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EVALUATING FAMILY HISTORY QUESTIONNAIRE VERSUS FAMILY PEDIGREE INTERVIEW FOR GENETIC DISORDERS IN PRIMARY HEALTH CARE, QASSIM REGION, SAUDI ARABIA

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ABSTRACT

Introduction: The use of family history as screening tool (pedigree or questionnaire) has been shown to increase the likelihood of detecting a patient at high risk of developing an inherited medical condition by 20% compared with medical record review alone. **Objectives:** 1.To assess the ability of the current used Family History Questionnaire, to identify genetic risk in the study population 2. assess the ability of family pedigree interview with patient, to identify genetic risk and compare the ability to identify genetic risk between family pedigree interview and currently used questionnaire. **Methodology:** Institutional based descriptive study, compared a random sample of reviewer currently used family history questionnaire in Primary Health Care (PHC) with standardized family history, pedigree questionnaire for evaluating risk of common genetic diseases among Qassim population. Same participant fills both questionnaire during the period from March 2015 to August 2015 among 289 participants. **Results:** currently used family file folders revealed that nearly 61.9% of total files had a family history of diabetes and 53% had a history of hypertension also 15.6% had asthma history. With the the involvement of Family Pedigree interview revealed that history of Genetic diseases 5.9% and Down syndrome was 5.9% and diabetes was 51.9%. There was significant difference was noticed with family pedigree interview. In comparison with regularly used family history questionnaire in primary care unit our study genetic family history questionnaire was succeeded in measuring familial genetic risk factors in higher percent for non-insulin dependent diabetes, cancer (colon breast, prostate, varies, etc.), hypertension and coronary heart diseases. **Conclusions:** Based on the study results, the currently used family record folders which were available in PHC not focussed on genetic diseases family history only focused on few selected diseases. Good evaluation of familial genetic risk is crucial to assess the potentiality of patient to get diseases by assessing genetic risk factors and in that order the physician may find preventive measures to decrease his patient disease potentiality in terms of reduction of cost expenditure, premature deaths and quality of productive life .

KEYWORDS: Genetic risk; Family pedigree, PHC, Different diseases, Qassim Region, Saudia Arabia.

INTRODUCTION

The family history has long been recognized as a risk assessment tool in primary care services, despite the recent stress on the importance of the family history in primary care, there appears to be a significant shortage considering obtaining complete and specific family history information in primary care practice. As an example "occupied, busy schedules of physician and limited knowledge, skills and training of primary care clinic physician regarding advances in human genetics and how to use effective screening questionnaire.[1]

It is important to the screening tool selected to be tailored to the practice setting and patient population, taking into consideration patient education level, cultural specifications and catch on the update in the family history periodically for new diagnoses within the family and throughout pregnancy as appropriate.[2] The family history also it contributes in making a diagnosis, determining risk, and assessing the needs for patient education and psychosocial support.[3]

Several methods have been established to obtain family medical histories; each has its own advantages and dis-advantages. A common tool used in general practice is the family history questionnaire or checklist. Direct patient questioning permits clarification of medical terminology that may be unclear to the patient. Any positive responses on the questionnaire should be followed up by the health care provider to obtain more detail, including the relationship of the affected family member(s) to the patient, exact diagnosis, age of onset, and severity of disease.[4]

1995; American Society of Human Genetics (ASHG), Suggested that analysis of the family history appears to be the most appropriate method for the initial identification and stratification of genetic risk for common and chronic disorders. In the case of heart disease, cancer and diabetes, the family history may reveal the magnitude of these disease risks. Typically, a family history of these disorders is associated with relative risks ranging from 2 to 5 times those of the general population.[5]

Another family history assessment tool, commonly used by genetics professionals, is the pedigree. The health care provider may decide to complete a detailed pedigree or refer the patient to a genetics professional for further evaluation. A pedigree basically shows at least three generations and involves the use of standardized symbols, which clearly mark individuals affected with a specific diagnosis to allow for easy identification. The pedigree assists in determining the size of the family and the mode of inheritance of a specific condition, and it may facilitate identification of members at increased risk of developing the condition.[2]

But the genetic pedigree interview is far too time-consuming to use in routine general practice. A simple structured format is needed to gather enough information on first degree relatives, grandparents and relevant second degree relatives to allow a preliminary assessment of genetic risk and also be suitable for review in a 10-minute consultation.[6] Moreover, the use of a family history screening tool (pedigree or questionnaire) has been shown to increase the likelihood of detecting a patient at high risk of developing an inherited medical condition by 20% compared with medical record review alone.[2]

OBJECTIVES

The objective of this study:

1. To assess the ability of a currently used Family History Questionnaire, to identify genetic risk.
2. To assess the ability of newly designed family history questionnaire, interview to identify genetic risk.
3. Compare the ability to identify genetic risk between newly designed questionnaire and currently used questionnaire.

