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HONR 4980

Genetic Counselors' Attitudes towards Telegenetics: The Good, The Bad, The In-Between

Abstract:

Given that the global coronavirus pandemic has necessitated a quick and vast transition to telehealth, the purpose of this study is to explore these changes in the field of genetic counseling. To examine the perceived effectiveness of telegenetics from the perspective of genetic counselors, I conducted in-depth, semi-structured interviews with two genetic counselors in which each participant was asked six questions. The interviews were recorded, transcribed, and then analyzed using standard thematic analysis techniques for qualitative data analysis. The analysis revealed two main themes: benefits and drawbacks of telegenetics, with two sub-themes under each. The major benefits are increased patient accessibility and work productivity, while the main drawbacks are limited control over the environment during the encounter and challenges to the provider-patient relationship. Findings were mixed, however, regarding whether telegenetics compromises the provider-patient relationship and so further research is needed in order to establish the if there is a negative correlation between telegenetics and the ability to connect with patients. The results of this study suggest that telehealth is viewed favorably among genetic counselors, although this study also identifies several areas for further improvement that are necessary to address, especially given that the widespread use of telegenetics is likely to continue long after COVID-19 has been contained.

Introduction:

Genetic counselors are individuals who help and support patients in informing and making decisions about their genetic health. In a normal setting the majority of genetic counseling meetings occur in person. However, in light of the recent SARS-CoV-2 global outbreak normal proceedings have been paused. In order to maintain proper COVID procedures social distancing was enforced, and as a result genetic counseling meetings have moved virtual. With the different delivery methods research has not established how genetic counselors view telegenetics, and the impact it has on the relationship of a genetic counselor and their patients.

Background:

Background of Genetic Counseling

Genetic counseling first began in the mid-1900s, which coincides with the beginning of genetic testing. While early genetic counseling primarily focused on family history modern genetic counseling has developed much farther. Nowadays genetic counselors are healthcare professionals who help to educate individuals and assist them in understanding and making decisions about their genetic health. Those who may benefit from genetic counseling include, but are not limited to those with a “child with abnormalities of growth, developmental delays, mental retardation, blindness or deafness” (Kumari, 2021, pg 2). Genetic counseling can also be helpful to those with a “previous child, parents or close relatives with an inherited disease, congenital anomalies, metabolic disorders, developmental disorders or chromosomal abnormalities” (Kumari, 2021, pg 2). As more genetic tests are discovered and more research is allotted toward understanding different diseases/disorders, genetic knowledge and the need for genetic counselors continue to grow. In genetic counseling there are also sub-specialties including, prenatal, oncology, neurology, cardiology, and pediatrics. Prior to the COVID-19 pandemic the majority of genetic counseling meetings were done in-person, or non-positive results were delivered by phone. However, in order to ensure the health and safety of people during COVID-19 pandemic most genetic counseling meetings have been moved virtually. Genetic counseling has dramatically increased since the mid-1900s, and it continues to become more important as genetic testing is becomes more readily available.

Effectiveness and Satisfaction of Genetic Counseling

With any healthcare job it is important that both patients and providers view their services as satisfactory. Having effective meetings is important in order to make sure that patients’ questions, needs, and quality of care have been properly met. It is important for genetic counselors’ to view their meetings as successful because it can prevent burnout and other psychological effects caused by this job (Aalfs et al, 2017). One group of researchers conducted a project to study how what patients viewed as being effective in a genetic counseling meeting. Prior to the meeting most counsees had no idea what the meeting would entail, but after the meeting most counsees had high levels of satisfaction (Macleod et al, 2002). Several areas of satisfaction were addressed by patients. It was found that patients were very satisfied that their concerns were heard and addressed, and that they had someone who would advocate for them. Patients also found it very helpful that the genetic counselor had the ability to order tests, screenings, and reach out to other healthcare specialists (Macleod et al, 2002). Some participants in this study expressed that they had been overlooked by the healthcare system. Genetic counselors addressed family history, heritability, and whether patients and their families were at risk or not. Being able to get answers to questions and know preventive steps to take all contributed to high satisfaction ratings (Macleod et al, 2002). Results from the study also stated that “a number of participants spoke of their feelings of relief to have the problem ‘adopted’ by the doctor. The perception was that the doctor not only understood the problem but was willing to get involved in the family’s situation.” (Macleod et al, 2002, pg 154). It is important to understand how both patients and genetic counselors perceive the success of their meetings in different delivery methods in order to determine their effectiveness.

