Original Article

Psychometric of Physicians' Awareness, Attitudes and Performance Questionnaire about Genetic Counseling and Testing

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Abstract

Introduction: Considering the key role of physicians in providing genetic counseling services and the lack of studies in this field in Iran, it seems necessary to design a valid and reliable instrument for measuring the awareness, attitude, and performance of general and specialist physicians in genetic counseling. **Materials and methods:** In this descriptive study, the design and psychometrics of the questionnaire were performed in 4 steps: first, defining the concept of awareness, attitude, and performance of general and specialist physicians in relation to genetic counseling by reviewing texts and articles; second, designing questionnaire items; third, determining the face and content validity by 10 university experts; and forth, determining reliability using Cronbach's alpha coefficient method. **Results:** The primary version of the questionnaire was designed taking into account 60 items during the first and second stages. In the third step, one item was removed and in the final version of the questionnaire 59 items and the content validity index (CVI) and content validity ratio (CVR) were reported to be 0.98 and 0.92, respectively. Reliability with Cronbach's alpha coefficient was determined 0.82. **Conclusion:** The final questionnaire with 59 items had appropriate psychometric properties. This questionnaire has the ability to be used by health care providers in health care systems to measure the awareness, attitude, and performance of physicians about genetic counseling. The need for further studies is suggested to measure the other types of validity, such as the structural validity of the questionnaire.

Keywords: *Attitude, awareness, genetic counseling, performance, physicians*

Introduction

Today, controlling infectious diseases and vaccine-preventable diseases has reduced infant mortality. Such conditions increase the share of genetic diseases in infant mortality and disability.[1] At present, the annual incidence of hereditary disorders and diseases in Iran as a country with an average income level is 3 to 5% of live births.[2] Due to their complex nature and long-term complications, these diseases lead to a continuous increase in hospital visits, widespread and continuous demand for medical services. On the other hand, the merely palliative, supportive, and temporary nature of treatment in most of these diseases causes lasting dissatisfaction in the patient and the medical team and ultimately the helplessness of the client and the service providers.[3] Today, in many common non-communicable diseases such as coronary artery disease and common cancers (breast cancer, gastrointestinal and colon cancers, etc.) and common mental illnesses such as depression, genetics can play an effective role in control and prevention programs. Applying genetic knowledge in disease management can increase the effectiveness of interventions related to these diseases.[4]

According to the Center for Disease Control and Prevention (CDC) in 2013, genetic testing has expanded to 2,200 diseases, with 2,000 available for clinical use. On the other hand, the development of genetic testing for cancer has many benefits for patient management, such as allowing medical professionals to diagnose patients with suspected cancer who have not developed any symptoms, and early detection of cancer to increase the likelihood of recovery, and perhaps treatments.[5] Genetic counseling, as an important link in the genetic service chain and translator of this knowledge to people, in many cases can calculate the risk of disease and how to prevent or reduce it for people, explain and plan. Genetic

How to cite this article: Mousavinasab SM, Khosravi S, Ramezanzadeh M. Psychometric of physicians' awareness, attitudes and performance questionnaire about genetic counseling and testing. Int J Prev Med 2022;13:134.

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Access this article online

Website: www.ijpvmjournal.net/www.ijpm.ir

DOI: 10.4103/ijpvm.IJPVM_608_20

Quick Response Code:

counseling is the process by which a family or individual who is potentially at risk receives the necessary clinical and laboratory examination, including assessments, clinical and laboratory consultations, and then, based on the diagnosis of the disease, patients or people at risk, the consequences of disease and the possibility of contracting or transmitting it to other relatives and preventive ways and interventions, adaptation to genetic disorders or treatment in the existing conditions and to make an informed decision, they are fully accompanied until they have a fair case to services.[6,7]

At present, the widespread need for centers active in the field of medical genetics, the importance of full knowledge of general practitioners as the first to deal with patients, has been considered. On the other hand, due to the presence of a limited number of geneticists to respond to the vast needs of society, the inability of individuals due to genetic diseases and low levels of literacy and family knowledge about genetics and referral of most patients at risk to doctors, may it can suggest the effective role of general practitioners and specialists in referring these people to genetic counseling.[8,9] Therefore, it seems necessary to assess the knowledge, attitude, and practice of general practitioners and specialists in relation to counseling and genetic testing.

