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Evaluation of a Clinical Decision Support System for the Prescription of Genetic Tests in the Gynecological Cancer Risk

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Abstract

Clinical Decision Support System (CDSS) has been implemented to support physicians about the medical prescription of genetic testing. CDSS is based on open source software. A CDSS for prescribing these genetic tests in BRCA1 and BRCA2 and preventing gynecological cancer risks has been designed and performed in the 'Virgen del Rocio' University Hospital. Clinical evidence demonstrates that BRCA1 and BRCA2 mutations can develop gynecological cancer, but genetic testing has a high cost to the healthcare system.

The developed technological architecture integrates open source tools like Mirth Connect and OpenClinica. The system allows general practitioners and gynecologists to classify patients as low risk (they do not require a specific treatment) or high risk (they should be attended by the Genetic Council), According to their genetic risk, recommending the prescription of genetic tests. The aim main of this paper is the evaluation of the developed CDSS, getting positive outcomes.

Keywords:

Clinical Decision Support Systems, Breast Neoplasms, Ovarian Neoplasms.

Introduction

Breast Cancer (BC) is the most common type of cancer in women in the western world. Besides, Ovarian Cancer (OC) ranks fifth in cancer death in women. However, difficulties in their diagnosis and therapeutic treatment imply high mortality, greater than 50% at five years from diagnosis. The International Agency for Research on Cancer estimated a worldwide incidence of 1.67 million new BC cases per year and over 0.23 million of OC [1].

The appearance of tumors of BC and OC is usually sporadic, but around 10-15% of diagnosed cases are heritable. BRCA1 and BRCA2 genes describe germinal mutations. They are inherent dominantly and with high penetrance in 7% of BC and around 11-15% of OC [2]. Therefore, BRCA1 and BRCA2 mutations increase the risk of BC and OC. Concretely, people with BRCA1 mutations have a 57% risk of developing BC and around 40% of OC. People with BRCA2 mutations have a 49% risk of developing BC and approximately 18% of OC [3-5].

Hereditary gynecologic cancer usually starts in younger people while this trend is not usual for sporadic cancer. Also, it has a more invasive histopathological pattern. However, its diagnosis is important because the result could be positive with determined treatments [6].

In this sense, to know the risk of having a hereditary cancer is essential for people with family history. Moreover, patients' anxiety and concern may be provoked if the risk stays unknown. The treatments for individual patients are different, depending on the BRCA1 and BRCA2 gene mutation. If the mutation is positive, patients will receive monitoring or preventative measures. Otherwise, patients can be relieved of anxiety. In this sense, there are some studies which demonstrated that patients are benefited by the genetic testing results [7-9]. These studies found out a significant decrease in patient concern and anxiety about developing cancer.

In clinical practice, general practitioners and gynecologists are the first in the attendance of the patient. However, the genetic tests to analyze BRCA1 and BRCA2 gene mutation have a high cost to the healthcare system. Previous studies also indicate that performing these genetic tests to the population is not cost-efficient [10]. In this sense, criteria were defined to identify patients with high risk for developing those mutations. They were agreed between the scientific society and the official organization [11]. Genetic testing is only recommended for patients with previous family and personal history. The CDSS was developed for this purpose and it will optimize the prescription and the care process avoids unnecessary patient referrals.

Besides, a recent study has assessed data privacy, security protection and health-promoting role modeling in the technology acceptance model. After controlling for several covariates, perceptions of usefulness, data privacy and security protection, and health-promoting role were all statistically significant factors that influenced the use of electronic personal health records. Those findings suggest that electronic health records users feel more protected and less concerned about privacy and security when their providers use electronic health records [12].

In this paper, this CDSS is functionally and clinically evaluated with preliminary results. Concretely, static rules defined by the Spanish Society of Clinical Oncology (SEOM) will be validated. The dimensions to be assessed are: Perceived Usefulness, Perceived Ease of Use, Social Norms, Facilitating Conditions and Intention to Use.

Methods

Specific questionnaires based on Technology Acceptance Model (TAM) were designed to measure clinical staffs' acceptance of the technology. The model was developed by David (Davis, 1989) and David et al. (David et al., 1989) and it's effective and proven in predicting the use of information and communication technologies. Besides, it has been demonstrated the leading information and communication technology (ICT) application areas for the TAM in health services: telemedicine, electronic health records, and mobile applications. The original TAM has been extended to fit dynamic health service environments by integration of components such as the theory of planned behavior and unified theory of acceptance and use of technology. Those variables frequently reflect the concepts of subjective norm and self-efficacy, but also compatibility, experience, training, anxiety, habit, and facilitators are considered [13].

