Family History of Cancer, Trend of Genetic Counselling and Screening in Karachi: A Survey among Students of Jinnah Sindh Medical University

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Abstract

Background: Family history is a significant tool to identify at risk population. For cancer, about 22% positive family history has been reported which includes breast, ovarian, endometrial, prostate and colorectal carcinoma. Documentation of family history of cancer facilitates the decision of screening test and counselling in susceptible subjects.

The aim of the present study was to observe the trend of genetic counseling and screening tests in subjects with documented positive family history of cancers and to identify the knowledge of familial cancers and related hereditary mutations among MBBS, BDS and Pharm D students.

Methods: A cross sectional, questionnaire-based survey was conducted at Jinnah Sindh Medical University from June to September 2019. A total of 162 MBBS, BDS and PHARM D students were included in the study. Data were analyzed by SPSS version 22.

Result: Approximately 42(25.9%), 67(41.4%) and 60(37.0%) participants had positive family history of cancer in first, second- and third-degree relatives, respectively. Most commonly reported familial malignancy 45(27.8%) was breast carcinoma. BRCA gene screening and mammography was prescribed to 6(3.7%) and 20(12.3%) subjects respectively with positive familial history of breast cancer. Family history of hereditary nonpolyposis colorectal carcinoma was positive in 16(9.9%). However, colonoscopy was prescribed to 7(4.3%) subjects. About 44(27.2%) subjects reported documentation of family history and 36(22.2%) of participants were advised genetic counselling or referred to a genetic counsellor (p= 0.000). Students of MBBS were more aware of familial cancers and hereditary mutations in comparison to dentistry and pharmacy students.

Conclusion: The most common reported familial malignancy was breast carcinoma and the least frequent was retinoblastoma. Documentation of family history of cancer, advised screening and genetic counselling was found to be inadequate in our clinical setup. Students of MBBS had a good knowledge of familial cancer and related hereditary mutations in comparison to dentistry and pharmacy students.

Key words: cancer; family history; screening; genetic counselling; documentation.

Introduction

Cancer is the primary cause of mortality in high income states and an ultimate cause of mortality in middle and low-income countries (1). It is estimated that by 2020 the incidence of cancer would constitute 10 million deaths and 15 million new cases per year (2). Proportion of the commonly diagnosed malignancies varies globally (3).

Family history is a health risk element for a number of diseases and has acquired global importance in genetics science for preventive medicine and public health. Reported positive family history was 22% for cancers, inclusively breast, ovarian, endometrial, prostate and colorectal carcinomas (4). In addition to environmental factors, positive family history and associated genetic mutations have a predominant role in development of cancer risk. Researchers have used family history of first-degree relatives as a marker for hereditary risk of cancer. Family history identifies outcomes of genetic mutations, environment factors and associated life style. Individuals with first-degree relatives are more prone to suffer from malignancies in comparison to general population (5).

Specific genetic mutations can multiply risk of certain cancers. For instance, hereditary non polyposis colorectal carcinoma is associated with mutated MLH1 and MSH2 genes. These mutated genes elevate the risk in family members up to 80%. In a similar manner, mutated BRCA1 and BRCA2 genes constitute 5–10% of breast and ovarian cancers and associated with familial risk of these malignancies (6). Breast cancer is the most common cancer of females in Karachi, Pakistan. However, mutations of BRAC1, BRAC2 have not been sufficiently studied in the local population. Cancer control programs emphasizing on population screening, routine breast clinical checkup of females and control of breast cancer are required (7). Cancers of breast, ovary, uterus and colorectal region are associated with familial risk and a screening test may be required for first- and second-degree relatives (8).

Cancer risk assessment includes documentation of history, identification of susceptible subjects, cancer screening recommendation and genetic screening test (9). Documentation and assessment of family history is a primary step which a family physician can take in reporting candidates who will obtain advantage from screening. Family physicians should emphasize on family history documentation and risk factors assessment (10). Studies demonstrate that family history of malignancy is not only significant in identifying first-degree relatives but second and occasionally third-degree relatives of cancer patients may also require screening and genetic counseling (11).

