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PAPER

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Factors influencing the decision to share cancer genetic results among family members: An in-depth interview study of women in an Asian setting

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Abstract

Objective: Reluctance to share hereditary cancer syndrome genetic test results with family is reported among Asian patients. This study aims to explore patient factors influencing result sharing with family, to improve overall testing uptake.

Methods: Participants were women with a personal/family history of breast and/or ovarian cancer who received a positive, negative, or variant of uncertain significance test result. In-depth interviews were conducted to theme saturation to explore facilitators and barriers for sharing results with family. Grounded theory with thematic analysis was applied in analysis and interpretation.

Results: Twenty-four women participated. Three themes representing facilitators emerged for all results categories: family closeness, involvement of families in the testing process, and perception of low emotional impact of results. In the positive result category, 2 facilitator themes emerged: presence of actionable results and perception of family members' acceptance. In the negative and variant of uncertain significance result categories, 2 themes representing barriers to sharing emerged: perception of no genetic or medical implication for family and result ambiguity.

Conclusion: Facilitators and barriers for result sharing are similar to those among Western women. A framework to explain Asian patients' decision-making process identifies optimal counselling opportunities to enhance communication with family.

KEYWORDS

Asian, cancer, cancer genetics, decision-making process, genetic testing, in-depth interviews, oncology, qualitative

1 | INTRODUCTION

Genetic testing for hereditary conditions has revolutionized the concept of preventive medicine. Testing allows for a personalized assessment of an individual's health risks and helps the individual to seek early disease prevention and/or treatment options.[1] Hereditary genetic information affects not only the person being tested but also their family members. Test information sharing with family members is key to further predictive testing.[1] In particular, for hereditary cancer syndromes, predictive testing of at-risk relatives has been shown to provide the greatest benefit at both the individual and population levels.[2] For example, predictive testing of families with a *BRCA1* or *BRCA2* mutation identifies currently asymptomatic family members and allows for intensified surveillance and/or risk-reducing interventions, which can result in improved survival.[3,4] Predictive testing can also identify relatives who have not inherited the familial mutation, saving them the burden

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of further investigation as *BRCA1/2* mutation carriers. Informing family members about test results is the first important step patients can take in promoting further predictive testing.

Studies have reported low rates of disclosure of *BRCA1/2* genetic test results to family members and lower uptake of predictive testing among Asian compared with Western populations.[5,6] Disclosure of genetic information to family is influenced potentially by the result itself, and by complex personal, cultural, and social factors.[7,8] There are the 3 possible test results in genetic testing, which complicates the decision-making process: (1) positive (a pathogenic mutation is found and indicates an increased risk of cancers), (2) negative (no pathogenic mutation is detected), and (3) variant of uncertain significance (VUS, an alteration in the gene sequence with unknown consequence on the function of the gene product or risk of causing disease).[9] The majority of existing literature in family communication has focused on the sharing of positive genetic test results, and little has been reported about information sharing among patients with negative or VUS results.[7,8,10,11]

We therefore conducted a qualitative study among Asian women who had recently undergone germline genetic testing for hereditary breast and ovarian cancer syndrome, who tested positive, negative, or VUS, to explore facilitators and barriers for information sharing with their family. Our goal was to construct a framework with which to understand the decision-making process for sharing results, to inform best approaches to improve predictive testing among patients of Asian origin.

2 | METHODS

2.1 | Study setting and participants

Singapore is a Southeast Asian multiethnic, multicultural island country of 5.5 million served by 7 public hospitals and 9 specialty centres. The National Cancer Centre Singapore is one of the 2 cancer specialty centres in Singapore. Presently, the Cancer Genetic Service at National Cancer Centre Singapore sees between 80 and 120 new cases per month for genetic testing of hereditary cancer syndromes.

Participants were women who had attended Cancer Genetic Service for genetic counselling. Participants were recruited based on the following criteria: (1) English speaking; (2) aged 21 and over; (3) referred for genetic counselling due to either a personal and/or family history of breast and/or ovarian cancer; and (4) underwent genetic testing for hereditary breast and ovarian cancer syndrome. We sought to explore facilitators and barriers for information sharing with the 3 types of genetic results. Thus, our goal was to recruit an equal proportion of patients with positive, negative, and VUS test results. We used a purposive sampling technique over 10 months (from December 2015 to September 2016). All patients were recruited face-to-face, by a clinical cancer geneticist or one of 2 genetic counsellors after receiving their test result. We obtained informed consent at the time of recruitment. The study was approved by the Singapore Health Services Centralised Institutional Review Board (CIRB2015/2012) and conducted in accordance with the Declaration of Helsinki.

