

The Use of Telemedicine for Genetic Counseling in Oncology

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A Utilização de Telemedicina para o Aconselhamento Genético em Oncologia

La Utilización de Telemedicina para la Asesoramiento Genético en Oncología

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Abstract

Introduction: The development of medicine and the discovery of the human genome revolutionized the knowledge, monitoring and treatment of cancer that favored the utilization of new methods of diagnosis, treatment for several types of neoplasms and genetic counseling. Although difficult to access, genetic counseling may allow the patients with suspected hereditary cancer, to reduce the rates of morbidity and mortality by this disease and ensure improvement in their quality of life. **Objective:** Demonstrate the state of the art of genetic counseling in oncology using the digital telemedicine tool. **Method:** An integrative literature review conducted between December 2018 and March 2019, using the PubMed, SciELO and BIREME databases, utilizing the words registered in the MeSH and Health Sciences Descriptors (DeCS), respectively, in English, Genetic Counseling AND Telemedicine OR eHealth AND Oncology. **Results:** 16 articles were found and of these, eight were excluded. Thus, seven articles that met the inclusion criteria showing data about patient satisfaction with cancer-oriented counseling services, its advantages and disadvantages remained. **Conclusion:** Although there are few studies addressing the experience of genetic counseling in oncology, this low cost practice has been shown to be efficient and well accepted by the patient and the professional, providing proper responses mainly in places of difficult access.

Key words: Genetic Counseling; Telemedicine; Oncology.

Resumo

Introdução: O desenvolvimento da medicina com a descoberta do genoma humano revolucionou o conhecimento, o acompanhamento e o tratamento do câncer, proporcionando a utilização de novos métodos de diagnóstico e tratamento para diversos tipos de neoplasias e permitindo o aconselhamento genético. O aconselhamento genético, embora de difícil acesso, pode vir a permitir aos pacientes com suspeita de câncer hereditário a diminuição dos índices de morbidade e mortalidade por essa doença e proporcionar uma melhora na qualidade de vida dos pacientes. **Objetivo:** Demonstrar o estado da arte do aconselhamento genético em oncologia, utilizando a ferramenta digital da telemedicina. **Método:** Foi realizada uma revisão integrativa de literatura, entre dezembro de 2018 e março de 2019, a partir das bases de dados PubMed, SciELO e BIREME, por meio das palavras cadastradas no MeSH e nos Descritores em Ciências da Saúde (DeCS), respectivamente, em inglês, *Genetic Counseling AND Telemedicine OR eHealth AND Oncology*. **Resultados:** Foram encontrados 16 artigos; destes, oito foram excluídos. Restaram sete artigos que atendiam aos critérios de inclusão, demonstrando dados sobre a satisfação do paciente para com os serviços de aconselhamento genético voltados para o câncer, assim como suas vantagens e desvantagens. **Conclusão:** Embora ainda haja poucos estudos relatando experiências da prática do aconselhamento genético em oncologia, essa prática tem se demonstrado eficiente, de baixo custo e de boa aceitação do paciente e do profissional, suprimindo necessidades principalmente nos locais de difícil acesso.

Palavras-chave: Aconselhamento Genético; Telemedicina; Oncologia.

Resumen

Introducción: El desarrollo de la medicina con el descubrimiento del genoma humano revolucionó el conocimiento, seguimiento y terapia del cáncer, permitiendo la utilización de nuevas técnicas de diagnóstico y terapias para diferentes tipos de neoplasias, además del asesoramiento genético. El asesoramiento genético, aunque de acceso difícil, puede permitir a los pacientes con sospechas de cáncer hereditario la disminución de los índices de morbilidad y mortalidad para esa enfermedad y proporcionar una mejora en su calidad de vida. **Objetivo:** Demostrar el estado del arte del asesoramiento genético en oncología utilizando la herramienta digital de la telemedicina. **Método:** Se realizó una revisión integrativa de literatura entre diciembre de 2018 y marzo de 2019, a partir de las bases de datos PubMed, SciELO y BIREME, por medio de las palabras catastradas en el MeSH y en el Descriptores de Ciencia de la Salud (DeCS), respectivamente, en inglés *Genetic Counseling AND Telemedicine OR eHealth AND Oncology*. **Resultados:** Se han encontrado 16 artículos; de estos, 8 fueron excluidos. Así, quedaron siete artículos que atendían a los criterios de inclusión demostrando datos sobre la satisfacción del paciente hacia con los servicios de asesoramiento genético dirigidos al cáncer, así como sus ventajas y desventajas. **Conclusión:** Aunque todavía hay pocos estudios relatando experiencias de la práctica del asesoramiento genético en oncología, esta práctica se ha demostrado eficiente, de bajo costo y de buena aceptación del paciente y del profesional, supliendo necesidades principalmente en los lugares de difícil acceso.

