

Knowledge of BRCA gene among young Pakistani adults: A short report

Shahir Muhammad Ibne Rasa¹, Daler Ahmad², Mishaal Hasan³, Agha Hashim Raza Khan⁴, Minahil Farhan⁵, Farzad Effan⁶

Abstract

A total of 271 young Pakistani adults responded to a self-designed multiple choice-based questionnaire ($\alpha = 0.83$) which was then used to assess the levels of knowledge regarding breast cancer susceptibility (BRCA) gene mutation. Overall knowledge levels were assessed using the sum score of each response; any possible significance between knowledge scores and educational backgrounds as well as gender were also tested. The results show that 161 (63.9%) of the sample population had awareness about BRCA gene mutation. Knowledge scores were comparable across both groups (medical and non-medical educational backgrounds) with 20 (13.8%) of medical and 14 (13.5%) of non-medical respondents demonstrating a high level of knowledge about the BRCA gene mutation and its testing. Neither gender nor educational background had a significant influence on knowledge scores. The results from this report suggest that awareness regarding BRCA gene was adequate, while knowledge levels were noted to be poor among the sample population.

Keywords: Breast cancer, Pakistan, Awareness, Knowledge, Gene.

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Introduction

Breast cancer is the most prevalent cancer in Pakistani women, accounting for 47.3% of all cancer cases, while ovarian cancer accounts for 4.4% of all cases, making it the third most prevalent cancer for women in the country.¹ In normal healthy individuals, breast cancer susceptibility (BRCA) genes function as tumour suppressor genes preventing uncontrolled cell growth and the development of cancers. However, mutations of these genes paradoxically predispose the patients to developing such malignancies.

Studies conducted globally have shown that 5-10% of breast cancer cases and at least 10% of all ovarian cancer

^{1-3, 5, 6}Lahore Medical and Dental College, Lahore, Pakistan; ⁴Azra Naheed Medical College, Lahore, Pakistan.

Correspondence: Shahir Muhammad Ibne Rasa.
e-mail: shahir.ibnerasa@gmail.com

ORCID ID: 0009-0005-5414-475X

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cases are hereditary.² Among hereditary ovarian cancers, BRCA gene mutation carriers accounted for more than 90% of such cases.³ Individuals with BRCA gene germline mutations have been seen to have an estimated cumulative risk of up to 87% for developing breast cancer and 44% for developing ovarian cancer by the age of 70.⁴ Men and women carrying the BRCA2 gene mutations have also been observed to have a higher risk of developing stomach cancers, pancreatic cancers, gallbladder and bile duct cancers, malignant melanomas, and prostatic carcinomas.⁵ Even among breast cancer patients, BRCA1 gene mutation carriers were much more likely to develop the aggressive triple-negative subtype than non-carriers.⁶ According to studies conducted in Pakistan, Rashid et al. (2006 & 2019) observed the prevalence of BRCA1 or BRCA2 mutations to be 55.4% in families with a history of breast and ovarian cancer, 27.4% for families with two or more cases of breast cancer, 12.3% for families with a single case of early-onset breast cancer, and 9.0% for single cases of early-onset ovarian cancer (≤ 45 years).^{7, 8} Rashid et al. (2019) also concluded that BRCA1/2 mutations were responsible for one in five cases of male breast cancer.⁷ Farooq et al. (2011) in their study attributed mutations in the BRCA gene to account for 11% of the total number of cases of both hereditary and sporadic breast and ovarian cancer.⁹ Therefore, recent trends suggest that BRCA gene mutations account for a significant proportion of hereditary as well as early-onset breast and ovarian cancer cases in Pakistan.

As a result, it becomes extremely important to understand the knowledge individuals have, both from the susceptible population as well as those associated with the medical profession, regarding this gene to avoid lapses in health care. A literature search revealed no local research on this topic and while some global studies were present that were relevant, none of them targetted the specific age range of this report and also did not explore the possible links between the effect of medical vs non-medical educational backgrounds on awareness and levels of knowledge.

Methods and Results

After obtaining ethical approval from the institutional review board of Lahore Medical & Dental College, data for this report was collected from July 2021 to October 2021. The sample population was set to include both male and female young Pakistani adults from various medical and

non-medical educational backgrounds. The inclusion criteria stated that respondents must be between the targetted age range of 18 to 30 years.

