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PATIENT FACING SYSTEMS

Telegenetics: an Update on Availability and Use of Telemedicine in Clinical Genetics Service

Irena Vrečar[1] · **Dimitar Hristovski**[2] · **Borut Peterlin**[1]

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Abstract Although telegenetics as a telehealth tool for online genetic counseling was primarily initiated to improve access to genetics care in remote areas, the increasing demand for genetic services with personalized genomic medicine, shortage of clinical geneticists, and the expertise of established genetic centers make telegenetics an attractive alternative to traditional in-person genetic counseling. We review the scope of current telegenetics practice, user experience of patients and clinicians, quality of care in comparison to traditional counseling, and the advantages and disadvantages of information and communication technology in telegenetics. We found that live videoconference consultations are generally well accepted by both clients and clinicians, and these have been successfully used in several genetic counseling settings in practice. Future use of telegenetics could increase patients' access to specialized care and help in meeting the increasing demand for genetic services.

Keywords Telegenetics · Genetic counseling · Clinical genetics · Telemedicine · E-genetics

Introduction

Telemedicine is increasingly used as an innovative approach to medical care delivery using electronic and communication technologies for diagnosis, monitoring, and therapy [1]. Areas using telemedicine include dermatology, radiology, psychiatry, cardiology, diabetes, fetal ultrasound, pediatric services, and more recently, clinical genetics (also called telegenetics) [2, 3].

The one in 17 Europeans who are affected by rare genetic diseases during their lifetime may require clinical genetic services [4]. For several diseases or group of diseases the expertise for diagnosis and management is scattered and not easily accessible in the national health systems. Therefore there is increasing demand for cross-border health services. Telegenetic services offer an opportunity to access genetic counseling services without the need for patients or professionals to travel. Moreover, telegenetics may provide an effective method for teleconsulting and professional knowledge exchange.

Additionally, an exponential increase in genetic knowledge based on new methodologies, including next generation sequencing tests, is leading to increasing demand for genetic services. The number of available genetic tests increased from 300 in the early 1990s to over 2000 in 2011 [5]. As we increasingly understand the role of genetics in common diseases, referrals are also expanding to the general population [6]. A solution for keeping pace with demand and improved technology might be provided by the introduction of information technology in genetics service delivery [5]. Alternative models of service provision, including genetic counseling over the phone and videoconferencing, have been explored in order to improve equality of access and cost efficiency as well as help in meeting an increasing demand for genetic services [1].

Since telegenetics implementation in practice is still rather limited and not widely accepted, we aimed to review the scope of telegenetics use, user experience (including patients' satisfaction and genetic counselors opinions), and existing barriers to wider availability and use of telegenetics services.

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✉ Borut Peterlin
borut.peterlin@guest.arnes.si

[1] Clinical Institute of Medical Genetics, University Medical Centre Ljubljana, Ljubljana, Slovenia

[2] Institute for Biostatistics and Medical Informatics, Faculty of Medicine, Ljubljana, Slovenia

Telegenetics consulting modalities

Telemedicine in general encompasses four major modalities:

1. *Live video (synchronous).* Using audiovisual telecommunications technology for live, two-way interaction between a person (patient, caregiver, or provider) and a provider. This type of service may serve as a substitute for an in-person encounter and is also known as "real-time".
2. *Store-and-forward (asynchronous).* Recorded health history (e.g., videos such as ultrasound, digital images such as x-rays and photos, and various test results) is transmitted through a secure electronic communications system to a practitioner, usually a specialist, who evaluates the case without live interaction.
3. *Remote patient monitoring.* Electronic transmission of personal medical data collected from an individual in one location (usually home) to a provider in a different location for use in care and related support.
4. *Mobile health (mHealth).* The use of mobile communication devices such as cell phones and tablet computers to provide health care and education.

