Cancer Biol Med 2025. doi: 10.20892/j.issn.2095-3941.2024.0519

LETTER

Attitude towards genetic testing for breast cancer susceptibility genes and choice of prevention strategies in Chinese women with or without breast cancer

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Breast cancer (BC) is now the most common cancer and the fifth leading cause of cancer-associated mortality among women in China[1]. Germline pathogenic variants (PVs) of BC susceptibility genes, such as the well-known *BRCA1/2* genes, increase the risk of BC and other cancers (ovarian and pancreatic cancer)[2,3]. Recent studies have demonstrated substantial benefits of poly (adenosine diphosphate ribose) polymerase inhibitors in the treatment of BC patients who carry *BRCA1/2* PVs[4].

Therefore, genetic testing for BC susceptibility genes has become an essential component of health care and has implications for screening, cancer prevention and therapeutic recommendations in individuals harboring PVs[5]. A better understanding of attitudes towards genetic testing and factors affecting utilization could influence clinical practice.

The demands of genetic testing have increased worldwide due to the feasibility of new gene sequencing techniques and the improved efficacy of targeted therapy. Previous studies have demonstrated a high level of interest in genetic testing in Western countries with 60%–90% of the population considering genetic testing[6-8]. Attitudes towards genetic testing varies based on individual characteristics. Specifically, individuals with the following characteristics have favorable attitudes towards genetic testing: younger age; a higher level of genetic knowledge with higher education; and higher income[7,8]. Furthermore, there are various motivations for genetic testing in different cultures, including awareness of a child's risk for cancer, knowing and reducing one's personal cancer risk, detecting cancer early, and helping family members make better health decisions[7,9]. However, no published studies have described the attitudes towards genetic testing for BC susceptibility gene and the influence on prevention strategies in the Chinese population.

We conducted a prospective survey study to explore attitudes and the factors influencing genetic testing for BC susceptibility genes, as well as prevention strategy choices when Chinese women carry PVs. We recruited women with or without BC from the Department of Breast Surgery at Peking University International Hospital between May 2020 and March 2023. The workflow of patient selection is shown in **Figure S1**. Participants were asked to complete a questionnaire on sociodemographic characteristics, attitude towards genetic testing, and intention to implement prevention strategies.

A total of 781 hospital-based women participated in this survey, including 516 (66.1%) women with BC and 265 (33.9%) without BC. Among the 516 participants with BC, the median age was 51 years (range, 22–87 years). Most participants had a low monthly income and education level. In addition, most women were married (97.5%) and had children (91.3%), while 13.4% had a family history of BC. Among the 265 participants without BC, the median age was 36 years (range, 13–90 years).

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Received November 21, 2024; accepted January 2, 2025.
Available at www.cancerbiomed.org
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Most participants without BC had a high monthly income and education level. Among women without BC, 78.9% were married and 63.8% had children and 10.6% had a family history of BC. The participant characteristics are summarized in **Table S1**.

Attitudes towards genetic testing

Overall, 52.1% (n = 269) of the women with BC and 52.8% (n = 140) of the women without BC had favorable attitudes towards genetic testing but only 17.6% (n = 91) and 9.4% (n = 25) of women with and without BC underwent genetic testing, respectively. Interestingly, the proportion of participants who were inclined to undergo genetic testing increased if testing was free [women with BC (76.4%) *vs.* women without BC (77.7%)]. In addition, 22.9% (n = 118) of the women with BC and 14.3% (n = 38) of women without BC had received a genetic consultation (**Table S2**).

Factors influencing attitudes towards genetic testing

Younger age (P = 0.003), higher monthly income (P < 0.001), higher education level (P = 0.001), having commercial insurance (P = 0.041), awareness of genetic testing (P < 0.001), and receiving a genetic consultation (P < 0.001) were correlated with favorable attitudes towards genetic testing for BC susceptibility genes in women with BC, while having commercial insurance (P = 0.010) and awareness of genetic testing (P = 0.024) remained favorable factors in women without BC (**Table 1**). Notably, a family history of BC had no significant impact in the two populations.

Motivations and concerns for genetic testing

A high proportion (50.4%) of women with BC cited 'To know children's risk of inheriting PVs' as the motivation for genetic testing, followed by 'To know the possibility of personalized precision treatment' (28.3%). A minority of women with BC chose 'To know personal risk of BC' (11.8%) and 'To know personal risk-reducing prevention strategies' (9.5%) as the motivations for genetic testing (**Figure S2A**). In contrast, about one-third women without BC (33.6%) cited 'To know personal risk of BC' as the main motivation for genetic testing. 'To know children's risk of inheriting PVs' was the motivation for genetic testing among 27.5% of individuals. Approximately 20% of women without BC each reported 'To know personal risk-reducing prevention strategies' and 'To know the possibility of personalized precision treatment' as the motivation for genetic testing (**Figure S2A**).