Furthermore, genetic education, counseling and screening are essential in cases with positive family history with genetic susceptibility to a particular cancer. Genetic counseling is required before performance of genetic screening. Subjects prone to inherited mutations and cancer syndromes should seek genetic counseling that will be useful in evaluation of risk of underlying cancer and need for preventive medicine and screening test (12).

The current study was designed to identify the prevailing clinical approach towards recording of family history of cancer, genetic counseling and screening tests in the relatives of cancer patients. Furthermore, this survey may prove to be a useful tool in assessing knowledge of familial cancers and related hereditary mutations among the medical students of Jinnah Sindh Medical University.

Materials and Methods

Study design, period and area: A student based cross sectional study was conducted in Jinnah Sindh Medical University (JSMU) during the period of June to August 2019. JSMU is one of the oldest and prestigious medical institutes of Karachi, which offers undergraduate program in Bachelor of Medicine and Bachelor of Surgery (MBBS), Bachelors of Dental Surgery (BDS) and Doctor of Pharmacy (PHARM-D) among a few others. Authorization to conduct the study was ascertained by Institutional Review Board of JSMU (JSMU/IRB/2019/-200).

Study population: Undergraduate medical students of Sindh Medical College (SMC), Sindh Institute of Oral Health Sciences (SIOHS) and Institute of Pharmaceutical Sciences (IPS) of JSMU with age range of 19 to 25 years were included in the current study. Students of JSMU enrolled in non-medicinal fields were excluded.

Sample size and sampling techniques: Sample size was calculated on open EPI software using population of 2550 (total population of above-mentioned institutes of JSMU was 2550, MBBS= 1750, BDS= 300, PHARM-D= 500). Anticipated % frequency was 12.9% (8), confidence level was 95% with 5% margin of error. With this calculation, the final sample size obtained was 162. Simple random sampling technique was applied to select the study subjects. All volunteers were asked to fill out the questionnaire in their free time within the university premises. Written informed consent was obtained from participants.

Data Collection: A self-administered structured questionnaire was used to collect the data. It consisted of open and close ended multiple-choice questions that have met the objectives of the study. Clinical oncology consultant assessed validity of the content of questionnaire. Furthermore, the questionnaire was pretested on 15 students (excluded in sampling) to certify the accuracy of content. The questionnaire was distributed to respondents. Participants were guaranteed the provision of anonymity. Theirs f part includes demographic data. The second part contains questions that were structured to assess the knowledge of students regarding hereditary cancers and particular genes mutated in familial cancers. The third part assessed prevalence of familial cancers in relatives of participants. The fourth part emphasizes on cancer history taking attitude from their family physicians, subsequent routine examinations, genetic counselling and screening.

We assigned one point (1) to a correct answer and zero (0) for don't know or an incorrect answer.

Data Analysis: Collected data was entered, coded and processed in SPSS Statistics version 22.0 for analysis. The current study is a descriptive study. Therefore, results were calculated in percentages. Chi-square test was applied to observe the association of different variables. A $P < 0.05$ was considered as statistically significant.

Results

A total of 162 medical students were interviewed. Distribution of medical students from the disciplines of MBBS, BDS and Pharm D was equal. Mean age was 21.3±1.52. Males were 38(23.5%) and females were 124 (76.5%). Approximately 42(25.9%), 67(41.4%) and 60(37.0%) participants have positive family history of cancer in first, second- and third-degree relatives, respectively. Moreover, 44(27.2%) participants reported that family history with respect to malignancy was documented by general physicians. Subsequently, 49(30.2%) of participants were advised for screening of cancers and 36(22.2%) were referred for genetic counselling. (Table 1)

Prescription of genetic counselling and screening test by physicians in subjects with positive family history of particular cancer was questioned. Comparative analysis of documentation of family history and advised genetic counselling and screening revealed a significant association. About 44(27.2%) subjects reported documentation of family history and 36(22.2%) of participants were advised genetic counselling or referred to a genetic counsellor (p= 0.000). Knowledge of familial cancers and related hereditary mutations among MBBS, BDS and Pharm D students was evaluated. Medical students had better knowledge in comparison to dentistry and pharmacy students (p=0.006). (Table 3)