Rural use of Telegenetics

The use of telegenetics is not something completely new. In fact it was used before the pandemic as a means to connect rural residents with genetic counseling. Nearly 20% of the American population resides in rural areas, while the majority of genetic counselors are located in urban

communities (Rhoads and Rakes, 2020). This geographic distance means that many rural residents lack access to the genetic counselors and have to travel long distances in order to receive care (Rhoads and Rakes, 2020). The use of telegenetics has allowed for rural residents to get access to genetic counselors without having to make a long commute. Several studies have been conducted regarding the overall success of telegenetics and it is said to be comparable with in person meetings mainly due to convenience and having a support system nearby (Rhoads and Rakes, 2020). It has been reported that the satisfaction rate for virtual genetic counseling meetings was high, and in some cases the satisfaction was higher for virtual meetings than in person meetings (Vrečar et al, 2017). One study also reported that, although there was high patient satisfaction, genetic counselors had more reservations about telegenetics due to the dependency on technology, picking up on nonverbal cues, and assessing patients' emotions and level of understanding (Vrečar et al, 2017). These studies demonstrate the importance of having telegenetics in order to provide rural residents with access to high quality care.

An additional study conducted in Australia studied the satisfaction of parents in the use of telehealth for pediatric genetic counseling visits (Hopper et al, 2011). From the data there was a very high overall satisfaction using videoconferencing as a means to meet with a genetic counselor. Although some of the parents would have preferred an in-person meeting, the remote meeting would allow them to be seen sooner. In this study a high definition video camera was used to transmit, record, and take still images of the meeting (Hopper et al, 2011). The camera allowed for the meeting to be live streamed with the genetic team, as well as be recorded, so the film could be referred to and reviewed at a later time. Three weeks after the initial videoconference an in-person meeting was arranged with the participants to meet with the same genetic team (Hopper et al, 2011). The use of the high-definition technology allowed for accurate measurements to be taken, and while there was some variability there was not a large enough difference to alter a diagnosis (Hopper et al, 2011). Overall this study demonstrated the ability to conduct genetic counseling meetings virtually while still obtaining accurate measurements.

Delivery Methods of Genetic Counseling

As time has progressed there have been more adaptations in format of how genetic counseling meetings are delivered. The article *Genetic Survey Delivery Models* provides an in depth look at the different methods of accomplishing genetic counseling. Overall, their study summarizes the benefits and barriers of the different delivery methods of genetic counseling. Benefits of in-person counseling include better adherence to risk management recommendations, the freedom to ask questions, and greater ability to build a relationship (Pierle and Mahon, 2019). The barriers of in-person counseling include it being time-intensive, dependent on geographical location for appointments, longer wait times for appointments, and dependent on the availability of providers (Pierle and Mahon, 2019). Benefits of telegenetics include the convenience for the patient and family, no travel time, better access for remote locations, and reduced wait times for appointment (Pierle and Mahon, 2019). The barriers to telegenetics include the technology requirements, having trouble gauging patient understanding and emotions, not having the patient's full attention, and variable billing (Pierle and Mahon, 2019). This study identifies different barriers and benefits to genetic counseling delivery methods in a time without COVID-19.

Satisfaction of Internet-Based Genetic Counseling

Besides the use of telehealth to connect rural residents with genetic counselors, one group of researchers studied the practicability of conducting virtual cancer genetic counseling services. In order to carry out the Internet based counseling participants were sent webcams and software to download onto their computer (Meropol et al, 2010). Participants were also provided with written instructions at an eighth grade reading level with computer screenshots. Prior to the meetings a connectivity test was done to make sure that the software was properly installed (Meropol et al, 2010). When looking at the results of this study, participants were satisfied with the technical aspects of having the genetic counseling meeting virtually. An unexpected finding in this study was that participants had such a high satisfaction of the technology used, even though most participants encountered some type of technological problem either before or during the meeting (Meropol et al, 2010). Regarding the evaluation of the meeting itself, participants were satisfied with the information they received, level of communication, and the ability to build a relationship with the genetic counselor (Meropol et al, 2010). In this study all of the participants agreed that they would recommend this type of meeting to others, but 29% of the participants also said that they would have preferred to have the meeting in person (Meropol et al, 2010). Overall, this study supports that telegenetics is feasible and can be used as an alternative to in-person genetic counseling meetings, although there are a few technical features that should be improved upon.