Moreover, increasing the psychological burden of one's children (46.1% *vs.* 34.3%) and oneself (28.5% *vs.* 42.3%) were the top two concerns for genetic testing in women with and without BC, respectively. Cost, as an important potential factor, was the third leading concern (10%–15%) for genetic testing. Few participants worried about the impact on insurance coverage (3.1% *vs.* 5.7%) or considered genetic testing not to be important (1.6% *vs.* 2.3%) in women with and without BC (**Figure S2B**).

Choices of prevention strategies

Of women with BC when they were susceptibility gene PV carriers, 30.8% (n = 357) intended to undergo prophylactic mastectomy (**Figure 1A**). Furthermore, most women without BC when they were susceptibility gene PV carriers [53.2% (n = 141)] chose early detection, while fewer women considered risk-reducing prevention strategies with 24.9% (n = 66) choosing prophylactic mastectomy and 20.0% (n = 53) choosing chemoprevention. Only 1.9% (n = 5) of the participants chose not to utilize a prevention strategy (**Figure 1B**).

This was the first study to assess attitudes towards genetic testing for BC susceptibility genes and the choice of preventive strategies in Chinese women with and without BC. The current study revealed that > 50% of Chinese women with and without BC have favorable attitudes towards genetic testing but only 15% underwent genetic testing. The lower intention and utilization of genetic testing for BC in the Chinese population compared to Western populations might be due to regional and population differences.

Differences in motivations for genetic testing were noted between women with and without BC. Women with BC were more concerned about their children's health, while women without BC cared more about their personal cancer risk, followed closely by their children's health. It has been reported in other Asian populations that the leading motivations for genetic testing in women with BC and women who are cancer-free were to help the family understand the

Table 1 Factors influencing attitudes towards genetic testing in women with or without breast cancer

Characteristics	BC women, *n* = 516		*P* value	Non-BC women, *n* = 256		*P* value
	Unfavorable attitude, *n* (%)	Favorable attitude, *n* (%)		Unfavorable attitude, *n* (%)	Favorable attitude, *n* (%)	
Participants	247 (47.9)	269 (52.1)		125 (47.2)	140 (52.8)	
Age, years			**0.003**			0.50
≤ 40	32 (13.0)	62 (23.0)		79 (63.2)	94 (67.1)	
> 40	215 (87.0)	207 (77.0)		46 (36.8)	46 (32.9)	
Monthly income			**< 0.001**			0.10
≤ ¥ 3,500	119 (48.2)	92 (34.2)		32 (25.6)	31 (22.1)	
¥ 3,501–5,000	57 (23.1)	65 (24.2)		19 (15.2)	15 (10.7)	
¥ 5,001–10,000	49 (19.8)	54 (20.1)		38 (30.4)	31 (22.1)	
¥ 10,001–20,000	11 (4.5)	36 (13.4)		23 (18.4)	43 (30.7)	
> ¥ 20,000	11 (4.5)	22 (8.2)		13 (10.4)	20 (14.3)	
Education			**0.001**			0.90
Lack of education	11 (4.5)	3 (1.1)		2 (1.6)	2 (1.4)	
≤ Senior high school	127 (51.4)	108 (40.1)		29 (23.2)	30 (21.4)	
Technical school	43 (17.4)	50 (18.6)		23 (18.4)	25 (17.9)	
Bachelor's degree	60 (24.3)	88 (32.7)		52 (41.6)	55 (39.3)	
≥ Master's degree	6 (2.4)	20 (7.4)		19 (15.2)	28 (20.0)	
Marital status			0.49			0.30
Married	242 (98.0)	261 (97.0)		102 (81.6)	107 (76.4)	
Unmarried	5 (2.0)	8 (3.0)		23 (18.4)	33 (23.6)	
Children			0.08			0.56
No	16 (6.5)	29 (10.8)		43 (34.4)	53 (37.9)	
Yes	231 (93.5)	240 (89.2)		82 (65.6)	87 (62.1)	
Medical insurance			0.15			0.56
Fully reimbursed	5 (2.0)	12 (4.5)		6 (4.8)	5 (3.6)	
Reimbursement rate ≥ 50%	179 (72.5)	202 (75.1)		79 (63.2)	93 (66.4)	
Reimbursement rate < 50%	54 (21.9)	51 (19.0)		37 (29.6)	35 (25.0)	
No reimbursement at all	9 (3.6)	4 (1.5)		3 (2.4)	7 (5.0)	
Commercial insurance			**0.041**			**0.010**
No	193 (78.1)	189 (70.3)		83 (66.4)	71 (50.7)	
Yes	54 (21.9)	80 (29.7)		42 (33.6)	69 (49.3)	
Family history of BC			0.12			0.93
No	220 (89.1)	227 (84.4)		112 (89.6)	125 (89.3)	
Yes	27 (10.9)	42 (15.6)		13 (10.4)	15 (10.7)	

