgical options (*n*=11). Two women, who were at moderate risk based on family history, who were found not to have a *BRCA*1/2 mutation, elected a breast conservation approach. Two women found to carry a *BRCA*1 mutation elected a bilateral mastectomy, with one also choosing RRSO. Women who were at high risk, based on family history, who were found not to carry a *BRCA*1 or *BRCA*2 mutation were given an 'inconclusive' result (*n*=10). Of these women, the majority (*n*=6) reported finding this result reassuring, and elected conservative treatment. The remaining four women, however, chose to have bilateral mastectomy as they felt their high risk status was sufficient to warrant preventative measures.

Implications for family members

Fifteen women reported that after their surgical decision-making needs had been met, a secondary implication of TFGT was for other family members, particularly for first-degree relatives such as children and siblings. Table 5 presents quotations summarizing participants' views on the actual and hypothetical implications of TFGT for patients and family members.

[Insert Table 5 about here]

Communication with family members about TFGT

While the majority (*n* = 13) of women reported feeling able to discuss TFGT with their family, and the expected implications, several felt that other family issues and the potential for family members to blame themselves in relation to hereditary factors, would make the discussion difficult initially. When relationships with specific family members were difficult, several enlisted the assistance of another family member, such as their mother, to contact those relatives. Many women (*n* = 11) felt a duty to inform other family members about their decision to have TFGT and the potential implications of a mutation positive result. Table 6 presents quotations illustrating participants' views on communicating with family members about TFGT.

[Insert Table 6 about here]

DISCUSSION

TFGT was found to be highly acceptable to women in this study, both among those who had and those who had not experienced it. Participants felt that it allowed them to make informed decisions regarding their treatment options, particularly in light of the increased risk of ovarian cancer and contralateral breast cancer. The majority of participants expressed relief or anticipated relief at receiving an inconclusive result, for the reassurance it gave them regarding their children's subsequent risk. On the other hand, if they were found to be mutation carriers, it was believed to offer the opportunity for

genetic testing of unaffected relatives and targeted preventative measures. The immediate clinical utility of this information has been noted in previous research. Schwartz et al. [19] reported 48% of newly diagnosed breast cancer patients with a *BRCA* 1/2 mutation opting for prophylactic mastectomy. Weitzel et al. [17] found that of 37 women with a recent diagnosis of breast cancer at the time of receiving genetic counseling, 32 proceeded to genetic testing, and of those who received a positive BRCA1/2 test result, 100% chose bilateral mastectomy. Women have also reported finding TFGT and genetic counseling extremely helpful when facing future medical decisions [27, 28].

One of the concerns, however, regarding TFGT has been the potential for an increased psychological burden for women recently diagnosed with breast cancer. A previous study [28] found that breast cancer patients who had been diagnosed within the year prior to genetic testing reported higher breast cancer-specific distress than those who had been diagnosed more than a year prior to testing. Ardern-Jones et al. [29] conducted focus groups with women who had been diagnosed with breast cancer before the age of 40, asking them to consider their response to an offer of TFGT, had it been available to them. They reported a range of attitudes, with the majority feeling that TFGT was "too much, too soon". The authors acknowledged, though, that the study was retrospective and asked hypothetical questions about reactions to a future scenario. Vadaparampil et al.[30], in a qualitative assessment of the impact of a surgeon referral letter on recently diagnosed breast cancer patients' uptake of BRCA genetic counseling, reported that of the women interviewed, few mentioned any implications of genetic counseling on their ability to cope with their recent cancer diagnosis. They did, however, find that women that did not attend genetic counseling reported the offer of genetic counseling and testing shortly after their diagnosis to be overwhelming.

Health professionals have also expressed reservations about offering TFGT to breast cancer patients which could be a barrier to any future, wider, introduction of TFGT [29, 31]. Specifically in a study by Lobb et al.[31], 34 cancer genetics practitioners considered that TFGT raised a number of ethical issues concerning decision-making and consent for women at a time of emotional vulnerability immediately following their breast cancer diagnosis. They also identified the need for practitioners to be aware of the unaffected family members' interest in genetic information, and the management of this service within familial cancer clinics.

Participants in our study, however, indicated that they believed they would be able to weigh up and integrate the implications of genetic testing, without causing undue psychological distress. One explanation for the discrepancy between our findings and some of the previous research may be that the context of TFGT is significantly different from that of conventional genetic testing, that is, genetic testing once cancer treatment is complete. TFGT offers significant and personally relevant information on which to base immediate treatment decisions. Participants in this study agreed that a major focus on the relevance of the genetic information for other family members would be better left until after their own cancer treatment was complete, and that the focus of TFGT should be on their own surgical decisions. In accord with the views expressed by women in our study, Schlich-Bakker et al.[32] found in a prospective study of 58 women who underwent TFGT, that there was no increase in psychological distress 12 months following testing, compared to a control group. Tercyak et al. [33] also reported that the genetic test result did not predict quality of life or distress in women who underwent genetic testing before definitive surgery.