Studies examining physicians' knowledge, attitudes, and practices have focused more on measuring the knowledge and attitudes of a particular group of physicians, such as general practitioners, specialty students, or medical assistant students or interns in relation to a particular genetic test. The tools used in these studies were appropriate for the purpose of the study, suitable for use in a specific group and population studied.[10-13] Stefania Bossia *et al.* (2014), in their study in Italy, examined the knowledge and attitudes of internal residents regarding genetic tests for breast and colorectal cancer using a 14-item questionnaire.[10] Hunter *et al*. (1998)[11] in their study in Canada to assess the knowledge and attitudes of general practitioners and specialists in relation to genetics, used a questionnair that contained 110 questions about their sources of genetic knowledge, confidence in providing genetic counseling, attitudes toward referrals to genetic counseling centers and knowledge of molecular genetic tests and attitudes about their use in clinic and population screening.

In another study conducted (2014) in the United States by Bonham *et al*.,[12] researchers have introduced a questionnaire to measure knowledge about genetic diversity and racial characteristics in clinical assessments. The researchers introduced and psychoanalyzed three scales: GKAI (to measure knowledge about genetic diversity), HPBR (to measure the beliefs of health professionals about the human race), and Racial Attributes in Clinical Evaluation (RACE) using racial attributes in clinical evaluations).

The GCSS scale for measuring patients' satisfaction with genetic counseling was also introduced and evaluated by Tercyak *et al*. In 2004 in a sample of 61 Colombian women receiving genetic counseling services.[13] This scale also assesses the level of patient satisfaction with the genetic counseling process.

Tan *et al*.[14] also designed a 19-item scale to assess the attitudes, knowledge, and practice of physicians specializing in genetic counseling in Lynch syndrome.

By reviewing studies conducted in the field of genetics in Iran, researchers are more likely to have assessed the attitudes and knowledge of medical students about genetic counseling, abortion therapy,[15] premarital genetic counseling in couples,[16,17] the educational needs of couples about thalassemia[18] and according to our research, little study has been done on measuring the knowledge and attitudes of physicians regarding genetic counseling and a standard scale to measure this important category has not been introduced.

However, due to the limited resources of health systems and the need to reduce the burden of non-communicable diseases, and the use of genetic prevention tools in the management of non-communicable diseases, the World Health Organization has limited access to these services in middle-income countries such as Iran and emphasized the need for its development[2] and considering the serious role of physicians in genetic counseling and the need to refer to genetic counseling, design and manufacture a valid and reliable tool to measure the knowledge, attitude and practice of physicians in relation to genetic counseling that is appropriate for Iranian culture; It seems necessary. Therefore, the present study aimed to determine the validity and reliability of the questionnaire of knowledge, attitude, and practice of general practitioners and specialists in relation to genetic counseling.

Method

In this descriptive and analytical study, the design and psychometrics of the questionnaire were performed according to the Waltz 2010 method in 4 stages:

1) Defining the concept of awareness, attitude, and performance of physicians in relation to genetic counseling by reviewing books, articles, and genetic theories. 2) Designing questionnaire items using available internal and external sources. 3) Determining the validity of the questionnaire. 4) Determining the reliability of the questionnaire.[19]

Step 1: In this step, by purposeful study and review of relevant articles, and books[20,21] with the concept of knowledge, attitude, and practice of physicians in relation to genetic counseling were defined.

Step 2: In this step, according to similar questionnaires, including the questionnaire "Assessment of knowledge, attitude and practice of physicians in relation to genetic counseling in Lynch syndrome" Tan *et al*.[14] and GKAI

scale to assess the knowledge of health professionals about genetic diversity.[12] The initial version of the questionnaire consisted of 60 items was designed.

Step 3: After preparing the questions, two methods of face validity and content validity were used to examine and determine their validity.

Step 4: In this step, the reliability of the questions was determined. The questions were examined in terms of internal reliability. Cronbach's alpha test was used to determine the internal consistency of the questionnaire and the questionnaire was given to 31 general practitioners and specialists.

Face Validity

To evaluate face validity, the question is whether the appearance of the tool is properly designed to evaluate the intended purpose? Formal validity takes place both qualitatively and quantitatively.