METHODOLOGY

The present institutional based Descriptive study was conducted at some primary health centres of Qassim region during the period from March 2015 to August 2015. Prior to commencing the study, ethical approval was obtained from the relevant Local Research Ethics Committees, Qassim University - College of Medicine. Descriptive study, compared a random sample of reviewer currently used formal family history questionnaire in PHC with newly designed family history questionnaire for evaluating risk of common genetic diseases in Qassim population. Already used family history questionnaire in primary care centers, which is part of family file will be filled and evaluated in contrast to newly designed family history questionnaire. The sample size calculation was done in respective to pilot study using EPI6 and it was estimated to be 289 randomly selected family files from four Primary Health Centres representative to PHCC in Quassim Region population. Data which obtained using both instruments were entered into a Microsoft Access Database; accuracy of database entry was determined by double checking one in two entries. The data were then analyzed in SPSS version 17 (SPSS Inc.). Age and sex of all participants and comparison of the family history information from the informants completing both instruments were described. Further, the pairs of scores for each informant were compared. Informed consent was taken and maintained confidentiality of the individual.

RESULTS

This study was conducted at four primary care centers at Qassim governorate a total 289 family files was randomly selected, 25. 8% of these files were for extended families and 74. 2% were nuclear families , each family heads was interviewed and completed two questionnaires, one the original questionnaire that must be completed at the opening of a family file at a health care center and the other the proposed family history questionnaire which was recommended by the study.

Table 1: Family history as screened by currently used questionnaire in PHC.

Available diseases family history	Currently used questionnaire n	%
Diabetes	176	61.9
Hypertension	154	53.3
Tuberculosis	9	3.1
Skin diseases	18	6.2
Asthma	45	15.6
Others	24	8.3

Table 1: showed the diseases which were considered in currently used family file questionnaire and revealed that nearly 61.9% of total files had a family history of diabetes and 53% had a history of hypertension also 15.6% had asthma history and 3% for tuberculosis and 6% had a history of skin diseases.

Table 2: Personal history as screened by currently used questionnaire in primary care centers

Available diseases history	Currently used questionnaire n %	
Allergies	24	8.3
Rheumatic fever	7	2.4
Tuberculosis	2	0.7
Epilepsy	3	1.03
Others	108	37.4

Table 2: Out of 289 study population, 8.3% (24/289) were showing allergy as personal history and only 07% were shown as Tuberculosis as self reported disease.

Table 3: The personal history and familial risk as shown in the proposed genetic questionnaire.

Disease	Personal history	Number of total family history	Percent of total family history	Degree of relation	Risk level
Blood disorder	2	11	3.8%		
				8 (1st degree)	8 (high risk)
Anemia	1	10		2 (2nd degree)	2 (moderate risk)
Leukemia & lymphomas	1	6		5 (1st degree)	5 (high risk)
				1 (2nd degree)	1(average risk)
Infertility		39	13.5%	25 (1st degree)	18 (high risk)
				14 (2nd degree)	7(moderate risk)
					14(moderate risk)
Respiratory disorder	27	95	32.9%	70 (1st degree)	55(high risk)
					15 (moderate risk)
Bronchial asthma	26	76	26.3%	6 (2nd degree)	6 (average risk)
Ch. obstructive lung disease	0	1	0.35%	1 (2nd degree)	1(moderate risk)
Cystic fibrosis	0	1	0.35%	1 (2nd degree)	1 (moderate risk)
Pulmonary edema	0	1	0.35%	1 (1st degree)	1 (high risk)
Tuberculosis	1	0	0.35%	1 (1st degree)	1 (moderate risk)
Neurological disorders	2	100	34.6%		
				1 (1st degree)	1 (moderate risk)
				17 (2nd degree)	17(average risk)
Alzheimer	0	18	6.2%	1 (1st degree)	1 (high risk)
Autism	0	1	0.35%	4 (1st degree)	4 (high risk)
				4 (2nd degree)	4 (moderate risk)
Depression	0	8	2.7%	15 (1st degree)	15 (high risk)
				4 (2nd degree)	4 (moderate risk)
Epilepsy	1	19	6.6%	10 (1st degree)	10 (high risk)
				4 (2nd degree)	4 (moderate risk)
Mental retardation	0	14	4.8%	4 (1st degree)	4 (high risk)
				17 (1st degree)	3 (high risk)
Schizophrenias	0	4	1.4%	15 (2nd degree)	14 (moderate risk)
Stroke	1	32	11%		15 (average risk)

Table 3: Depicts that 32.9% of patients had family history for respiratory diseases mainly bronchial asthma and 13% had a history of infertility and 3.8% had a history of severe anemia with all categories under such diseases. At the same table history of neurological disorders was 34% under this is epilepsy, stroke,Alzheimer, mental retardation and depression.