In another study [14], it was found that the original TAM constructs had a significant impact on the staffs' behavioral intention to adopt HIS in paraclinical departments. The user behavior factors are essential for successful usage of the system and should be considered. It provides valuable information for hospital system providers and policy makers in understanding the adoption challenges as well as practical guidance for the successful implementation of information systems in paraclinical departments.

The TAM model is used to predict the use of technologies, based on two main characteristics: Perceived Usefulness and Perceived Ease of Use. Perceived Usefulness refers to the degree to which a person believes that using a system will improve the performance of a given job. The Perceived Ease of Use aims to assess to what degree a person believes, using a particular system, makes less effort to perform their tasks. The TAM aims to explain the causes of the acceptance of technologies by users. In that sense, it is considered that a person's perceptions of the Perceived Usefulness and Perceived Ease of Use of a system are conclusive in determining their Intention to Use it. According to this model, there are external variables that have a direct influence on the Perceived Usefulness and Perceived Ease of Use. In our study, different variants of TAM are included to evaluate different dimensions.

In order to evaluate the Clinical Decision Support System for the prescription of the BRCA1, BRA2 genetic tests in the prevention of hereditary breast and ovarian cancer, a personalized questionnaire was designed for evaluation by Gynecology based on TAM. The dimensions to be assessed in this questionnaire were: Perceived Usefulness, Perceived Ease of Use, Social Norms, Facilitating Conditions and Intention to Use.

Specifically, the items of the questionnaires assessed for ech dimension are as follows:

Perceived Usefulness

1.1. I believe that the developed system will facilitate coordination with primary care professionals in the integrated assessment of the risk of Hereditary Breast and Ovarian Cancer.

1.2. I believe that the system developed will facilitate coordination with genetics professionals in the integrated assessment of the risk of Hereditary Breast and Ovarian Cancer.

1.3. I believe that the system developed will provide professionals with a useful Clinical Decision Support System for prescribing genetic tests, unifying the criteria for information and referral, where appropriate, to genetic counseling units.

1.4. I believe that the system developed will allow people who express to their family doctor/gynecologist their concern about suffering/being able to suffer in the future from hereditary family breast/ovarian cancer because of their personal or family history, resolve doubts, diminish their worries and their level of anxiety.

Perceived Ease of Use

2.1. I think it would be easy for me to learn how to use the developed system.

2.2. I think it would be easy to acquire the necessary skills to use the developed system.

2.3. Overall, I think the developed system will be easy to use.

Social Norms

3.1. My specialty colleagues would like me to use the developed system.

3.2. My superiors would like me to use the developed system.

3.3. Genetic colleagues would like me to use the developed system.

3.4. Primary care colleagues would like me to use the developed system.

Facilitating Conditions

4.1. I think I will have the technical assistance available to solve problems associated with the developed system.

4.2. I think I will have the necessary resources to use the developed system.

Intention to Use

5.1. I intend to use the system developed for the Integrated Risk Assessment of Hereditary Breast and Ovarian Cancer.

5.2. I intend to use the system developed to manage and visualize primary care patients who are referred to as Genetic Counseling.

5.3. I intend to use the developed system to be able to visualize and know the results of the genetic tests of the patients registered in the developed system on which they decide to perform the BRCA1 BRCA2 genetic tests.

The questionnaire was designed by exchanging questions of different dimensions. Next, the correct use of the application in the departments of Gynecology and Mammary Pathology and Oncology of the HUVR was verified. Individually, in the consultations of Breast Pathology, Gynecological Oncology and in the Department of Maternal-Fetal Medicine, Genetics and Reproduction of the HUVR and the consultations of Gynecology of the Specialties Centre. Dr. Fleming from Seville.

The correct use of the application in the different computers had been checked. A training session was held with the 16

gynecologists. User manuals of the application were also provided based on the professions of the users: primary care physicians, gynecologists and geneticists. After the training, the system was validated with the defined TAM questionnaire and the results are shown in the next section.

Results

16 users completed the questionnaires. Each item is rated from 1 to 5, where 1 represents the most negative, and 5 the most positive. and NK/NA in the case of not knowing or not answering. The results are shown below:

Table 1 – Results of questionnaires

Dimensions	Item	Mean	Typical Deviation
	1.1	4,19	1,33
	1.2	4,20	1,15
Perceived	1.3	4,53	1,06
Usefulness	1.4	4,37	0,88
	Mean	4,32	0,98
	2.1	3,87	1,26
Perceived	2.2	3,85	1,21
Ease of Use	2.3	3,62	1,36
	Mean	3,71	1,23
	3.1	4,06	1,34
Social Norms	3.2	4,87	0,34
	3.3	4,25	1,36
	3.4	4,21	1,25
	Mean	4,34	0,78
Facilitating	4.1	3,47	1,50
Conditions	4.2	3,47	1,50
	Mean	3,47	1,44
	5.1	4,27	1,33
Intention to	5.2	4,60	0,91
Use	5.3	4,62	0,80
	Mean	**4,48**	0,81

The table shows the average score of each item, grouped by dimensions. It is observed that the highest score, and therefore the one that obtained the best rating from health professionals, was the Intention to Use.