2.2 | Study design

We chose a qualitative approach to elicit not only known patient factors but also previously unreported issues related to genetic test information sharing with family among Asian women. In-depth interviews were used rather than focus groups because of the topic's sensitivity and privacy and the potential for emotional responses in participants. In-depth interviews have the advantage of allowing for exploration of personal thoughts, behaviour, and values in a safe environment and avoid the influence of peer judgement.[12]

2.3 | Data collection

We designed an open-ended, semistructured interview guide to explore key areas, based on our previous clinical experiences and a literature review.[13-15] The draft interview guide was pilot-tested on colleagues and 1 volunteer patient not participating in the study. Following the pilot, the research team consisting of clinicians, health services researchers, sociologists, and an educator added expertise and knowledge of local health literacy and education among patients to refine the final question guide and probes (Table **S1**). The question guide explored participants' reactions to their genetic results, who they shared the results with immediately after receiving them, and the reasons why they did or did not share their results. Interviews were conducted in English, by one interviewer (L. Y. R.) and one moderator (S. S.) who were sociology researchers not involved in the participants' clinical care and who had no relationship with participants prior to the interview. Interviews were conducted in person, in private rooms that ensured participant privacy and confidentiality. Interview duration ranged between 30 and 60 min. All interviews were audio-recorded and transcribed verbatim. Interviews were conducted between 2 days and 9 months (average: 45 days) after participants received their genetic test result. Participants were given a SGD40 voucher to cover transportation costs. Data analysis occurred concurrently with data collection to ensure theme saturation. Interviews were conducted to saturation, defined as 2 consecutive interviews with no new themes identified for each of the 3 result categories (positive, negative, and VUS).

2.4 | Data analysis

Thematic analysis is a search for themes that emerge as being important to the description and explanation of the issue or phenomenon being examined.[16,17] Pattern recognition is used to identify categories of emerging themes relevant to the research question. Two researchers (S. L. and Z. O.) analysed the transcripts manually for emerging themes and key quotes following Braun and Clarke's[18] 6-step method (Figure S1). Each independently read and coded the first 3 deidentified transcripts to identify major themes. They then met to construct a coding schema, which was independently applied to the subsequent transcripts. The coding schema included only themes that were relevant to the research question. The coders met again after applying the schema to all transcripts, to arrive at consensus about the major themes for each type of test result (positive, negative, and VUS) and any new themes not previously identified. All the transcripts that were previously coded with preliminary coding schema were subsequently reanalysed with the final code schema. Themes common to

the 3 result categories were collated, and those unique to each result category were separated. Potential discrepancies were discussed and resolved. A third, adjudicating researcher (D. L.) participated in resolution of disagreements.

We adopted a grounded theory approach, using the major themes generated, to construct a framework to understand participants' decision-making process about sharing their genetic results with family members.[19] We chose an inductive approach[20] and applied grounded theory[21,22] to the data throughout the process, by interpreting the themes in light of the pertinent research question (ie, exploring the factors that influence the sharing of genetic test results).

Study rigour was addressed through dependability, credibility, transferability, and the use of thick descriptions.[23] Dependability was assured by precise record keeping and maintaining an event log to establish an audit trail. Credibility was accomplished using peer debriefing, in which thematic analysis was discussed with a third (noncoder) researcher, and by inclusion of multiple perspectives (geneticist, clinician, primary care physician, sociologist, and educator) in the analytic and interpretive process. The use of purposive sampling to select participants provided representative views and allows for transferability. Thick descriptions were achieved by analysing a large volume of qualitative data within the transcripts.

3 | RESULTS

Sixty-four eligible women were approached in our clinic between December 2015 and September 2016; 32 women consented to take part, and 8 dropped out before being interviewed, either due to scheduling conflict or withdrawal of consent. Twenty-four participants completed the interviews (Table 1). The final themes consisted of 3 major themes common to the 3 results categories: 2 unique themes for the positive and 1 unique theme each for the negative and VUS results categories. Theme saturation was achieved for each of the 3 categories after 8, 7, and 7 transcripts for positive, negative, and VUS results, respectively. The common and unique themes are described and explored in detail below.