Table 1: Family history of cancer, trend of genetic counseling and screening tests in subjects with documented positive family history of cancers. (n= 162)

Variables	Responses n (%)
Do your first-degree relatives have a history of cancer?	42(25.9%)
Do your second-degree relatives have a history of cancer?	67(41.4%)
Do your third-degree relatives have a history of cancer?	60(37.0%)
Which type of familial cancer did they suffer from?	
Breast	45(27.8%)
Ovarian	13(8.0%)
Prostate	19(11.7%)
Colorectal	16(9.9%)
Endometrial	6(3.7%)
Retinoblastoma	5(3.1%)
Medullary thyroid cancer	9(5.6%)
Total	110(67.9%)
Documentation of family history, advice of genetic counseling and screening test:	
Has physician ever documented your medical and family history concerning cancer?	44(27.2%)
Has physician advised genetic counseling or referred you, your relatives or family members to a genetic counselor considering the positive familial risk of cancer?	36(22.2%)
Has physician ever explained to you or any of your relatives about routine examination and screening relevant to your cancer family history?	49(30.2%)
Opinion regarding modes of improved attitude and practice of genetic counseling and screening	
Online familial cancer registration application Advertisement on media to create mass awareness Awareness programs in educational institution Family physician should document familial history and do counseling for familial risk of cancer	27(16.7%) 46(28.4%) 50(30.9) 39(24.1%)

Table 2: Advised genetic counselling and screening test by Physicians in subjects with positive family history of particular cancer. (n=162)

Diagnosed cancer types N (%)	Advised cancer screening	N (%)	P value*
Breast 45(27.8%)	Breast cancer screening: Breast self-examination Mammography Breast MRI BRCA screening No advised screening	 17(10.5%) 20(12.3%) 3(1.9%) 6(3.7%) 30(18.5%)	0.000
Colorectal 16(9.9%)	HNCP screening: Stool test for occult blood Sigmoidoscopy Colonoscopy CT Colonography No advised screening	 9(5.6%) 2(1.2%) 7(4.3%) 4(2.5%) 30(18.5%)	0.000
Ovarian 13(8.0%)	Ovarian cancer screening: CA 125 Screening Transvaginal Ultrasound No advised screening	 6(3.7%) 8(4.9%) 32(19.8)	0.000
Endometrial 6(3.7%)	Endometrial cancer screening: Transvaginal Ultrasound Endometrial biopsy No advised screening	 3(1.9%) 12(7.4%) 26(16.0%)	0.014
Retinoblastoma 5(3.1%)	Retinoblastoma screening: Regular eye examination from birth to 3 years No advised screening	 6(3.7%) 31(19.1%)	0.000
Prostate 19(11.7%)	Prostate cancer screening: Digital rectal examination Prostate specific antigen No advised screening	 3(1.9%) 15(9.3%) 31(19.1%)	0.000
Thyroid 9(5.6%)	Thyroid cancer screening: Ultrasound FNAC MEN2 screening No advised screening	 3(1.9%) 5(3.1%) 6(3.7%) 28(17.3%)	0 .000

Table 3: Knowledge of familial cancers and related hereditary mutations among MBBS, BDS and Pharm D students. (n=162)