On the other hand, the dimension where the professionals detected the most inconveniences was in the Facilitating Conditions. This assessment can be attributed to the learning curve of the application, given that the questionnaire was administered after training. The clinicians carried out specific questionnaires to measure acceptance, using the TAM Model, at the end of the training sessions. The results of these questionnaires are described below.

Within the Perceived Usefulness dimension, it stands out as the most valued item "*I believe that the system developed will provide professionals with a useful Clinical Decision Support System for prescribing genetic tests, unifying the criteria for information and referral, where appropriate, to genetic counseling units*" with an average score of 4,53 while the least valued was "*I believe that the developed system will facilitate coordination with primary care professionals in the integrated assessment of the risk of Hereditary Breast and Ovarian Cancer*" with an average score of 4,19. However, this item was also rated positively, taking into account that the maximum value of the scale is 5.

Within the Perceived Ease of Use dimension, it stands out as the most valued item "*I think it would be easy for me to learn how to use the developed system*" with an average score of 3,87 while the least valued was "*Overall, I think the developed system will be easy to use*" with an average score of 3,62.

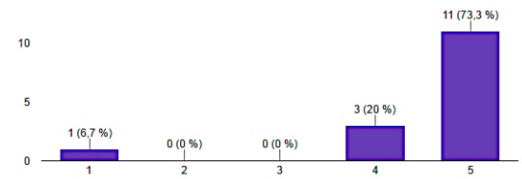

Figure 1 – "I believe that the system developed will provide professionals with a useful Clinical Decision Support System for prescribing genetic tests, unifying the criteria for information and referral, where appropriate, to genetic counseling units" (NK/NA: 1 user)

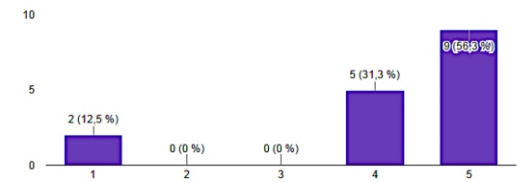

Figure 2 – "I believe that the developed system will facilitate coordination with primary care professionals in the integrated assessment of the risk of Hereditary Breast and Ovarian Cancer"

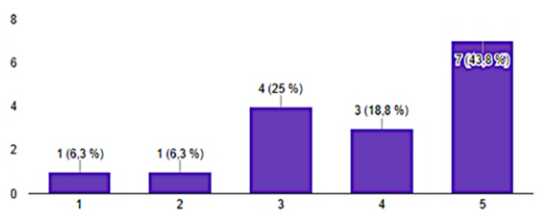

Figure 3 – "I think it would be easy for me to learn how to use the developed system"

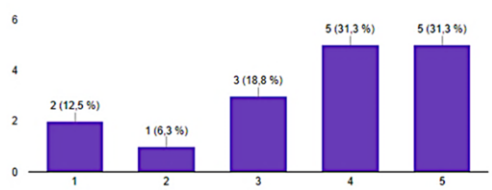

Figure 4 –" Overall, I think the developed system will be easy to use"

Within the Social Norms dimension, it stands out as the most valued item "*My superiors would like me to use the developed system*" with an average score of 4,87, while the least valued item was "*My specialty colleagues would like me to use the developed system*" with an average score of 4,06. However, this item was also rated positively, taking into account that the maximum value of the scale is 5.

Within the Facilitating Conditions dimension, the two items scored an average score of 3,47.

Finally, within the Intention to Use dimension, the most valued item is "*I intend to use the developed system to be able to visualize and know the results of the genetic tests of the patients registered in the developed system on which they decide to perform the BRCA1 BRCA2 genetic tests*" with an average score of 4.62 while the least valued was "I intend to use the system developed for the Integrated Risk Assessment of Hereditary Breast and Ovarian Cancer" with an average score of 4.27. However, this item was also rated positively, taking into account that the maximum value of the scale is 5.