The tool for data collection was a self-designed multiple choice-based questionnaire which was designed with the assistance of the Pathology department of the institution, following AMEE Guide No. 87 and The Checklist for Reporting Results of Internet E-Surveys (CHERRIES).^{10,11} The reliability of the knowledge section of the instrument was checked using IBM SPSS version 26 item analysis yielding a Cronbach Alpha of 0.83. Non-probability convenience sampling method was chosen and the questionnaire was administered as a voluntary open survey in English language via the internet using Google Forms. Respondents were limited to one response per email address and were informed about the research and its specifics before filling out the questionnaire. No data that could be used to identify any of the respondents was collected.

The questionnaire consists of a total of 24 questions, of which 11 questions directly test the respondent's knowledge and awareness of the BRCA gene mutations. The remaining questions are related to demographic characteristics and some open-ended questions to explore possible limitations to broader BRCA1 and 2 gene testing among the general population, according to the respondent.

Respondents were then scored based on their responses to the 11 questions relating to the knowledge and awareness regarding BRCA gene mutations. Each correct response was awarded a score of 1, each incorrect response was awarded a score of 0. The "I don't know" option, added to minimise guessing, was also treated as incorrect and scored as 0. The expected knowledge and awareness scores were between 0 and 11 and the obtained number scores were converted into percentage scores. Each respondent's percentage score was then assessed based on Bloom's cut-off point.¹² Those who obtained a score of 80% and above

were considered to have a high level, while those with scores between 79% and 61% were considered to have a moderate level. The scores of 60% and below were considered low level.

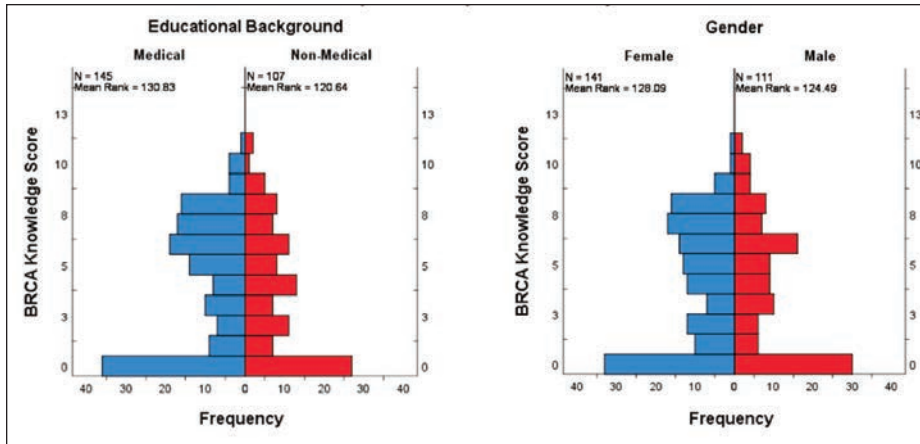
Data compilation and analysis was done using IBM SPSS version 26. Descriptive statistics were used to define the socio-demographic characteristics of the sample population. Normality of the data was assessed using the Kolmogorov Smirnov test. The non-parametric Mann-Whitney U test was used to determine any possible statistical significance between the knowledge scores of the respondents and their educational backgrounds as well as gender. A p -value ≤ 0.05 was considered to be statistically significant for all variables.

A total of 271 responses were submitted. After excluding data from 19 respondents who either did not fall within the specified age range of 18 to 30 years or provided incomplete information, the final sample consisted of 252 participants. Among the final sample, 141 (56.0%) were females, with a mean age of 22.58 ± 2.08 years, and 145 (57.5%) were from a medical educational background, which included people who were obtaining/had obtained degrees in medicine, dentistry, or allied health sciences. The remaining respondents included, but were not limited to, people with educational backgrounds in arts, business, engineering, finance and accounting, law, and social sciences.