TABLE 1 Demographics of in-depth interview study participants, National Cancer Centre, Singapore, 2017

		Genetic Result (N = 24)		
Demography		**Positive[8]**	**Negative[8]**	**VUS[8]**
Median age, y (range)		48 (31-64)	44 (35-60)	43 (27-60)
Race	Chinese	4	6	8
	Malay	3	2	0
	Others	1	0	0
Marital status	Married	4	6	5
	Single	3	1	2
	Divorced	1	0	1
	Widowed	0	1	0
Children	Yes	6	4	5
	No	2	4	3
Personal history of breast and/or ovarian cancer	Yes	7	7	8
	No	1	1	0
Family history of cancer	Yes	8	6	7
	No or unsure	0	2	1

Abbreviation: VUS, variant of uncertain significance.

3.1 | Common themes for all 3 result categories

3.1.1 | Family closeness

Regardless of the results, the majority of participants expressed their willingness to share their results based on the degree of perceived "closeness" with their family members. The willingness of the participants to share the information regardless of the results (positive, negative, or uncertain) was defined by level of emotional intimacy or bonding. None of the participants defined closeness by degree of blood relation. The quotes below represent these feelings:

> *Me, my sisters and my family members are quite close, that's why I share [my genetic result] with them. (P1, positive result, 54 y)*

> *Immediately after I got my result, I called my parents [to tell them the genetic result]…. I am very close to them. (N5, negative result, 35 y)*

> *There's nothing that we keep from each other in my family, so I shared the information [genetic result] with them [my family]. (U7, VUS result, 44 y)*

3.1.2 | Involvement of families in the testing process

Participants also reported that they were more likely to share their genetic result with the family members who were aware of or involved in their decision to undergo genetic testing. Participants expressed a sense of "duty to inform," in that they felt indebted or bonded with the person who either encouraged or supported their decision to be tested. The feeling persisted regardless of whether the genetic test results were positive, negative, or uncertain and is seen in the following quotes:

> *They [sister and family members] knew I went for test, that's why I let them know the genetic results. (P1, positive result, 54 y)*

> *I shared it [my results] with my husband because he's the one that knows about my appointment…. I would share whatever results…negative or positive with him. (N1, negative result, 38 y)*

> *Before I took the [genetic] test, I talked to my daughter. My daughter was the one who suggested the testing…. So I did talk to her [about my result]. (U6, VUS result, 60 y)*

3.1.3 | Perception of emotional impact on family

Participants reported that they would delay sharing or would not share their genetic result if they believed it would add mental or emotional burden to their family members. These participants attempted to protect their relatives from perceived negative emotional effects of the

test results, even if the results were negative. Perceived emotional impact included fear, anxiety, and worries. Participants felt that sharing of results would be burdensome to their family members and that by not sharing they could protect their relatives.

> *I am waiting for the right time to talk [about my positive result] to my two girls, because I don't want to frighten them. (P3, positive result, 46 y)*

> *My parents are elderly and I do not want them to worry.... I didn't really share [my negative result] with them.... [Because] I knew it would still cause them a lot of worry. (N1, negative result, 38 y)*

3.2 | Unique themes: positive result category

3.2.1 | Presence of actionable genetic results

A strong motivating factor for participants to share their results with family was to influence disease outcomes for their family, by delivering a "warning" that allows family to take precautions. This opinion was expressed as follows:

> *I shared [my positive result] with my brothers, sisters, and nephews and nieces, since it's a genetic condition...to let them take precaution, maybe yearly check-up after they get married or...remove the ovary etc. (P2, positive result, 50 y)*

> *I shared my positive result to let them [my family] be aware of what's going on so they can do their own research and get themselves prepared. (P3, positive result, 46 y)*

3.2.2 | Perception of family members' understanding and acceptance

Participants voiced the opinion that they were more likely to share positive results if they perceived that family members would be likely to understand the genetic results. Participants felt that family members with higher education level and emotional maturity were associated with better understanding about positive results. Participants who believed their relatives would not understand test results expressed unwillingness to share the genetic result. The following quotes show some examples:

> *They [my siblings] are not educated, especially the older sister, and they don't believe in genetic testing. I did not share my positive result with them because they are very sensitive. But my brother, he's educated so I told him [my result]. (P5, positive result, 64 y)*

> *I only shared the positive result with my elder daughter.... Because she is mature, and understanding, plus she will Google if she doesn't know certain things and wants to find out more. (P3, positive result, 46 y)*

3.3 | Unique theme: negative result category

3.3.1 | Perception of no genetic or medical implication of results

Participants who tested negative expressed that they chose not to share results because they perceived that negative results had no implications or consequence for their family, and sharing was perceived as "not helpful" or "not necessary." For example, they said:

> *This genetic information is for myself. If there's implication for my family, I would share with them.... Since its negative, I don't have to share it with them. (N1, negative result, 38 y)*