In face validity, the questions were given to 10 participants as well as people specializing in the fields of genetics and tool design, so that the questions were evaluated from three perspectives: level of difficulty, appropriateness, and ambiguity. In quantitative form validity, which is the search for the importance of items from the participants' point of view, the quantitative method of item effect is used to reduce and eliminate items. If the impact score obtained for each item is more than 1.5, the item is deemed suitable for subsequent analysis and is retained.[20]

In the present study, in order to determine the quantitative face validity, a questionnaire was given to 10 qualified general practitioners and specialists in gynecology, pediatrics, internal medicine, and oncologist. After completing the questionnaire by the target group, face validity was calculated using the following formula (item effect method).

Impact Score = Frequency (%) × Importance

Content Validity

Content validity refers to the extent to which the tool questions represent the content and purpose of the topic. Content Validity Index (CVI) is the most common quantitative method used by researchers to determine the content validity on multiple-choice scales, and this method is based on the degree of relevance of items based on the judgment of a panel of experts. This index was used to ensure that the items of the tool were designed to measure the awareness, attitude, and performance of physicians in relation to genetic counseling in the best possible way or not? In the present study, the content validation index and content validity ratio (CVR) were used to confirm the content validity. Content validity ratio was invented by Lawshe. To calculate this ratio, members of the panel of experts judge each item as necessary.[22,23]

Content Validity Index Calculation

To calculate this index, three criteria of simplicity and fluency, relevance, clarity and transparency are used using the Likert scale with 4 options for each item. Content validity index was calculated using the following formula:

$$CVI = \frac{\text{number of specialists who gave the score of 3 - 4 to the item}}{\text{total number of specialists}}$$

Calculate the Content Validity Ratio

To calculate this ratio, the opinions of 10 experts and people in genetic counseling and instrument design were used. Each item was examined using three spectra: item is necessary, the item is useful but not necessary, and the item is not necessary. The content validity ratio was calculated using the following formula.[24]

The calculated ratios for each item are compared with the numbers provided by Lawshe, and if the value of the ratio obtained is greater than the values in the table, the content validity of that item is confirmed.

$$CVR = \frac{\text{number of essential answer for each item} - \frac{\text{total number of participants}}{2}}{\frac{\text{total number of participants}}{2}}$$

Results

According to the findings of the study of articles, books, the concept of knowledge, attitude, and practice of general practitioners, gynecologists, internal medicine, pediatrics, and oncology in relation to genetic counseling were determined; Thus, physicians' knowledge includes knowledge of cases in which couples need genetic counseling (such as having or family history of disorders such as phenylketonuria, learning disability, early onset cardiovascular disease, and various cancers, etc.), knowledge about the role of genetic factors in cause of diseases as well as knowledge about the stages of genetic counseling and other examples of genetic knowledge include knowledge of the information which is passed to the counsultand during genetic counseling and providing available options to adapt to the existing risk and problem and knowledge about genetic tests.

The attitude towards genetic counseling refers to the attitude of physicians towards the cost, the time required for counseling, the stress of counsultand during the counseling process, the commitment of physicians to refer patients in need of genetic counseling, and the need of specific guidelines for physicians to refer to genetic counseling. Performance axis refers to the performance of physicians in referring people in need of genetic counseling and recommending genetic testing in the past 6 months. It also refers to all the things that doctors need to do to motivate and effective referring.

In the second stage, the relevant questions were designed inspired by similar questionnaires.[12,14] The initial version of the questionnaire consisted of 60 items so that in the field of awareness it had (36 items) in the area of attitude (10 items) and in the area of performance (14 items).

Face Validity

Calculating the item impact score: After calculating the item impact score index, considering that the values of the tool items were higher than 1.5, none of them were deleted at this stage, and all of them in terms of the target group were considered important and appropriate and was maintained for next steps.

Content Validity Index

Calculation of content validity ratio: The results of calculating this ratio showed that out of 60 items, the values of content validity ratio in 3 items were lower than the values presented in the table for 10 people (0.62). However, at this stage, no item was removed due to a low score, and after reviewing the relevant items and applying the comments and suggestions of panel members, a 60-item questionnaire was given to panel members to determine the content validity ratio. At this stage, out of 60 items, 1 item obtained values lower than the scores presented in the table and was removed, and the number of items reduced to 59.