From the four modalities listed above, the most frequently used in telegenetics is the first one (live video). It can be done with a standard desktop computer equipped with a video camera and a microphone, or with special dedicated video conferencing hardware such as a telemedicine cart. In telegenetics, video and audio applications are used to provide real time, online counseling or evaluation of patients. Virtual consultation largely involves synchronous video communication between genetic counselor and patient. There is the possibility of using various supportive tools similar to those used for in-person genetic counseling, like simultaneously viewing disease information brochures or supportive drawings [7]. Telegenetics has primarily been used for patients who do not require a physical examination; however, pilot studies in children suggest that morphological assessment with real-time, high-definition images transmitted by videoconference could be performed [3]. In its advanced mode telemedicine includes using a specialized portable workstation, which enables visualizing and measuring body parts and functions [8].

Although not yet used a lot in telegenetics, the second modality (store-and-forward or asynchronous) has a lot of potential for use. It can be especially helpful for communication between genetics specialists for getting opinion on rare cases or for help in the interpretation of the results of new genetic tests. For this modality, if the amount of data to be transferred is small, secure email can be used. However, if the amount of data to be transferred is large then cloud services are more appropriate. In any case, it is recommended that any sensitive personal data is encrypted before leaving the originating computer.

Telegenetics implementation in health systems

Telemedicine is particularly appropriate for care in which verbal communication takes precedence over physical examination [1]. Since genetic counseling consists largely of communication, information exchange, education, and psychosocial care, telemedicine shows great promise for implementation in genetics. Such care involves both communication between professionals and patients, and exchange of expert opinions between genetic professionals about patients [8].

First application of telegenetics was reported in the area of cancer in UK in 1998, when a pilot project was conducted in Wales [9]. Since then most reports on telegenetics implementation focus on hereditary cancer genetic counseling, where physical examination is usually not essential; effectiveness, cost and feasibility of using technology in this setting have been evaluated [10–12]. Current genetic practice requires at least two genetic sessions including pre- and post-test counseling, preferably in person. Telegenetics in cancer genetic counseling has been shown to improve access in underserved areas and for elderly patients for lower cost with acceptable satisfaction rates [11]. One hereditary cancer counseling study showed that the two most important characteristics of a cancer genetic service for the study respondents were direct one-on-one counseling and the competence of the genetic provider [13].

Predictive genetic counseling involves several genetic counseling sessions following a predictive testing protocol, which can prove to be time-consuming and logistically difficult in remote areas. Telehealth predictive testing for Huntington's disease was shown to be an acceptable alternative in improving access where geographic barriers may further deter individuals from predictive testing in already difficult situations [14].

Telemedicine has furthermore been used for diagnosis in pediatric and fetal genetics, where high definition images of the assessed children and fetal morphology on ultrasound can be transmitted by videoconference [3, 15, 16]. Quality of the transmitted image and bandwidth transmissions are especially important in fetal ultrasound, another important telemedicine application, as quality of ultrasound assessment in fetal medicine vary significantly between tertiary centers and general gynecological practice [16].

The use of telegenetics has been explored through various pilot studies, conducted in Australia, Canada, Netherlands, United Kingdom and in the United States, but so far these new approaches to service delivery seem to be rather slow to implement in routine clinical practice, where wider acceptance might be gained via standardization and clear expert recommendations.

Patients' satisfaction

Regarding patient experiences in telegenetics, all the studies report high levels of patient satisfaction [1, 8]. Telegenetics is

shown to be comparable favorably to in-person genetic counseling with respect to patient satisfaction and knowledge [5]. A randomized trial of cancer telegenetics versus in-person counseling showed high satisfaction among telegenetics counseling participants [11]. An Australian study of parents' satisfaction in a pediatric setting found it was an acceptable alternative to a standard face-to-face consultation. This study involved genetic assessment of intellectual disability, including morphological exam of the child [3]. In a randomized study by Gattas on assessment of telegenetics (mostly oncogenetics) consultations patients responded positively when asked if they would be happy to use telemedicine in the future [17]. Patients' satisfaction levels were generally similar in the telegenetics group and in the comparison group receiving traditional face-to-face counseling. As benefits to telegenetics, patients reported convenience, reduced travel time and associated costs, and reduced waiting times [1]. Some patients also said they felt more comfortable, at-ease and receptive at home than in a hospital setting.