> *I don't see the need [to share negative result with my daughter] since we don't have to take action or change management. (N6, negative result, 36 y)*

3.4 | Unique theme: VUS result category

3.4.1 | Perceived ambiguity of the genetic result

Similar to the negative result participant category, when there was no future action to take, the willingness to disclose among participants who had received a VUS result decreased. In addition, participants reported that because they had difficulty converting a VUS result into meaningful information, they preferred not to share the information with their relatives so as to avoid misunderstanding and creating "false alarm." This is exemplified by the quote below:

> *Since [the result] it's unknown, I don't think there's any cause for concern.... I don't think it's worth mentioning. If they ask about it I will share, but I don't think it is worth getting alarmed over. (U4, VUS result, 27 y)*

3.5 | Proposed framework for decision-making process in sharing of genetic result

The emergent themes allowed construction of a framework to understand cognitive processes that patients undergo when making a decision about sharing genetic test results with family (Figure 1). Our model suggests that some factors predate the genetic testing event and are independent of the test result. These include perceived intimacy with family members, awareness and support of relatives, and preconceived ideas surrounding their "duty to inform." Other factors depend on the test result being positive, negative, or uncertain. The perceived presence of actionable long-term implications is a key motivator to share a positive result. Hesitancy to share is experienced when there is a perception of burden to the family members or perceived lack of medical impact. Our model suggests that the 2 optimal time

points for intervention to facilitate the sharing of genetic results with family members are (1) *before* patients make the decision to proceed with testing and (2) at the time of result delivery to the patient.

4 | DISCUSSION

In our qualitative study of Asian women receiving genetic test results for hereditary breast and/or ovarian cancer, we found that a strong family bond and perceived intimacy, greater involvement of family members in the testing process, perception of low psychological burden, and presence of actionable implications are motivators for sharing positive, negative, or uncertain results. These factors are similar to those reported among Western women.[7,8,10,24]

Our finding that intimacy of relationships (defined by participants as "a feeling of closeness" or "high level of involvement") is a key factor is similar but not equivalent to the findings from 2 Western studies[8,10] reporting that disclosure about a positive result was more likely with first-degree relatives than with second-degree relatives and lowest with third-degree relatives, among *BRCA1/2* mutation carriers. In our study, "closeness" was defined by emotional attachment rather than proximity of blood relation.

The theme of perceived burden of results on family as a barrier to information sharing from our study concurs with a prior study suggesting that sharing of results was perceived as a stressor among *BRCA1/2* mutation carriers receiving their genetic results.[11] Another study from the perspective of health care providers also recognizes patients' desire to shield relatives from distress.[25] This protective behaviour has been recognized mainly in studies exploring the sharing of positive genetic results.[25] Our study extends the observation to different result categories. Our finding suggests that, rather than the type of result, the patient's perception of their relative's ability to cope with the information was the key factor affecting the decision to share the information. The perception of relative's ability to cope could be interpreted in the emotional or cognitive domain. If they perceived an emotional burden, such as worry, sadness, or fear, patients tended not to share the genetic information. Likewise, patients preferred not to share their genetic results if they perceived disbelief, lower education level, and immaturity in their relatives.

Our theme of "duty to inform" family members has previously been reported in a study on inherited cardiac conditions.[26] We found that this feeling of "obligation" was common among participants regardless of the genetic results. We propose that the "duty to inform" tendency is motivated not only by a desire to "save someone's life"[26] as previously suggested but also by the need to update the family on outcomes of the testing. In Chinese (and the wider Asian) culture, medical decisions are often made as a family.[27] This Confucian belief or value of family-centricity may, in our view, be powerful enough to drive the decision to share genetic results, whatever they may be.[28]

In the negative and uncertain result categories, the 2 barriers (perceived lack of medical implications and uncertain implications) have policy implications for counselling. These barriers may be addressed, for example, by providing more information to patients and their family before testing. A previous study[8] found that patients who were well supported by health care professionals had a better experience when sharing their genetic result with family. Strategies can be implemented to support patient sharing of negative or uncertain genetic results to reassure and provide some certainty to family members who are not at risk.