Table 4: Personal history of Preganancies and familial risk as shown in the proposed genetic questionnaire.

Disease	Personal history	Number of total family history	Percent of total family history	Degree of relation	Risk level
Pregnancy related dise.	13	52	18%		
Abortion	10	19	6.6%	12 (1st degree) 7 (2nd degree)	12 (high risk) 7 (average risk)
Infant death within 1st 2 y	0	6	2%	6 (1st degree)	6 (high risk)
Still birth	3	9	3.1%	9 (1st degree)	9 (high risk)
Birth defect	0	11	3.8%		
Club foot	0	0	0	-----	-----
Clift lip and palate	0	6	2%	4 (1st degree relative) 2(2nd degree relative)	4 (high risk) 2 (moderate risk)
Congenital hip dislocation	0	1	0.35%	1 (1st degree relative)	1 (high risk)
Poly dactyl	0	2	0.7%	2 (1st degree relative)	2 (high risk)
Hydrocephalus	0	1	0.35%	1(2nd degree)	1 (high risk)
Ocular diseases	5	41	14.2%		
Blindness	1	17	5.9%	11 (1st degree relative) 6 (2nd degree relative)	7(high risk) 4(moderate) 6 (average risks)
Glaucoma	1	5	1.7%	5 (2nd degree relative)	5 (average risks)
Cataract	3	18	6.2%	11 (1st degree relative) 7 (2nd degree relatives)	1(high risk) 10(moderate risk) 7(average risks)
Cataract and glaucoma	0	1	0.35%	1 (2nd degree relatives)	1 (average risk)

In table 4 pregnancies related disease history was about 18%, ocular diseases were 14.2%, and birth defects was 3.8%.

Table 5: Personal history and familial risk as shown in proposed genetic questionnaire.

Disease	Personal history	Number of total family history	Percent of total family history	Degree of relation	Risk level
Skin diseases	5	44	15.2%		
Atopic dermatitis	0	5	1.7 %	3 (1st degree relatives) 2 (2nd degree relatives)	3 (high risk) 2 (average risk)
Psoriasis	1	3	1%	1 (1st degree relatives) 2 (2nd degree)	1 (high risk) 2 (average risk)
Vitilligo	4	11	3.8%	5 (1st degree relatives) 6 (2nd degree)	5 (high risk) 6 (average risk)
Coronary heart disease	0	11	3.8%	6 (1st degree relatives) 5 (2nd degree)	6 (high risk). 5 (average risk).
Hypertension	25	122	42.2%	73 (1st degree) 49 (2nd degree)	52 (high risk). 19 (moderate risk) 49 (average risk)
Cardiac anomalies	0	3	1%	3 (1st degree relatives)	3 (high risk)
Hypercholesterolmeia	0	3	1%	3 (1st degree relatives)	3 (high risk)
Renal diseases	0	25	8.7%		
Renal failure	0	14	4.8%	9 (1st degree relatives) 5 (2nd degree)	9 (high risk) 5 (average risk)
Renal stones	0	6	2%	3 (1st degree relatives) 3 (2nd degree)	2 (high risk) 1 (moderate risk) 3(average risk)
Nephritic syndrome	0	1	0.35%	1st (degree relatives)	1(moderate risk)
Nephrotic syndrome	0	1	0.35%	1st (degree relatives)	1(moderate risk)
Nephrectomy	0	1	0.35%	1 (2nd degree)	1(average risk)
Genetic diseases	0	17	5.9%	10 (1st degree)	10 (high risk)
Down syndrome	0	17	5.9%	7 (2nd degree relatives)	7 (average risk)

In table 5: Reveals that skin disease history was 15.2% and hypertension was 42.2% coronary heart was 3.8%, renal disease was 8.7%, Genetic diseases 5.9% and Down syndrome was 5.9%. All those history data were accompanied with degree of relativity and age of the affected relative at early disease which cited as risk factor ranking criteria.