Acceptance of TFGT, and the positive attitudes towards it, did, however, come with a proviso. Participants in our study expressed the view that while they felt TFGT should become part of standard care; they acknowledged that it did have extended impacts on other family members. They recommended it be offered to all women that meet predetermined selection criteria, but felt it required adequate decision-related support.

Women in our study also recommended TFGT be offered before surgical decisions needed to be made. The advantage of offering testing shortly after diagnosis was that it allowed complex decisions about surgical procedures to be addressed at a time when most relevant, so that treatment could be streamlined and targeted. Silva et al. recommended TFGT be offered while patients are having neoadjuvant or adjuvant chemotherapy, prior to radiotherapy [6]. Additionally several studies have now shown that TFGT can be incorporated successfully into clinical practice [14, 17, 18] with TFGT being offered before definitive surgical treatment [19, 30], during systemic therapy[18], immediately following biopsy or during chemotherapy[17].

It is important to acknowledge several limitations of this research. The study was retrospective in that some women were asked to reflect on their past experience of TFGT. Furthermore, others who had not had had TFGT were asked about their hypothetical views. A strength of the study is that it provides the first qualitative exploration of women's actual experiences of TFGT, whether from low, moderate, or high risk families. As such, it offers in-depth information on the experiences and acceptability of TFGT among breast cancer patients who are likely to be targeted for this type of testing in the near future.

Participants expressed the view that the advantages of TFGT outweighed the disadvantages; that with adequate education and support, TFGT could be incorporated into standardized care for young women recently diagnosed with breast cancer, and that is was not “too much too soon”.

CONFLICT OF INTEREST

The authors have no conflicts of interest, financial or otherwise, to declare.

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Table 1. Participants' demographic and medical characteristics, by group, at time of interviews

Variable	**Group 1 (*n*=14)[a]**	**Group 2 (*n*=12)[b]**	**Total sample (*N*=26)**	
	M *(SD)*	M *(SD)*	M *(SD)*	
Mean age at interview (SD)	41.6 (4.8)	43.0 (4.9)	42.3 (4.8)	
Mean age at diagnosis (years, SD)	39.9 (4.9)	41.6 (4.9)	40.7 (4.9)	
Time since diagnosis (years, SD)	1.4 (0.7)	1.0 (0.0)	1.2 (0.6)	
Highest level of education	*n*	*n*	*N*	%
No post-school qualification	4	3	7	27
Post school qualification	10	9	19	73
Marital Status				
Married or co-habiting	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
Family risk status				
Low	2	0	2	8
Moderate	2	10	12	46
High	10	2	12	46

*Hodgkin's lymphoma; Melanoma; [a]Group 1 denotes women who had TFGT, actual decision-making; [b]Group 2 denotes women who had not had TFGT – hypothetical decision-making. **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.

Table 2. Quotations regarding the actual and hypothetical acceptability of TFGT.

Concept	Quotation
TFGT is acceptable	I think it's like being pregnant and being offered you know being 40 and being offered the amnio and the this and the that you know, I think it's, you, you know if you didn't know these things afterwards you'd look back and say well why didn't they tell me, so (1, C, 017)
	I think I would have wanted it because that would have given me, I suppose, further options. Whether or not I would have still done what I did is, you know, beside the point. But if you know that there is a gene within you that is more likely to develop a cancer elsewhere in the body as well, then it would give me more information to help my own family, you know. So, yeah (2, C, 064)
TFGT is acceptable, with some conditions	Only if it's relevant to them. (1,NC,025)
	I would have [had TFGT] but if it meant that I'd have to wait a month, I actually would find that quite stressful. I'm thinking the longer it's there, the more opportunity for the cells to spread. (1,C,053)
	Yes, I – but then I would have been guided by Dr [name deleted] and Dr [name deleted] in that if they said to me "Look, if you want to have it done, you can do it, but I'm telling you that it's unlikely" then I wouldn't have done it [okay, okay], but if they're saying to me "Look, there's only one member for example of your family, that's died of breast cancer, but that's enough [okay], and I'm suggesting you have the test done" I would have had the test done and I would have probably gone ahead and had a bilateral if that had been the advice (2, NC, 034)

Table 3. Participants' actual and hypothetical views on TFGT compared to other tests and whether TFGT should be incorporated into the standard testing processes