Palabras-chave: Asesoramiento Genético; Telemedicina; Oncología.

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INTRODUCTION

The development of oncogenetics is relatively recent and it is estimated that 5% to 10% of the cancer cases are hereditary. In these cases, there is the possibility of preventive measures to reduce the risks and early detection with increase of the chances of cure for certain types of cancer. Based in risk mapping for each individual, it is possible to identify whether the chances of developing cancer are high or not and adopt the required measures. Genetic counseling is a routine procedure in some developed countries and in them, mortality by several tumors has diminished satisfactorily, a factor that may have contributed for the fall of the mortality¹.

Some general findings alert for a possible genetic cause, among them, the presence of rare tumors, age lower than usual, several cases of cancer in the family, especially in first- and second-degree relatives and history of two or more neoplasms in the same person. In some cases, the presence of certain neoplasms combined to age and/or gender of the patient can lead to the indication of the evaluation, as, for instance, breast cancer before 50 years old or breast cancer in males. In certain cases, the tumor histology on its own is indication for counseling and genetic tests as is the case of ovary epithelial carcinomas and carcinoma of the choroid plexus².

Genetic counseling is based in the personal history, family history and specific findings of physical exam and results of diagnostic tests, particularly the genetic. The extensive family history with particular search for benign, malignant neoplasms and autism in the family, is essential to identify whether there is possibility of hereditary predisposition to malignant neoplasms or if there are signs of sporadic condition. In practice, a genealogy tree of the family is created manually or through software. Computer programs can, through mathematics algorithms, help to quantify the risk of genetic predisposition of the patients².

If after the mapping there is suspicion of hereditary cancer, procedures for molecular diagnosis are performed whenever they are available. The oncogeneticist should know what the genes are to be evaluated and which technique to be used in each case. Only then when confirmed the presence of the flaw of the gene that can favor the appearance of cancer, pathogenic variant or probably pathogenic, the patient receives specific guidelines for prevention and tracking of neoplasms².

Genetic syndromes as the Li-Fraumeni Syndrome, Lynch Syndrome, Hereditary Breast and Ovarian Cancer Syndrome, Familial Adenomatous Polyposis, Multiple Endocrine Neoplasms and Von Hippel Lindau Syndrome are some examples of familiar genetic syndromes that increase the chance of cancer and that has protocols of

follow up for preventive management. Once identified the genetic alteration responsible for the syndrome in a certain family, the early diagnosis and the adoption of preventive measures in non-affected relatives is possible².

The syndrome of predisposition diagnosis is a medical act, but physicians and professionals from other areas of expertise can help in the genetic counseling. In some countries, there are professionals specifically skilled to be genetic counselors³.

The regulation of telemedicine in Brazil needs updating, some as those addressed by the revoked Resolution Federal Council of Medicine (CFM) number 2.227/2018⁴. Currently, it is regulated by Resolution CFM number 1,643/2002⁵ and defined as a model of services where accredited experts provide remote care through “interactive methodologies of audiovisual communication and data with the objective of assistance, education and research in Health”⁵. This modality of service is especially important for patients living in poor medical resources areas. Works conducted formerly suggest that telemedicine in genetics, telegenetics, allows to provide access to services for those who face geographic barriers, improve cost-effectiveness and meet the increasing demand for services⁶. This issue was also emphasized in the Process-Consultation number 8.732/2009 – REPORT CFM number 17/2018, that emphasizes that genetic counseling can and must be done through electronic communication means, with the existing profile of demography of the specialty and the lack of expert professionals in medical genetics in Brazil⁶.

Because of the low availability of professionals experts in the practice of genetic counseling in oncology and the raising number of referrals for evaluation in relation to the syndrome of genetic predisposition to cancer, the present article intends to demonstrate the state of the art of genetic counseling in oncology, utilizing telemedicine as a tool to ensure access to information.

METHOD

An integrative review of the literature was conducted between December 2018 and March 2019. Firstly, to obtain the number of studies about the utilization of telemedicine in the practice of genetic counseling in oncology an exploratory study with the objective of calculating the amount of indexed articles in different databases was conducted, among which: a) Scientific Electronic Library Online (SciELO); b) Latin American and Caribbean Health Science Literature (LILACS); and c) U. S. National Library of Medicine (PubMed).