4.1 | Clinical implications

Three phases are recognized after genetic test results are received by patients: the decision-making phase, disclosure phase, and reaction phase.[8] Our proposed framework focuses on the first, decision-making phase and offers optimal timing for effective interventions by

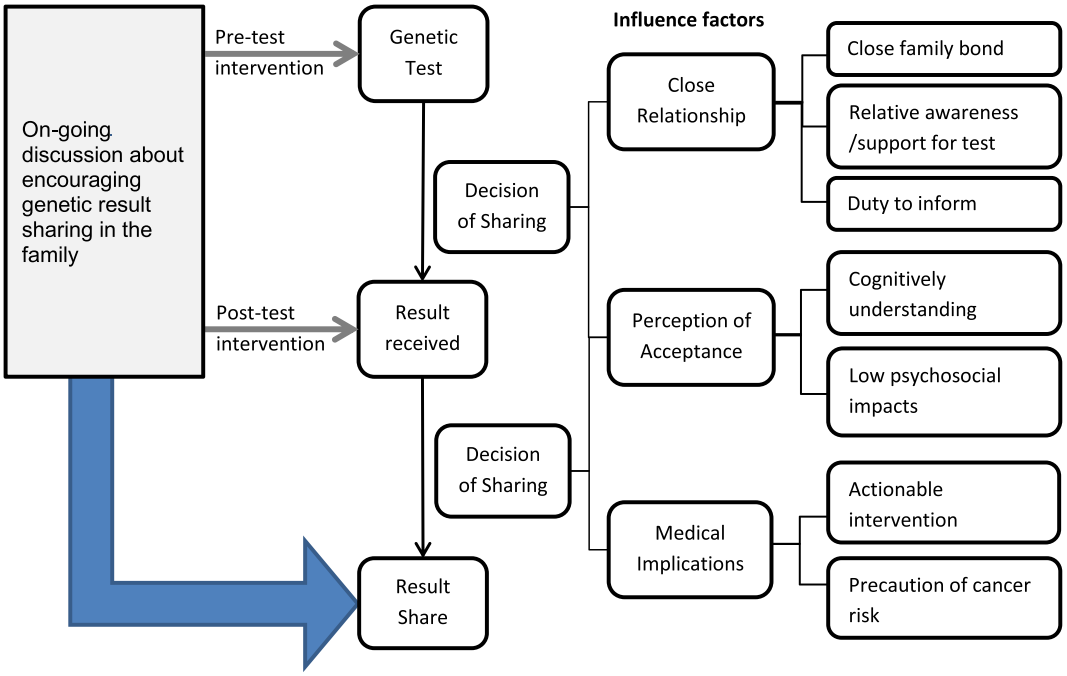

FIGURE 1 Proposed framework for Asian women's decision-making process in sharing genetic results, National Cancer Centre, Singapore, 2017

providers. We recommend that genetic counsellors and health care professionals have ongoing discussions regarding how and when to communicate genetic test results with family. In particular, they should address the periods before and after testing. On the basis of the known facilitators and barriers to result sharing, the discussion should take advantage of family closeness and involvement and cultural beliefs and norms and include clinical implications and patient's perception about the family acceptance. Patients should also be offered a pretest discussion together with family members and the option to bring their family members with them when receiving test results.

Our study contributes to a sparse literature from the perspective of Asian women. The strengths of our study include balanced sampling to represent all 3 test result categories, reaching theme saturation for each category. We also had culturally diverse representation and identified barriers and facilitators similar to those reported in Western cultures, as well as unique to the Asian culture.

4.2 | Study limitations

Our study has some limitations. Transferability of our model may be dependent on setting. Our interviews were limited to English-speaking patients. However, we may still have missed the perspectives of non-English-speaking Asian population, which represent about 17% of the population in Singapore[29] and non-Chinese populations. In addition, our study has an over-representation of Chinese over Malay and Indians because the Chinese are the predominant ethnic group in Singapore.[30] Future studies will be needed to explore specific cultural beliefs among different ethnicity and other non-English-speaking groups to distinguish among attitudes and beliefs about genetic result sharing. In addition, other socioeconomic determinants such as educational levels and income levels were not included in our study, and we were unable to determine the proportion of our participant's educational background. Our service sees predominantly patients affected with cancer as we are a physician-based referral tertiary centre; our results may not generalize to an unaffected individuals. We did not explore the distinction between intimacy and kinships during the interview. Therefore, more studies would be needed to explore which one (intimacy or kinship) is more important to the participants in sharing genetic information.

5 | CONCLUSIONS

Factors influencing the sharing of genetic information with family members among Asian women are largely similar to those reported among Western cultures. A proposed framework about sharing test information highlights 2 important time points for the implementation of interventions to enhance communication of genetic results with family.

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CONFLICT OF INTEREST

J. N. receives research funding from Astra Zeneca for ovarian cancer research. The rest of the authors declare that there is no conflict of interest.

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