From the total sample population, 161 (63.9%) of the respondents answered "Yes" when asked if they were aware of the BRCA gene mutation. Of the 145 respondents with a medical education background, 120 (82.8%) were aware, while among the 107 non-medical respondents only 41 (38.3%) stated they were aware of the gene mutation. When asked how much the respondents thought they knew about the BRCA gene on a scale of 0 - 5, the majority, i.e. 77 (30.6%) of the sample population, selected a score of "0". Of the total sample, 102 (40.5%) stated that their source of information was their medical educational

Table: BRCA Questionnaire - Knowledge Section.

Questions	Correct	Incorrect	I don't know
1. Is the BRCA 1/2 gene present normally in everyone?	89 (35.3%)	36 (14.3%)	127 (50.4%)
2. Can the BRCA 1/2 gene mutation be transmitted genetically?	136 (54.0%)	16 (6.3%)	100 (39.7%)
3. According to your knowledge, is there any criteria for getting tested?	126 (50.0%)	13 (5.2%)	113 (44.8%)
4. Are you aware at what age a person should get tested for the BRCA 1/2 gene mutation?	75 (29.8%)	61 (24.2%)	116 (46.0%)
5. Does the BRCA 1/2 gene mutation only affect females?	85 (33.7%)	61 (24.2%)	106 (42.1%)
6. Does everyone with BRCA 1/2 gene mutation get cancer?	98 (38.9%)	35 (13.9%)	119 (47.2%)
7. Are individuals from certain geographical regions or populations at a higher risk of developing the BRCA 1/2 gene mutation?	95 (37.7%)	18 (7.1%)	139 (55.2%)
8. Is it possible to get Breast/Ovarian cancer without BRCA 1/2 gene mutation?	111 (44.0%)	18 (7.1%)	123 (48.8%)
9. Is it possible to get other forms of cancer with the BRCA 1/2 gene mutation?	82 (32.5%)	23 (9.1%)	147 (58.3%)
10. According to your knowledge, are there any risks associated with the BRCA 1/2 gene mutation test?	60 (23.8%)	49 (19.4%)	143 (56.7%)
11. Is the BRCA 1 gene mutation more dangerous than BRCA 2?	44 (17.5%)	19 (7.5%)	189 (75.0%)



n=252		n=252	
Mann-Whitney U	8385.000	Mann-Whitney U	7602.000
Wilcoxon W	18970.000	Wilcoxon W	13818.000
Test Statistic	8385.000	Test Statistic	7602.000
Standard Error	565.824	Standard Error	568.298
Standardized Test Statistic	1.109	Standardized Test Statistic	-0.393
Asymptotic Sig. (2-sided test)	0.267	Asymptotic Sig. (2-sided test)	0.694

Figure: Independent-samples Mann-Whitney U test result. institute or being medical practitioners.

Table shows the percentages of the answers to the knowledge and awareness section of the questionnaire. For 9 out of the 11 questions in this section, "I don't know" was the most frequent response. Among the total sample, 161 (63.9%) of the respondents had a low level of knowledge regarding the BRCA gene mutation, while 57 (22.6%) had a moderate level. Those with a high level of knowledge accounted for 34 (13.5%) of the respondents.

Results of the Mann-Whitney U test (Figure) revealed knowledge scores in respondents with a medical education (median=5.00) did not differ significantly from respondents with a non-medical education (median=4.00) with $p=0.27$. While respondents with a non-medical background accounted for a larger percentage of participants with low knowledge levels when compared with those from a medical background, 76 (71.0%) vs 85 (58.6%) respectively, similar distribution was seen for high knowledge levels, 14 (13.1%) vs 20 (13.8%) respectively. No statistically significant difference was observed with gender (Female median=4.00, Male median=4.00) as well with $p=0.69$.

Regarding limitations to BRCA gene testing among the general population, 191 (75.7%) of the respondents felt lack of awareness/incomplete knowledge was a factor, while 146 (57.9%) felt the high cost of genetic testing played a role. The respondents also felt that the social stigma and the psychological stress attached with potentially testing positive were other reasons for lack of testing, accounting for 73 (29.0%) and 65 (25.7%) of the sample population

respectively. When asked about effective methods to raise awareness regarding BRCA gene mutations, 194 (76.9%) opted for social media, 149 (59.1%) for government programmes and awareness campaigns, 137 (54.4%) for integration into the educational curriculum, and 95 (37.7%) opted for awareness through news outlets. 226 (89.7%) of the respondents wanted to know more about the BRCA gene mutation and its testing.