Table 1 Continued

Characteristics	BC women, *n* = 516		*P* value	Non-BC women, *n* = 256		*P* value
	Unfavorable attitude, *n* (%)	Favorable attitude, *n* (%)		Unfavorable attitude, *n* (%)	Favorable attitude, *n* (%)	
Previous cancer diagnosis			0.49			0.75
No	242 (98.0)	261 (97.0)		123 (98.4)	137 (97.9)	
Yes	5 (2.0)	8 (3.0)		2 (1.6)	3 (2.1)	
Have you heard of genetic testing for BC susceptibility genes?			**< 0.001**			**0.024**
No	135 (54.7)	100 (37.2)		70 (56.0)	59 (42.1)	
Yes	112 (45.3)	169 (62.8)		55 (44.0)	81 (57.9)	
Have you ever received a genetic consultation?			**< 0.001**			0.30
No	212 (85.8)	186 (69.1)		110 (88.0)	117 (83.6)	
Yes	35 (14.2)	83 (30.9)		15 (12.0)	23 (16.4)	

BC, breast cancer.
The bold represents *P* value with statistical significance.

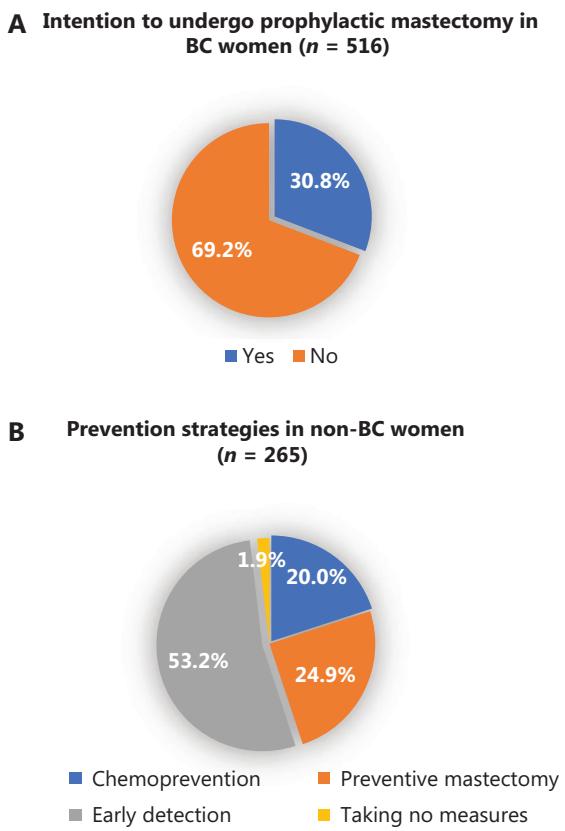


Figure 1 Participants' choice of prevention strategies when they carried breast cancer susceptibility genes pathogenic variants among breast cancer (A) and non-breast cancer women (B). BC, breast cancer.

cancer risk and to learn what to do to reduce the individual cancer risk[9].

There was a lower proportion of women [women with BC (30.8%) and women without BC (24.9%)] considering prophylactic mastectomy with a hypothetical susceptibility gene mutation in the current study compared to other studies. A study reported that > 50% of healthy young Korean women were inclined to undergo risk-reducing mastectomy if they were *BRCA1/2* mutation carriers[10]. Another study found that 30% of young women with BC indicated that their knowledge or concern about genetic risk affected their surgical decisions. Among these women, 86% of *BRCA1/2* mutation carriers and 51% of non-carriers chose bilateral mastectomy[6]. Culture differences and body image considerations might make the rate lower.

In summary, we found that > 50% of Chinese women have favorable attitude towards genetic testing. Nearly one-third of women with BC preferred to undergo prophylactic mastectomy and women without BC who carry PVs were more likely to prefer early detection. The results of the current study will facilitate genetic counseling in Chinese women.

Grant support

This work was supported by grants from the National Natural Science Foundation of China (Grant Nos. 82272932, 81974422, and 92359201).

Conflict of interest statement

No potential conflicts of interest are disclosed.

Author contributions

Conceived and designed the analysis: Yuntao Xie
Collected the data: Xue Yu, Furong Kou
Contributed data or analysis tools: Xue Yu, Furong Kou
Performed the analysis: Furong Kou
Wrote the paper: Furong Kou, Yuntao Xie.

Data availability statement

The data generated in this study are available upon reasonable request from the corresponding author.

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Cite this article as: Yu X, Kou F, Xie Y. Attitude towards genetic testing for breast cancer susceptibility genes and choice of prevention strategies in Chinese women with or without breast cancer. Cancer Biol Med. 2025; 22: 28-32. doi: 10.20892/j.issn.2095-3941.2024.0519