Concept	Quotation
TFGT is different	Oh no it's so different, because all the other tests are only for you but this gene test is for your family as well as yourself. (1,NC,076)
	Yeah, I do. Not so much in the mechanics of it, more in terms of the implications because it's not just the pathology of the cancer, it's - yeah, it's the implications for ovarian cancer which is something that you know I've always been a little bit worried about. (2,NC,033)
TFGT is just another test	I think it's one more test that you'd have in that period where you're having all your others. When you're first diagnosed, they then send you off for CAT scans and bone density and so just the one more test to go through.(1,C,053)
	No. It's just another test. Yeah, nothings you know. It will help in your decision but you've got to do so many tests anyway so you know what's one extra test. You know you do countless tests anyway so it's just one extra test you know just taking a bit of blood so it's nothing you know, nothing extra really. Your just doing oh I can't even remember how many I did. It's just another test you know yeah. (2,C,065)
TFGT as part of the standard treatment process	Absolutely. Yeah, I think it would benefit a lot of women and to be part of the whole – what do you call it, screening – I don't know, screening – to be part of the whole – yeah, it's absolutely necessary. I'm sure there's a lot of people in my situation it has to be. (1,C,061)
	Knowing that it's just part of the whole process. That it's just – yeah, you are waiting on all the other tests results anyway, you're waiting on that one too, you're not then sitting down and discussing all the possible implications of the genetics test, and the implications to the family before you even know (1,NC,045)
TFGT as part of standard treatment process, with some conditions	I think so. If the people have got you know a strong family history I think that maybe it would be good, yeah. (2,C,065)
	Spelt out, yeah, that's right. You've always got to I guess take the care that just because it's part of the process doesn't mean that these bits are bleeped over and taken for granted sort of thing which is always a risk. (2,NC,073)

Table 4. Participants' actual and hypothetical views regarding the advantages and disadvantages of TFGT

Concept	Quotation
Advantages of TFGT	*All in one go* If you've just had a lumpectomy, and you've had surgery, and then you find out you're genetically positive, they you've got to go back and have more surgery. So from that aspect you'd want to know early. (1,C,061) I would rather know beforehand, before the treatment than have to face it afterwards. If I found out I carried that gene, they'd all have to go through a whole lot of new emotions anyway, so as raw as it is, I think I'd rather handle everything all at once. (2,C,018) *Guides decision-making* If it had come back positive then you know you have the ability to decide on appropriate medical action. I like that it's more targeted. (2,NC,034) *To assist family members* It's the future. I've got what I've got. I've got to deal with it, but it's the next generation down. If we knew it was genes, then these kids could be on track with it. (1,NC,076)
Disadvantages of TFGT	*Increased anxiety* I'm wondering what it would be like to actually get the diagnosis and a positive BRCA result and have not only am I going to die but my family. That might just be too much. (1,C,055) The fear of the unknown whilst you're waiting for the results. I see that as a very real thing, particularly when you're first told you have cancer. (2,NC,034) *Cost* Look no I think that's a difficult one because the cost in the private, you know I've got top level private health cover and it has cost me so far $20,000 about. And I'm about to go and have the other breast off and on and the gap on that's going to be about another $6000, $7000 and you know it is terribly expensive you know never mind the loss of earnings, it is an incredibly expensive business having breast cancer, you know a really expensive. And that's not counting the taxis home from chemo and you know etc etc. (1,NC,052)

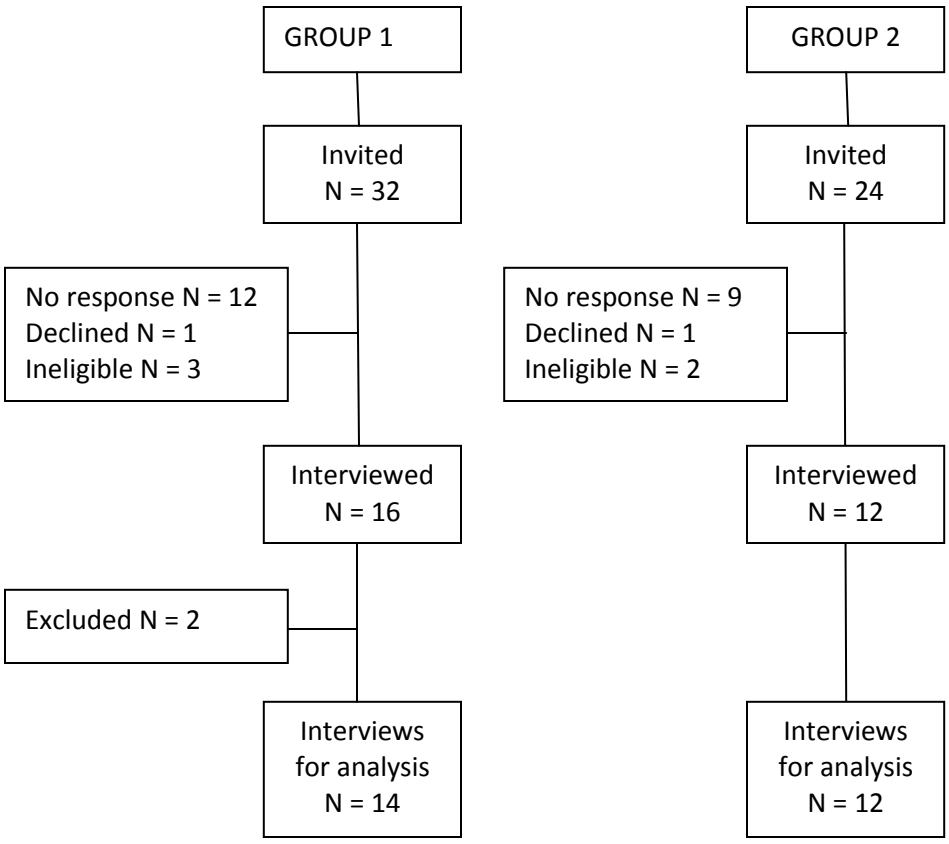
Table 5. Participants' actual and hypothetical views on the implications of TFGT for oneself and family members