Discussion

The aim of this report was to assess the awareness and level of knowledge of young Pakistani adults with regards to BRCA gene, their role in breast and ovarian cancers and its genetic testing. The results showed that among the sample population 161 (63.9%) stated that they were

aware of the BRCA gene and its testing; however, knowledge about the BRCA genes was poor as only 34 (13.5%) of the respondents fell into the category of high level as defined by this report. These results were fairly in line with other studies conducted worldwide in which awareness of the genes and its testing was high but actual knowledge about them was on the lower side.¹³⁻¹⁵

Furthermore, neither gender nor educational background seemed to play a significant role in the levels of knowledge and awareness. This was contrary to the authors' expectations of respondents from a medical background having higher levels of knowledge and awareness of the topic, especially since other studies conducted worldwide show that medical professionals overall have a higher level of knowledge and awareness about BRCA and its genetic testing.^{16,17} One possible explanation for this could be the inclusion of dental professionals and individuals associated with allied health sciences to the medical education demographic as both are not required to know the specifics of breast cancer and its genes as part of their course outlines. Another explanation could be the lack of experience and clinical exposure when it comes to breast/ovarian cancer and its genetic testing as the sample population included relatively younger individuals. This seems to be consistent with one study conducted in Malaysia that tested the knowledge and attitudes regarding BRCA genetic testing among Malaysian nurses in which the authors observed that years of experience substantially impacted knowledge levels, with junior

nurses having less knowledge when compared to nurses with 10 or more years of experience.¹⁸

The results of this report should, however, be interpreted keeping in mind the following limitations. The study population was selected using a non-probability convenience method instead of a random method of sampling. This, coupled with the relatively smaller sample size, may affect the ability of the results of this report to be generalised. Additionally, despite the efforts of the authors, there was no way to adequately control and estimate the degree of guessing from the respondents due to the "Yes" and "No" nature of the questionnaire. Furthermore, due to a lack of research on this topic among the Pakistani population, appropriate comparisons of results were not possible.

Conclusion

The results from this report suggest that while awareness regarding BRCA gene among young Pakistani adults may be adequate, especially among those with a medical educational background, knowledge about the gene and its testing, however, was poor across the entire sample population regardless of educational background and gender. An overwhelming majority of the study participants displayed an interest in wanting to learn more about this topic with social media as their preferred source for raising awareness. Considering how prevalent breast and ovarian cancers are among the Pakistani population, it becomes absolutely essential to improve the knowledge of the general population on this topic which will encourage individuals at high risk to seek professional advice which in-turn will help in early diagnosis and management of this life-altering disease.

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Disclaimer: None.

Conflict of Interest: The authors of this study would like to declare as a potential conflict of interest that the chairperson of the IRB, Prof. Abdul Majeed Chaudhry, was also the principal of the institute in which the authors were enrolled at the time of obtaining ethical approval for this study.