After investigating the Descriptors of Health Sciences (DeCS), the following descriptors were selected “*Aconselhamento Genético*”, “*Telemedicina*”, “*Telessaúde*”

and “Oncologia” for the comparative study in the databases SciELO and LILACS. After searching the Medical Subject Headings (MeSH), the following descriptors were chosen “Genetic Counseling”, “Telemedicine”, “eHealth” and “Oncology” for the comparative study in the databases PubMed.

The inclusion criteria were articles from January 2009 to October 2018 in Portuguese and in English; and the exclusion criteria were independent literature reviews in relation to the methodology applied. Because of the inexistence of articles in the databases SciELO and LILACS, only articles of the database PubMed were used, resulting in 16 articles. After the application of the exclusion and inclusion criteria, seven articles remained whose characteristics are shown in Table 1 and in Table 2, the advantages and disadvantages with the results.

RESULTS

Through the search, it was encountered 16 articles selected with inclusion and exclusion criteria. Therefore, seven articles were included.

DISCUSSION

The use of telegenetics is increasingly seen as an alternative of access to information on health in remote areas⁷. A previous study of telegenetics-pilot involving patients of Maine, counseled for several genetic conditions, including the susceptibility to cancer, demonstrated a positive impact for medical

decision taking among the study subjects and high level of confidence in the healthcare providers. The study concluded that while moving the information instead of the patient, telemedicine promises to improve healthcare services and tears down the barrier of where and when genetic services are not provided⁸.

The articles evaluated demonstrated the patient’s satisfaction with the services of cancer genetic counseling, because many advantages as accessibility to this type of service and possibility of consultation without travel were met. Overall, few already expected disadvantages were observed for the services of telemedicine as the concern with the risk of violation of confidentiality of the health information⁹⁻¹¹. Safety and privacy issues with electronic communication were the most common disadvantages reported, which was also found in other studies which utilize health information technologies and communication^{12,13}.

Hilgart et al.¹⁴ demonstrated that, although the interviewee preferred the consultation in-person, telemedicine could benefit the persons who cannot travel and that have scarce accessibility because of their place of residence both because of the distance or transportation means¹⁴.

When compared the practice of virtual *versus* in-person counseling, a telegenetics randomized study discovered that the services of telemedicine are less expensive than in-person counseling still associated with high satisfaction among the clients, being a viable alternative and financially better for health systems, in addition of granting access in remote places to genetic information^{10,15}.

Table 1. Characteristics of the studies analyzed

Author/ year	n	País Country	Câncer Cancer	Aconselhamento Counselling
Bradbury et al., 2016	98	USA	Colorectal, breast and ovary	Pre and post-test
Buchanan et al., 2015	162	USA	Colorectal, breast, ovary, other/multiple	Pre and post-test
Zilliacus et al., 2010	18	Australia	Breast and Ovary	Post-test
Hilgart et al., 2012	225	Wales	Breast/ovary, colorectal, stomach and other	--
Zilliacus et al., 2010	15	Australia	Breast and Ovary	--
McDonald et al., 2014	149	USA	Breast and Ovary	Pre-test
Zilliacus et al., 2011	195	Australia	Breast and Ovary	Pre and post-test

Table 2. Characteristics of teleconsultation and advantages and disadvantages of genetic counselling utilizing telemedicine

Author/year	Teleconsultation	Advantages	Disadvantages
Bradbury et al., 2016	Virtual consultation was realized with a genetic counselor of University of Pennsylvania. In the pre-test counseling with support of the local clinic and technical support. In the post-test counseling, 63% of the patients who needed this counseling had the local physician present, the other patients had follow-up consultation	Low cost, without necessity of long travels to large cities, possibility of scheduling the counseling in parallel with consultation in-person with the local physician who follows up the patient	Technical difficulties. Two participants felt lack of in-person support of the counselor
Buchanan et al., 2015	One only counselor of Duke University attended all the patients via telemedicine from the university to the regional clinic where the patient was being followed up. The counselor and the patient communicated in real time with a screen and visualized the documents commonly used during counseling in a second screen	Lower cost per patient through telemedicine (\$ 106) in comparison with in-person counseling (\$ 244)	Rate of 32% of the participants reported it would be a disadvantage not to have in-person contact with the counselor
Zilliaccus et al., 2010	In this study, 18 participants had genetic counseling for telemedicine utilizing the system Tandberg™. The sizes of the monitors were, in average, 42" screens. The cameras allowed a horizontal view of 72° and a vertical of 43.5°. The cameras utilized have the capacity of accommodating three persons (genetic counselor, patients and support person) and also have the capacity of broadening to capture the upper part of the body and the face of one person (usually genetic clinician) to augment the facial expression and make the consulting more welcoming	Reduction of travels and associated costs	Lack of emotional support
Hilgart et al., 2012	Investigators through databases of previous studies invited patients and staff to participate of a study to evaluate the accessibility and viability of telemedicine in the services of cancer genetics. Many aspects of telemedicine for genetic counseling proposed in the study were highly acceptable for the patients, including an electronic version of the questionnaire of family history, with emails for cancer genetic consultations and support to computerized decision	Facility of access to information via e-mail and other electronic methods without the necessity of traveling to a reference center	Impersonality and lack of human contact because of electronic methods
Zilliaccus et al., 2010	This work planned to evaluate the satisfaction and experience of the professionals who realized genetic counseling through telemedicine in oncologic patients, and, therefore, semistructured interviews were performed with geneticist physicians providing services in New South Wales, Australia. The interviews explored experiences, objectives perceived in the service, satisfaction, advantages and disadvantages of technology	It was verified more effectiveness, minimization of travels and associated costs, more access to rural areas	No creation of bonds