Content Validity Ratio & Content Validity Index

The overall score of content validity ratio was calculated in a 59-item questionnaire CVR = 0.92. Also, the content validity index for all items is above 0.79 and its total value was calculated as CVI = 0.98 [Table 1].

Reliability

Cronbach's alpha coefficient was used to determine the reliability of the 59-item questionnaire. The total reliability of the questionnaire in a sample of 31 eligible physicians was value of 0.82. It was calculated separately in the field of awareness the value of 0.78, in the field of attitude the value of 0.63, and in the field of performance the value of 0. 90 obtained.

It should be noted that in the mentioned questionnaire, the scale of answering the questions in the awareness domain with 35 questions is yes = 1 or no = 0 and the total score is 0-35, the questions in the attitude domain with 10 questions are in the form of 3-choice Likert I agree = 2, I have no opinion = 1 and I disagree = 0 and total score is 0-20 and in this domain 6 questions are inverse. Also, the response scale in 3 questions in the performance domain is yes = 1 or no = 0 and the remaining 11 questions are 6-point Likert based on the percentage of referrals during the last 6 months and the total score is 0-58.

Discussion

The aim of this study was to design and psychrometer the questionnaire of awareness, attitude, and performance of general practitioners and specialists in relation to genetic counseling. The results of the study showed that the designed tool has appropriate validity and reliability to measure the awareness, attitude, and performance of general practitioners and specialists in relation to genetic counseling. Ghasemi *et al*. (2007)[15] in their study in Yazd, designed a questionnaire with 11 items to assess the knowledge and attitudes of medical students about medical abortion and genetic counseling. 7 items of this questionnaire were used to assess students 'knowledge about genetic counseling, genetic tests available during pregnancy, and the time and indication of abortion, and another 4 items were used to assess students' views on selected abortion in case of fetal disorders. In the present study, the need for genetic counseling, the role of genetic in disorders, the stages of counseling, and existing tests are the most important questions in the field of knowledge of the questionnaire. However, Ghasemi *et al*. did not mention the scale of answering the questions of the questionnaire in the areas of knowledge and attitude, as well as psychometric information (validity and reliability) of the questionnaire.[15]

Tan *et al.* (2014) in Australia designed and psychoanalyzed a questionnaire based on their qualitative study on the facilitators and barriers to genetic counseling in Lynch syndrome, a comprehensive review of relevant sources and articles, and the Delphi method to measure knowledge, attitude and practice of professionals Health about genetic counseling in Lynch syndrome. To determine the content and face validity of the questionnaire, 10 members of the panel consisting of oncologists, gynecological oncologists, geneticists, and clinical and social researchers were present. The final version of the 19-item questionnaire provided, which included demographic information, referral indications, barriers and facilitators for referral to counselors, and the role of physicians in supporting the provision of genetic services.

One of the most important aspects of the above questionnaire is measuring the barriers and facilitators of physicians' performance to refer their clients to genetic counseling. Because highlighting barriers and facilitators of genetic services can improve the information and knowledge of physicians as well as their patients and the development of initiatives in the provision of genetic services, which in turn leads to positive behaviors and better health outcomes for patients.[25] Surely, in the above questionnaire in the field of physicians 'referral performance, physicians' performance was evaluated only in relation to 3 DNA mutation tests, IHC (ImmunoHistoChemistry) and MSI tests. The three options: yes, no and I am not sure, designed for Lynch syndrome. While in the questionnaire of the

Table 1: Content Validity Ratio (CVR) and Content Validity Index (CVI) of each item for Questionnaire of awareness, attitude and performance of general practitioners and specialists in relation to genetic counseling