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References

1. The Punjab Cancer registry [Internet]: Punjab Cancer registry report 2020.[Online] [Cited 2022 May 19]. Available from: URL: http://punjabcancerregistry.org.pk/reports/PCR_2020.pdf
2. Carroll JC, Cremin C, Allanson J, Blaine SM, Dorman H, Gibbons CA, et al. Hereditary breast and ovarian cancers. *Can Fam Physician*. [Online] [Cited 2022 June 15]. Available from: URL: <https://www.cfp.ca/content/54/12/1691>.
3. Russo A, Calò V, Bruno L, Rizzo S, Bazan V, Di Fede G. Hereditary ovarian cancer. *Crit Rev Oncol Haematol*. 2009; 69:28-44. <https://doi.org/10.1016/j.critrevonc.2008.06.003>.
4. Ford D, Easton DF, Bishop DT, Narod SA, Goldgar DE. Risks of cancer in BRCA1-mutation carriers. *Breast Cancer Linkage Consortium*. *Lancet*. 1994; 343:692-5. doi:10.1016/s0140-6736(94)91578-4.
5. Breast Cancer Linkage Consortium. Cancer risks in BRCA2 mutation carriers. *J Natl Cancer Inst*. 2022; 91:1310-6. doi:10.1093/jnci/91.15.1310
6. Chen H, Wu J, Zhang Z, Tang Y, Li X, Liu S, et al. Association Between BRCA Status and Triple-Negative Breast Cancer: A Meta-Analysis. *Front Pharmacol*. 2018; 9:909. doi:10.3389/fphar.2018.00909
7. Rashid MU, Muhammad N, Naeemi H, Khan FA, Hassan M, Faisal S, et al. Spectrum and prevalence of BRCA1/2 germline mutations in Pakistani breast cancer patients: results from a large comprehensive study. *Hered Cancer Clin Pract*. 2019; 17:27. doi:10.1186/s13053-019-0125-5.
8. Rashid MU, Zaidi A, Torres D, Sultan F, Benner A, Naqvi B, et al. Prevalence of BRCA1 and BRCA2 mutations in Pakistani breast and ovarian cancer patients. *Int J Cancer*. 2006; 119:2832-9. doi:10.1002/ijc.22269
9. Farooq A, Naveed AK, Azeem Z, Ahmad T. Breast and Ovarian Cancer Risk due to Prevalence of BRCA1 and BRCA2 Variants in Pakistani Population: A Pakistani Database Report. *J Oncol*. 2011; 2011:632870. doi:10.1155/2011/632870
10. Artino AR Jr, La Rochelle JS, Dezee KJ, Gehlbach H. Developing questionnaires for educational research: AMEE Guide No. 87. *Med Teach*. 2014; 36:463-74. doi:10.3109%2F0142159X.2014.889814
11. Eysenbach G. Correction: Improving the Quality of Web Surveys: the Checklist for Reporting Results of Internet E-Surveys (CHERRIES). *J Med Internet Res*. 2012; 14:e8. doi:10.2196%2Fjmir.2042
12. Bloom BS. Learning for Mastery. Instruction and Curriculum. Regional Education Laboratory for the Carolinas and Virginia, Topical Papers and Reprints, Number 1. [Online] [Cited 2022 July 11]. Available from: URL: <https://eric.ed.gov/?id=ED053419>.
13. Amin TT, Al-Wadaani HA, Al-Quaimi MM, Aldairi NA, Alkhateeb JM, Al-Jaafari AA. Saudi women's interest in breast cancer gene testing: possible influence of awareness, perceived risk and socio-demographic factors. *Asian Pac J Cancer Prev*. 2012; 13:3879-87. doi:10.7314/apjcp.2012.13.8.3879
14. Kinney AY, Croyle RT, Dudley WN, Bailey CA, Pelias MK, Neuhausen SL. Knowledge, attitudes, and interest in breast-ovarian cancer gene testing: a survey of a large African-American kindred with a BRCA1 mutation. *Prev Med*. 2001; 33:543-51. doi:10.1006/pmed.2001.0920
15. Young AL, Butow PN, Vetsch J, Quinn VF, Patenaude AF, Tucker KM, et al. Family Communication, Risk Perception and Cancer Knowledge of Young Adults from BRCA1/2 Families: a Systematic Review. *J Genet Couns*. 2017; 26:1179-96. doi:10.1007/s10897-017-0125-4
16. Bellcross CA, Kolor K, Goddard KA, Coates RJ, Reyes M, Khoury MJ. Awareness and utilisation of BRCA1/2 testing among U.S. primary care physicians. *Am J Prev Med*. 2011; 40:61-6. doi: 10.1016/j.amepre.2010.09.027

17. Van Riel E, Wárlám-Rodenhuis CC, Verhoef S, Rutgers EJ, Ausems MG. BRCA testing of breast cancer patients: medical specialists' referral patterns, knowledge and attitudes to genetic testing. *Eur J Cancer Care (Engl)*. 2010; 19:369-76. doi:10.1111/j.1365-2354.2008.01065.x
 18. Liu KT, Wan Rosli WR, Yusuf A, Lean Keng S. Malaysian nurses' knowledge and attitudes regarding BRCA genetic testing. *Belitung Nurs J*. 2021;7:493-9. doi:10.33546/bnj.1802.
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Author Contribution:

AA: Introduction, methodology, result analysis and discussion.

AY: Review introduction, methodology, result analysis, discussion and helped in conclusion.