to be continued

Table 2. continuation

Author/year	Teleconsultation	Advantages	Disadvantages
McDonald et al., 2014	The genetic counseling visit was realized through telemedicine and assessed by the application of a questionnaire with 27 questions that evaluated demographic data, personal and family history of cancer, as well as evaluation of risk of susceptibility to cancer, knowledge about genetic tests in oncology, acceptability of characteristics that reflect models of services of genetic counseling, qualification and evaluation of genetic counseling	Accessibility of contacting a qualified professional providing care and counseling remotely	--
Zilliacus et al., 2011	The genetic counseling visits involved a geneticist physician via telemedicine, in addition to a local present geneticist counselor with the patient in its origin city in individuals with hereditary breast and ovary cancer, where one group received virtual counseling and other, in-person. The participants filled out self-applicable questionnaires before and one month after genetic counseling, demonstrating the expectations and evaluating the service	Telemedicine consultation had better performance than in-person consultation in meeting the expectations	--

In relation to the biopsychosocial aspects involved in genetic counseling and cancer diagnosis itself, Zilliacus et al.¹¹ did not find statistically significant difference in the topics generalized anxiety, depression, perception of empathy of the genetic clinician and perception of empathy of the genetic counselor when compared to virtual genetic *versus* in-person counseling¹¹.

In Wales, it was reported the case of a woman with recent diagnosis of cancer where telemedicine was unable to meet her needs of psychosocial support, said necessities that can be explained by the phase of acceptance of the disease experienced by the patient with the recent diagnosis¹⁴.

In regard to the patients expectations, in a North American article, genetic counseling in oncology had significant better performance than in-person counseling to meet the patients expectations, which can also be justified by the fact that is something new and unknown, creating surprises for the interviewees¹⁶.

In USA, in relation to the professional angle of genetic counseling, it was noticed that when the consultation occurred with the presence of the genetic counselor, of the physician and of the patient, ensuring co-participation, it has occurred a high level of information exchange, offering a useful structure for genetic counseling in telemedicine, complementary roles among the professionals and effective services provided¹⁷.

Remarkably, telemedicine, through videoconference offers an alternative model of genetic counseling for

hereditary cancer in rural populations and difficult geographic access. This method allows geneticist physicians to remain in their base hospital, connecting with the patients by videoconference for genetic counseling. Telemedicine by videoconference is being used successfully in other medical areas as psychiatry¹⁸, dermatology¹⁹, pediatrics²⁰ and oncology²¹.

CONCLUSION

With the increase of the indications of genetic tests for investigation of hereditary predisposition to cancer, there is an increasing necessity of providing access of quality to genetic counseling to ensure equity of access in different geographic regions. Videoconference appears to be a viable technological alternative.

Although there are few studies reporting experiences of genetic counseling in oncology, in some countries, this modality of consultation has been shown to be effective, low cost and good acceptance of the specialist and the patient, meeting demands mainly in places of difficult access.

The model of virtual genetic counseling can be an important tool in underdeveloped and in development countries where there is difficulty of access to health services. Always bearing in mind that technology is a support tool in specific cases and in no circumstance replaces the work of the professional physician.

CONTRIBUTIONS

The authors contributed equally in all the phases of the study and approved the final version to be published.

DECLARATION OF CONFLICT OF INTERESTS

There is no conflict of interests to declare.

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