Table 6:Personal history of Cancers and Metabollic diseases and familial risk in proposed genetic questionnaire

Disease	Personal history	Number of total family history	Percent of total family history	Degree of relation	Risk level
Cancer	0	53	18.3%		
Cancer lung	0	10	2.8%	3 (1st degree relatives) 7 (2nd degrees)	3 (high risk) 7 (average risk)
Cancer brain	0	2	0.6%	1 (1st degree) 1 (2nd degree)	1(high risk) 1 (average risk)
Cancer breast	0	1	0.35%	1 (1st degree)	1(high risk)
Cancer colon	0	8	2.8%	2 (1st degree) 6 (2nd degree)	2 (high risk) 6 (average risk)
Cancer liver	0	8	2.8%	4 (1st degree) 4 (2nd degree)	4 (high risk) 4 (average risk)
Metabolic diseases	32	150	51.9%	76 (1st degree)	24 (high risk.) 52 (moderate risk)
Diabetes mellitus	29	132	45.7%	56 (2nd degree)	56 (average risk)
Obesity	3	4	1.38%	4 (1st degree)	4 (high risk.)

Table 6: Depicts that all cancers categories history was 18.3%, diabetes mellitus history was 45% and obesity was 1.38%..

DISCUSSION

A family health tree is a guide to an individual's or family's genetic makeup. Many common disorders such as diabetes, heart disease, cancer and mental illness may have a genetic basis. If health care physicians are aware of their patient's family health history, it may be possible to predict, prevent or treat health problems that have affected previous generations.[7]

Family history remains one of the most powerful genetic tests to identify individuals at risk of genetic disorders when laboratory tests are not available,it can be used to identify single gene disorders or chromosomal abnormalities that affect family members. More frequently the family history will identify families with increased susceptibility to disorders such as diabetes or hypertension that diseases rarely caused by single gene mutation and often influenced by environmental condition and lifestyle.[8]

There is a significant history of an individual or individuals affected with a disorder that has a known or suspected genetic basis, or if multiple family members are affected with a particular disorder, the pedigree should be extended to identify additional affected family members and to as certain the inheritance pattern.[9]

Family history has traditionally served as a raw predictor of disease risk in patients. Recently, more attention has been paid to defining the best approach to eliciting this history and how it might better guide disease prevention protocols. Frezzo et al and Scheuner[3,5] discuss the development of a self-administered family history questionnaire for patients that appears easy to use and informative. Scheuner notes the "need for improvement in [family history documentation]" by primary care physicians and that this questionnaire appears a viable substitute for the time-consuming three-generation pedigrees[5,10]

The Agency for Health Care Policy and Research spear headed the "Putting Prevention into Practice Program" (PPIP). PPIP offers materials that enable clinicians to comply with recommended prevention guidelines and publishes handbooks encouraging patients to take an active role in disease prevention. These materials include an annual adult health risk profile for patients, containing direct questions about family history of hypertension, cardiovascular disease, hyperlipidemia, gastrointestinal and gynecological cancers, and glaucoma.[10]

Frezzo et al.[3] found that the most commonly reported disorder in genetic assessment questionnaire is coronary artery disease by 38.5% compared to 28.2% discovered by ordinary family history questionnaire,in our study only 3.8% were discovered by genetic history questionnaire compared to 0% by regular family history questionnaire, Frezzo also found that his genetic risk assessment questionnaire assessed 29.5% of his study population as increased risk of non-insulin dependent diabetesmellitus compared to 19.2% of at risk population discovered by ordinary family questionnaire. In our study our genetic risk assessment questionnaire found 52% of the study population at risk of developing diabetes compared to 60% by ordinary family history questionnaire.

Concerning breast, ovarian,prostate and colon cancers Frezzo questionnaire found 17.9% of his study group familial risk compared to 6.4% of the ordinary questionnaire group which is matched with our study as we found that 18.3% of our study population at risk of cancer by genetic questionnaire compared to zero percent by regular questionnaire.[3,11]

This study demonstrates that a majority of our study population has one more significant genetic risk factors for which preventive medical recommendations can be suggested. A family history is a collection of information about the health history of an individual's biological relatives. Fundamentally, collecting a family history is an inexpensive, noninvasive screening procedure.[12] For family histories to improve health outcomes, the information collected must be accurate, the risk to the patient identified and effectively communicated, and appropriate action taken by provider and patient. Each of these steps presents significant challenges.

CONCLUSIONS

Based on the study results, the currently used family record folders which were available in PHC not focussed on genetic diseases family history only focused on few selected diseases. Good evaluation of familial genetic risk is crucial to assess the potentiality of patient to get diseases by assessing genetic risk factors and in that order the physician may find preventive measures to decrease his patient disease potentiality in terms of reduction of cost expenditure, premature deaths and quality of productive life**.** Target family history analysis reveals good evaluation of familial genetic risk is crucial to assess the potentiality of patient to get diseases by assessing genetic risk factors and in that order the physician may find preventive measures to decrease his patient disease potentiality in coming future.

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