Concept	Quotation
Implications for the patient	*Focus on oneself* You are there primarily for yourself in the first instance, and making decisions that affect you and saving your life, and whatever you find out for your family, for me, is more about forewarning them to be more diligent, but it's not my primary reason for doing it. (1,C,053) I would say just for myself at the initial because it's going to be, that's the first, you're only thinking about yourself. I mean maybe if you were to speak to a mother, a person who's got a young daughter or something, her answer would be completely different. But I would say myself for now and obviously then you know you will get the results and then you can start asking the questions and what you need to do with the, in regards to your family members. (2,NC,021) *Impact on surgical decisions of a mutation positive result* I mean obviously down the line we knew because my process took a while and um because I know when I made the decision after seeing Dr [name deleted], that um that I was a carrier because we knew to take off the second breast, so that's what, and it was just crystal clear for me as to what had to be done. (1,C,017) Right, well I would then say "Let's talk seriously about getting rid of the other breast definitely, absolutely, positively", and there are a few minor advantages to that cosmetically in that I haven't yet had my reconstruction, where I'm to be reconstructing two breasts, like now I'd be reconstructing one and perhaps having the mastectomy on the other at the same time, that would be a very satisfying solution to that. (2,NC,034)
Implications for family members	*Impact on family members of a mutation positive result* But it would mean that I would have to consider having a double mastectomy, and it would be something that my daughter, and my nieces, and my sister would all have to know about it. Yes, they would have to find – I mean, I know my sister doesn't know all that much about the genetic side of things. You know, it's important to know – it's really important to know. (2,C,068) And it was also going to mean that I was going to be bringing bad news to my family because I was going to be telling them that I had this mutation, and my sister, you know, might have to be thinking about that too, although she's ten years older than me but – I sent her off to go and get her mammogram and ultrasound as soon as I was diagnosed anyway but (1,C,072)

Table 6. Participants' actual and hypothetical views regarding communicating with family members about TFGT

Concept	Quotation
Potential barriers	It was only because my dad had reached the stage that he'd become terminal, so we had that going on as well. We were trying to deal with that whilst trying to help my mum care for my father whilst trying to start going though the whole thing myself. And I guess my father and I are both sensitive to each other's vulnerabilities, so in one way it was a bit difficult. (1,C,047)
	I was a little bit nervous about how my mother would react. I thought my mother might be thinking it was her fault, but other than that, it was not particularly difficult. I mean, I approached it as its relevance to me and my treatment. I tried, when I discussed it with them first off, almost to shy away, or not make a big deal of it being inherited. Even though I mentioned it, I probably mentioned it being a mutation in my personal make-up rather than inherited. I let that come into the conversation at a subsequent time. (1,C,072)
Duty to inform	More of a duty to warn because I didn't want to make a moral judgement that they should, so I didn't say you should go and get tested, I said I have got tested, we, I don't have, I don't have any markers, however I have you know spent two separate hours with the director of the family cancer unit, who's an oncologist and a geneticist and her view is that we have you know a definite predisposition and a significant family history. So you know I'm taking action on that and, and she has said that if you want to do testing or stuff, she's happy to send the information over to you know your GP or something. So I, it was more of a duty to warn in that sense (1,NC,052)
	I'm surprised because usually the way it seems to work is that you know a new development or a new diagnostic tool happens and it gets introduced and then it just becomes the correct part of management. That's what I think happened with say colonoscopies or mammograms if you've had a first degree relative with that type of cancer. There was no umming and arhing about should you, should people be told or not. Why is it that we are wondering about this rather than just implementing it? (2,NC,015)
Enlisting help from family members	I think – my family networks sort of works through my mum, she's the one who sort of tells people things and so forth. And my sister, [name deleted], she would have probably done all that because at the time I was probably a bit fragile to sort of – (2,C,064)

Figure 1 – Progress of participants through the study.



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