Relevant items and areas	CVI	CVR
Scope of awareness		
Which of the following requiers genetic counseling?		
1. A couple who intend to get married and have phenylketonuria in their family?	1	1
2. A couple who have a child with learning disability and intend to conceive again.	1	1
3. A woman who has planning to become pregnant and her father has died of a heart attack at the age of seventy	0.88	02
4. A person whose close relatives include people with cardiovascular disease under the age of 50.	1	0.8
5. A couple who have a child with asthma.	0.9	0.8
6. Parents whose neonate has a positive test for phenylketonuria screening.	1	0.8
7. A person who has multiple cases of breast cancer in her family and is worried about getting it.	0.9	0.8
8. A person with family history of infertility.	1	1
9. A woman who has a history of one spontaneous abortion for no apparent reason.	1	1
10. Couples with minor thalassemia.	1	1
11. A pregnant woman who is at high risk for screening in the first trimester.	1	1
Genetic factors are effective in causing which of the following diseases?		
12. Cancer	1	1
13. Cardiovascular Diseases	1	1
14. Diabetes	1	1
15. Cleft palate	1	1
16. Schizophrenia	1	1
17. Lumbar disc herniation	1	1
18. Congenital pyloric stenosis	1	1
which of the following is one of the steps in genetic counseling?		
19. Diagnosis of a problem or disease	1	1
20. Estimating the risk of recurrence in the family	1	1
21. Decision making about treatment of an affected individual	1	1
22. Decision making about abortion of affected fetus	1	1
What information is passed on to people during genetic counseling?		
23. The medical diagnosis and its implications in terms of prognosis and possible treatments.	1	0.8
24. The mode of inheritance of the disorder and the risk of developing and/or transmitting it to the next generation	1	0.8
25. The options and items available for dealing with the risks	1	0.8
26. Estimating the cost of treating an affected individual	1	0.8
27. Selection and introduction of the treating physician	1	0.8
*Which genetic disorders can be examined with the available tests?		
28. Chromosomal	1	1
29. Single gene	1	1
30. Susceptibility to multifactorial disorders	1	0.8
31. Prenatal diagnosis of multifactorial disorders	1	0.8
Which of the following would you recommend for someone who needs genetic counseling or genetic testing?		
32. I refer the person to the nearest genetic counseling center.	1	0.78
33. I refer to the most equipped genetic center regardless of distance and cost.	1	0.78
34. I give the patient the necessary information and let him choose.	1	0.78
35. Do you know enough about the process of how to refer a patient to genetic counseling?	0.88	0.78
36. Do you have access to a genetic counseling service?	1	1
Scope of attitude		
37. In my opinion, genetic counseling does not help consultand in comparsion to the cost that ultimately imposes	1	1
38. In my opinion, genetic counseling does not help the consultand in comparsion to time it takes from she/he	1	1
39. Because genetic diseases have no definitive cure, genetic counseling cannot help a patient with a genetic disease	1	1
40. Genetic counseling can help a patient’s family to manage disease	1	1
41. Genetic counseling can reduce the birth rate of affected neonates	1	1
42. All physicians are required to refer their patients for genetic counseling if necessary	1	1
43. Genetic counseling can help improve community health indicators	1	1
44. Genetic counseling is the sole responsibility of geneticists	1	1

Contd...

Table 1: Contd...

Relevant items and areas	CVI	CVR
45. In my opinion, it is necessary to provide doctors with a specific guide for referring patients to genetic counseling	0.9	0.62
46. I think genetic counseling increases stress of consultand	0.8	1
Scope of performance		
47. Have you used a specific guideline to select your patients for referral to the relevant center?	1	1
48. Do you have connection with genetic counseling centers?	1	1
49. Do you provide the necessary explanations in the time of patient referral in order to gain his/her confidence about keeping his/her secrets?	1	1
Please pay attention to the following issues and respond based on your referral experience of the last six months		
50. Infertile couples after routine examinations	1	1
51. Repeated abortions of unknown cause	1	1
52. A family have a child with learning disability or genetic disorder	1	1
53. Stillbirth with unknown cause	1	1
54. Before marriage or pregnancy in consanguineous marriages	1	1
55. A person with a family history of cancer.	1	1
56. A person with a family history of early onset cardiovascular disease	1	1
57. The family of an affected person who needed screening	0.8	0.88
58. People in need and interested in genetic counseling	0.09	0.62
59. What percentage of cases who needed genetic testing were reffered to the lab?	0.88	0.78
60. Other cases of referral to genetic counseling (please mention the case in the following columns with percentage).	0.88	0.78

present study, in the field of performance, the performance of physicians in using a specific guideline for referral as well as referral function in various diseases and genetic conditions (cancers, recurrent miscarriages, cardiovascular problems, consanguineous marriage and.) is measured in terms of the percentage of referrals, which of course is due to differences in the field under study.