	Students of various sub-specialties			
Knowledge Variables:	MBBS n (%)	BDS n (%)	Pharm D n (%)	P value
Do you believe cancer can be hereditary?	52(38.2%)	44(32.4%)	40(29.4%)	0.006
Which of the following cancers are hereditary?				
Prostate Cancer	33(44.6%)	23(31.3%)	18(24.3%)	0.000
Breast Cancer	48(40.0%)	30(27.5%)	31(28.4%)	0.001
Ovarian Cancer	43(44.8%)	27(28.1%)	26(27.1%)	0.001
Colorectal Cancer	39(54.9%)	18(25.4%)	14(19.7%)	0.000
Thyroid Cancer	29(37.7%)	23(29.9%)	25(32.5%)	0.015
Endometrial Cancer	41(46.1%)	26(29.2%)	22(24.7%)	0.001
Retinoblastoma	35(49.3%)	17(23.9%)	19(26.8%)	0.000
Mutations: Genes mutated in familial cancers				
Breast: BRCA1 and2	41(50.0%)	27(32.9%)	14(17.1%)	0.000
Prostate: BRCA1,2 and HOXB13	18(54.5%)	4(12.1%)	11(33.3%)	0.013
Colorectal: APC	27(56.3%)	10(20.8%)	11(22.9%)	0.000
Endometrial: PTEN, PIK3CA, TP53	23(76.7%)	1(3.3%)	6(20.0%)	0.000
Ovarian: BRCA1,2 and TP53	32(51.6%)	17(27.4%)	13(21.0%)	0.000
Medullary thyroid: MEN	24(63.2%)	7(18.4%)	7(18.4%)	0.000
Retinoblastoma: RB	29(57.3%)	15(27.8%)	10(18.5%)	0.002

Discussion

Family history is a health risk element for cancer. Genetic education, counselling and screening are essential in cases of positive family history with genetic susceptibility to a particular cancer. In the current study a considerable number of responses from students showed varying frequency of malignancy in first, second- and third-degree relatives. The most commonly reported malignancy was breast cancer 27.8%. A study conducted in Pakistan agrees by reporting 25.6% of cancer patients who had positive family history of malignancy with breast cancer as the most frequent type (13). However, oral cancer has also been frequently reported (14). Prostatic carcinoma was reported as 11.7% in our survey. In Europe and North America however, the reported frequency (39%) was much higher (15). Another study conducted on a large scale in Pakistan recorded the following frequencies of malignancies including prostatic 2.4%, colorectal 5.9%, ovarian 2.5%, thyroid 1.4% and endometrial 4.8% carcinomas (16). Proportion of familial medullary thyroid cancer in our survey was 5.6% which is comparable with a past study suggesting that familial medullary thyroid cancers are underestimated (17). These data enlighten the region wide distribution of various malignancies, globally. Physicians can play a fundamental role in genetic risk evaluation, counselling and screening (18). According to our findings, 27.2% and 30.2% participants acknowledged documentation of family medical history and subsequent screening tests of cancer by general physicians, respectively. Mammography and colonoscopy were advised in a small number i.e. 12.3% and 2.5% of family members of breast and colorectal cancer patients, respectively. Moreover, only a few respondents related to ovarian cancer patients were advised CA125 screening (3.7%) and transvaginal ultrasound (4.9%). Scheuner et al. is in agreement with inadequate documentation of family history in clinics by primary care practitioners (19). However, research published in the American Journal of Oncology showed a comparatively higher frequency (42.7%) of patients being referred for genetic counselling and testing with a positive family history of breast and colon cancer (20). Another study reported advised colonoscopy in 50.4% of participants (21). This comparative analysis shows the difference in approach and practices of general practitioners between advanced and less developed countries.

Studies on ovarian cancer revealed about 11%–65% risk of acquiring this malignancy in first- and second-degree relatives, respectively. Genetic mutations in BRCA1, BRCA 2 genes, Lynch II syndrome, and Li-Fraumeni syndrome have shown significant association. Currently in Pakistan, CA 125 levels and transvaginal ultrasound are being advised for ovarian cancer screening. However, both of these are not considered as satisfactory screening markers due to a low sensitivity and specificity (22).

Conclusion

The most common reported familial malignancy was breast carcinoma and the least frequent was retinoblastoma. Documentation of family history of cancer, advised screening and genetic counselling was inadequate in our clinical setup. Students from the discipline of medicine possessed comparatively better knowledge of familial cancer and related hereditary mutations in comparison to dentistry and pharmacy students.

Recommendations

A mobile application inquiring family history of cancer from individuals may help physicians and oncologists to identify at risk population. This may also be useful in preventing a large number of deaths occurring due to familial cancer each year.

Limitation

Study population included students from only one public sector university.

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Ethical consideration: Study is ethically approved by Institutional review board (IRB) of JSMU.

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