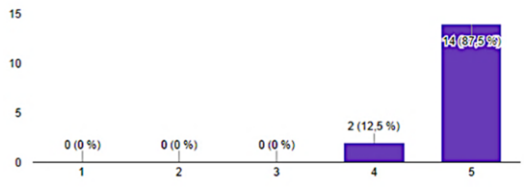

Figure 5– "My superiors would like me to use the developed system"

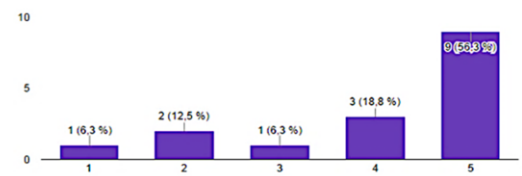

Figure 6 – "My specialty colleagues would like me to use the developed system"

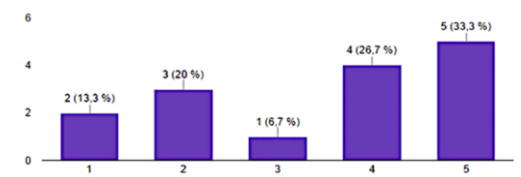

Figure 7 –" I think I will have the technical assistance available to solve problems associated with the developed system" (NK/NA: 1 user)

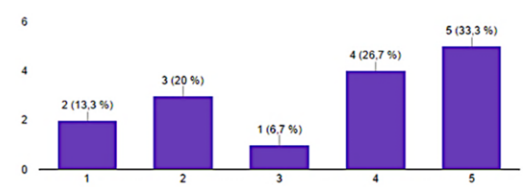

Figure 8 –" I think I will have the necessary resources to use the developed system" (NK/NA: 1 user.)

Discussion

The dimension of the questionnaires that obtained the best rating from health professionals was the Intention to Use. Therefore, this score is relevant for predicting the future use of the developed tool in this project. Within the Intention to Use dimension, the most valued item was "*I intend to use the developed system to be able to visualize and know the results of the genetic tests of the patients registered in the developed system on which they decide to perform the BRCA1 BRCA2 genetic tests*" with an average score of 4.62, meaning other items of this dimension also rated positively.

Besides, the dimension where the professionals detected the most inconveniences was in the Facilitating Conditions, due to the learning curve of the application, given that the questionnaire was administered after training. The two items scored an average score of 3,47, and these scores could be improved. For instance, guaranteeing and providing the required technical assistance and the necessary resources to use the developed system.

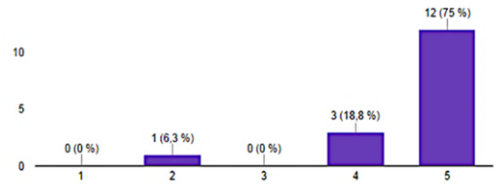

Figure 9–"I intend to use the developed system to be able to visualize and know the results of the genetic tests of the patients registered in the developed system on which they decide to perform the BRCA1 BRCA2 genetic tests"

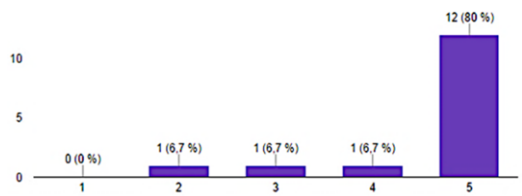

Figure 10 – "I intend to use the system developed for the Integrated Risk Assessment of Hereditary Breast and Ovarian Cancer" (NK/NA: 1 user)

Finally, other dimension with a low score is Perceived Ease of Use, where stands out as the most valued item "*I think it would be easy for me to learn how to use the developed system*" with an average score of 3,87 while the least valued was "*Overall, I think the developed system will be easy to use*" with an average score of 3,62. So it is essential to promote the realization of training sessions and the use of user manuals. Besides, these items rated positively, taking into account that the maximum value of the scale is 5.

In general, the results of the questionnaires show favorable and positive feedbacks. Therefore the use of the developed system is recommended to clinicians for supporting the prescription of genetic tests in the gynecological cancer risk.

According to analysis of the feedbacks, the developed CDSS will be useful for clinical practice. As a result, a tool adapted to the needs and preferences of clinicians, who have also formed part of the project's research team for the definition of requirements, the design of the tool and the implementation of clinical knowledge in the CDSS, has been achieved.

We are working on the development of Artificial Intelligence and Data Mining algorithms. So that through these predictive algorithms can potentially improve the recommendations following the guidelines of the SEOM for the prescripting genetic tests, since currently there are a high percentage of patients, who were prescribed the test and the result turned out

negative. In this way, the system will evolve into a learning healthcare system.

Predictive models are using information collected in electronic medical records, in addition to that referred to in clinical guidelines, to develop more efficient algorithms, with greater precision in prescribing the genetic tests. In this way, we are analyzing the set of variables relevant to this prescription, as well as their association with the target variable.

Conclusions

In this study, a CDSS for prescribing the genetic tests in BRCA1 and BRCA2 and preventing gynecological cancer risks was designed and evaluated in the Virgen del Rocío University Hospital. The developed technological architecture allows general practitioners and gynecologists to classify patients as low risk (who are not required a specific treatment) or high risk (who should be attended by the Genetic Council). Also, it provides recommending related to prescription of genetic tests and their genetic risk of suffering gynecological cancer.

An evaluation of the developed CDSS, for usefulness perceived by clinicians, the perceived ease of use and the intention to use, was also carried out and the results showed positive feedbacks.

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