COVID-19 prenatal GC

Since the outbreak of COVID-19 there has been one study published regarding the use of telehealth for high risk pregnancies during COVID-19, based on research conducted in New York City, one of the global epicenters. There are many different healthcare professionals a woman may interact with while pregnant, and one of these people is a genetic counselor. In this study the roles of different medical professionals who may be involved in a high-risk pregnancy are altered to fit into a time of telehealth. Due to the pandemic all genetic counseling meetings were conducted using telehealth (Aziz et al, 2020). At home saliva kits have been sent directly to a patient's home to collect samples that can then be directly shipped to laboratories to have genetic testing done (Aziz et al, 2020). To limit in-person contact any blood tests or noninvasive prenatal screening have been coordinated to coincide with scheduled ultrasounds (Aziz et al, 2020). Overall this research showed how genetic counseling was able to adapt to a remote setting.

Although previous studies have been conducted about genetic counseling and telegenetics, my project addresses the topic under the conditions of living in a pandemic. This study examines genetic counselors' perceptions of telegenetics when in person meetings were not feasible. The purpose of this research is to understand how genetic counselors' feel about the use of telehealth.

Methods:

This study examines genetic counselors' perceptions about the positive and negative factors associated with telegenetics during COVID-19. This project took place in the spring semester of 2021 spanning from January to May. A preliminary literature search was conducted to gain background knowledge and set the basis for interview questions. Two in depth, semi-structured interviews were set up with genetic counselors to ask questions from a survey in February of 2021. Participants were recruited through email and networking with a member of the American

College of Medical Genetics. This member of the ACMG passed along a research flyer to additional genetic counseling contacts. These interviews were recorded and then transcribed, where qualitative data was collected.

The questions asked were broad in nature, where meaningful and detailed explanations were provided by the genetic counselors. Before the interviews were recorded each participant was asked if it is alright, that they do not have to answer any question if they are not comfortable doing so, and that they are free to stop the interview at any time. In this report each of the participant's privacy will be protected using pseudonyms. The sample consists of two women, Jennifer and Lindsey. Jennifer is a white, middle-aged woman who specializes in pediatric genetics, with some emphasis on pediatric cancer predisposition. Lindsey is a non-white middle aged woman who works in both pediatric and adult general genetics as well as cranial facial inpatient services. Both genetic counselors have less than five years experience in genetic counseling and work in the Los Angeles, California area, although their service radius extends much farther. For each interview 45 minutes were set aside, although neither meeting lasted that long. A total of six questions were asked to each genetic counselor. This set of questions were

1. How long have you been a genetic counselor? Do you have a specialty within Genetic Counseling?
2. Pre-COVID what was your primary delivery method?
3. During COVID how has your delivery method changed?
4. How do you feel about telegenetics in comparison to face-to-face meetings?
5. Do you think that the patient-provider relationship has changed as a result of doing more virtual meetings? How? Do you feel that the relationship has weakened or strengthened?
6. What is your overall satisfaction of conducting genetic counseling meetings virtually?

Using the audio from the recorded meetings each interview was transcribed verbatim. Each transcript was read and analyzed to determine the common themes. The qualitative data was analyzed using interpretive descriptive thematic analysis techniques (Braun & Clarke, 2006). The first investigator used line by line coding when analyzing the transcripts, where while reading each transcript notes were written off to the side. Complementary to the first investigator, a second investigator analyzed the transcripts. Both investigators met regularly where they came to a general consensus on the codes and themes. The common themes that the transcripts were organized into were benefits and drawbacks.

Results:

The data suggests that both participants view telegenetics favorably. For the purpose of this research each participant identified both positive and negative aspects of telegenetics. Below I describe how I identified the two main benefits of telegenetics as increased patient accessibility and productivity and the two main drawbacks as a lack of control over the environment during patient encounters and challenges to the patient-provider relationship.

Benefits

The results suggested that the greatest benefit of telegenetics was the increased accessibility for patients. Both participants mentioned that telehealth helped by eliminating travel times. By having meetings conducted virtually patients no longer had to commute and drive long distances to meet with genetic counselors. Not having to travel also helped to decrease stress levels and money spent. When commenting on travel times, Jennifer said that

“[in] our service radius we get patients all the time from the, the Central Valley’s all the way like in Fresno...Where Santa Barbara which is like hours north, people will drive all day. So we actually have been able to reach a lot more patients.”

A second benefit to telegenetics that both participants agreed on was the increased productivity. By getting rid of the commute, as well as no longer having a limited time frame to meet with patients face-to-face genetic counselors have been able to use the once lost time to their benefit. Each participant mentioned that they were able to both see more patients and spend more time with them. Jennifer said that “I can spend more time with patients and not have to worry about getting them out of a clinic room quickly so that the next patient can come into a physical room.” She also recalled that when doing in-person meetings that

“you only have a certain amount [of rooms] for a certain amount of time and you have X number of patients. You really have to keep things moving otherwise everything gets backed up.”