Trecyak *et al*. (2004) in Colombia examined the validity and reliability of the GCSS (Patient Satisfaction with Genetic Counseling) scale in a sample of 61 women receiving genetic counseling services for breast and ovarian cancer. This scale has 6 items and the answer scale is a 5-point Likert (strongly agree, agree, have no opinion, disagree, strongly disagree). Cronbach's alpha test was also used to assess the reliability of this questionnaire that 90% was obtained.[13,26]

Bonham *et al*. (2014)[12] in the United States, designed and psychoanalyzed three scales: GKAI, HPBR, and RACE in a sample of 787 interns; In order to design the items of each scale from cognitive interviews and holding a panel of experts, exploratory factor analysis was used. To determine the validity of the structure Cronbach's alpha method was used to determine the reliability of the scales. The results of the study showed that the GKAI scale is a desirable scale for evaluating the knowledge of health professionals about genetic diversity. Race and genetic diversity in estimating the risk of diseases with Cronbach's alpha (61.3%) and RACE scale having a range with Cronbach's alpha of 86.7%, is a valid tool for measuring the use of racial characteristics in clinical evaluations of health professionals.

In a 2019 study by Haja *et al*. In North Carolina on the knowledge, attitudes, and experience of primary care physicians in relation to individual genetic testing, the GKAI Bonham *et al*. Used.[12] According to the authors, the lack of a credible tool to measure physicians' actual knowledge that leads to behavioral change, and the fact that the questions in the field of knowledge were more relevant to the general principles of genetics, and the lack of questions about integrating genomic information into practice, including limitations of this questionnaire.[27]

In the questionnaire of the present study, in the field of awareness, questions were about the indication of genetic counseling in various diseases, the role of genetic factors in causing diseases such as cancer, cardiovascular disease, diabetes, cleft palate, etc., stages of genetic counseling, information which is passed on to the client during the consultation, the uses of existing laboratory tests and existing genetic centers, the extent of their access and communication, and how they are referred.

In another questionnaire designed by Hunter *et al*. (1998)[11] in Canada to measure the knowledge and attitudes of general practitioners and specialists about molecular genetic testing, the scale contained 110 questions in eight areas. Demographic information 9 questions, Genetic information sources 16 questions, Genetic education 6 questions clinical experiences about genetic diseases 19 questions, doctors' confidence to manage specific genetic situations 4 questions Physicians' views on molecular genetic testing and patient referrals 22 questions, knowledge about available genetic tests 17 questions, physicians' functional attitudes toward 3 specific clinical genetics situations 10 questions, and physicians' views on screening, the population is 7 questions. Despite addressing all aspects of genetics in the items of the above questionnaire, but in

the study, there is no evidence of psychometric information of the questionnaire and on the other hand, considering the number of items in the questionnaire, it seems to answer in the group of physicians according to a particular job is time-consuming.

One of the limitations of the present study was the lack of construct validity to determine the subscales of the questionnaire and also the use of a Cronbach's alpha method to determine the reliability of the questionnaire. However, due to the busyness and difficulty of re-accessing qualified samples (general practitioners and specialists) and the fact that in the Cronbach's alpha method, one step test is needed to provide an estimate of the reliability of the test, so the researchers used this method to determine the reliability of the questionnaire and due to the problems mentioned, it was not possible to use the retest method.

Conclusion

The results of the study showed that the designed questionnaire with desirable psychometric properties is a suitable tool for measuring the awareness, attitude, and performance of general practitioners and specialists in relation to genetic counseling. On the other hand, this questionnaire is appropriate for Iranian culture and can be used by health care providers and geneticists. The use of this questionnaire in clinical trial research to determine the effect of interventions in increasing the knowledge and attitude of physicians in the field of genetics, as well as the validity of the structure along with the reliability of subscales in future studies, is recommended.

Acknowledgements

The present study is taken from a research project with the code of ethics IR. BPUMS.REC.1399.008 of Bushehr University of Medical Sciences, which was approved by the research council of this university. Researchers express their gratitude and appreciation to the esteemed professors, members of the panel of experts, general practitioners, and specialists participating in the research.

Received: 27 Jan 21 **Accepted:** 14 Sep 21
Published: 11 Oct 22

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