An online survey showed 80 % of parents of patients with Down, Williams', and DiGeorge syndrome showed an interest in mobile health (m-health) applications, wanting to become more involved in their child's disease management [18]. The conclusion would be that telehealth applications can lead to patient empowerment and are perceived as useful for many patients with genetic conditions, not only for those living far from healthcare centers.

Genetics professionals opinions

The number of genetic counselors using telegenetics is low [6, 8]. An online survey completed by 104 European genetics professionals showed telephone genetic counseling was used by 17%, while videoconferencing facilities were available to 24%, only 9% of them used these for patient counseling [8].

Health care professionals have a moderately positive attitude toward telegenetics, but they are concerned about technical imperfections, lack of rapport building, and the difficulty in detecting nonverbal cues and assessing patient emotions and understanding [2, 8, 19]. There is some concern, particularly among nursing staff, that communication and technical restrictions may impair their ability to observe the nonverbal behavior of patients [1]. However, in one study, genetic counselors' experience with telephone communication of BRCA test results showed that videoconferencing with the use of visual aids allows for at least some non-verbal communication and might therefore be preferable to telephone communication [19].

Genetic practitioners in New South Wales, Australia perceived videoconferencing favorably due to increased efficiency and convenience for clinicians, minimized travel for the patient, reduced costs, and increased access to rural areas [2].

Barriers to availability and use of telegenetics, lack of national regulations and privacy issues

Several practical and psychological barriers contribute to the limited use of telegenetics in Europe. Genetic professionals participating in the study on telemedicine uptake reported various barriers to greater current use of telemedicine, including practical constraints (lack of resources and supportive regulations), lack of professional support / knowledge, to lack of perceived suitability and need [8]. Practical barriers like lack of technical equipment with appropriate communication bandwidth and knowledge also have an impact.

Ensuring data safety and confidentiality of medical information on the web is a growing concern [20]. Encryption of the connection to ensure a secure exchange of sensitive patient information is advised by American Telemedicine Association [8]. Currently, the majority of telegenetics consultations are done with patients visiting local medical institutions, which in most cases have implemented various technical measures for data safety and confidentiality. However, in the future, increased number of telegenetics consultations are expected to happen with patients participating from their homes, which will create new challenges in regard to security and confidentiality.

Related information technologies are having an impact on telegenetics. The introduction of web-based family history tools and pedigree drawing programs enables online genetic burden assessment, saves time, and prepares for genetic consultation [21], while electronic health records and new e-health applications are likely to increase telegenetics facilitation [8].

Conclusions and future perspectives

Telemedicine shows great potential for implementation in genetics, where a high level of expertise can be scattered in various specialized genetic centers worldwide. Genetic service is predominantly based on counseling and increasing demand for genetics services calls for implementation of new approaches to service delivery including the use of new technologies. Although telemedicine has been successfully implemented in various other medical specialties, its usage in genetics has so far been relatively scarce. Our review shows recent implementations of new technologies for genetics consultations are promising with mostly positive experiences reported both from patients and genetic professionals. Our group has tested the telegenetics application as part of the international collaboration project SIGN (Slovene Italian Genetic Network) in the field of reproductive genetic counseling, where we established good satisfactory response rates among the patients (unpublished). We conclude experiences with telegenetics implementation are generally positive in the

various fields that have so far been tested, with high levels of satisfaction and improved knowledge among patients. Inspite of these findings and the fact of the first telegenetics implementation taking place over 18 years ago, there has been no widespread adoption of telegenetics in routine clinical praxis. This could partly be explained by the lack of international expert guidelines that would include opinions on patient data safety, maintaining quality of care and improve awareness among professionals. In order to overcome these barriers, we suggest standardization of the telehealth in genetics as a tool and establishing formal expert recommendations, formulated by the responsible genetic committees with the aim to foster wider acceptance of telegenetics as a valid genetic counseling service delivery mode.

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