Having genetic counseling appointments have allowed for there to be a smaller waitlist to schedule an appointment and more time spent one-on-one with a genetic counselor.

Drawbacks

Along with the advantages of telegenetics, study participants also noted a few disadvantages. Based on the participants’ responses the primary barrier of conducting genetic counseling meeting remotely is not being able to control the environment. There are several different factors that contribute to this theme including internet connection, disruptions, and the setting the meeting is conducted in. In an in-person meeting the counselor and patient typically meet in a doctor’s office in a quiet room. However, with the ability to have meetings online, patients can choose to take the meeting anywhere. Lindsey was able to see how having meetings at home can be both good and bad. She mentioned that

“some people are more comfortable in their home actually, and other people, like we have kids running around and their wifi isn’t working and they’re not so technology savvy. Or we have had people take a call from like the parking lot of McDonalds.”

The other participant Jennifer spoke of a time where she disclosed diagnostic results of cancer predisposition and the patient was so upset she ran from the room leaving the genetic counselor stuck there. These virtual meetings allow for more distractions and interruptions, as well as connection issues and patients leaving meetings prematurely. One instance of a patient leaving early occurred when a patient hung up a call after learning they were negative for Huntington’s disease. Jennifer spoke about her frustration that “there was like no way to kind of follow through on, like, how are you feeling about this like where are you going, let’s talk about this.” With telegenetics you lose the private environment that a clinic space offers. The lack of control

over the environment also includes technology issues. When asked what she would like to change in order to improve the quality of telegenetics, Lindsey responded

“I wish more patients would know to test their wifi. We should, they would be aware, like could you be in a quieter space, be somewhere where you’re really comfortable and not distracted. And I wish that there was more ways to give like equitable access, you know, somebody who’s Spanish speaking or not really good with tech or doesn’t have a Smartphone, like how can we make it seamless for them to have equal access to this type or care.”

Jennifer also mentioned that

“connection issues are the worst because like someone [is] sort of frozen and then they, and then they move again and, you know, without a fluid. Yeah without a fluid connection it’s hard to really see that transition that someone’s going through.”

Overall the environment and good internet connection play a critical role in the success of virtual counseling.

A second theme that I identified under disadvantages to telegenetics is a compromised ability to connect with patients. This connection with patients includes both physical exams and emotional connections. When it comes to genetic conditions there are certain features and characteristics that may be an indicator of a certain ailments. Jennifer said

“sometimes when you see features of a syndrome on telemed, it’s not quite as precise. You can’t appreciate certain skin markings, so if someone really needs a detailed physical exam by an MD, they’re going to that in person clinic.”

Although some of these features can be shown over the video conference or sent as individual photo attachments it can be difficult to truly appreciate the features through a screen, and that is why in person physical exams are quite valuable. However, when asked about conducting physical exams virtually Lindsey said that

“I think it can be a little harder to do a physical exam, but sometimes we ask them to send pictures to us or you know come closer to the camera and sure, we don’t get the full physical exam sometimes, but, um, I think it doesn’t change what testing we do and if results come back that we need to observe their hands more than we’ll, we’ll focus on that.”

Another aspect of connecting with patients occurs at the emotional level. In a typical in-person setting genetic counselors are able to pick up on nonverbal cues, subtle motions or changes, to help identify how a patient is feeling. The two participants had slightly conflicting responses to picking up on non-verbal cues virtually. Lindsey said that “in terms of picking things up no, I think I’m doing just as well on that, the same reaction, like if the video is placed right I can still pick things up and I’m not afraid to explore it.” On the other hand Jennifer said that it is harder for her to pick up on things and that she has to say things up front, pick up on subtle movements, and directly ask how the patient is feeling. Further research will need to be conducted in order to truly understand the impact that telegenetics has on the patient-provider relationship. Lindsey said that “I haven’t seen any bad changes. I’ve really seen them [the patients] still connect to us,

still trust us, still need us; no real change.” However, Jennifer feels that it has been more complicated to get patients engaged and assessed.

Overall, both respondents predict that telegenetics is here to stay, even once COVID-19 is under control. COVID-19 has caused our healthcare system to evolve, including the practice of genetic counseling. Although the hope is to return to in-person meetings once COVID-19 restrictions are lifted, it is unlikely that the use of telegenetics will completely go away.

Discussion:

The results of my thesis seem to be quite comparable with previous studies. From my literature review telegenetics was first implemented in order to allow for genetic counselors to meet with rural residents. One such study stated

“Many rural residents do not receive genetic counseling or testing when needed because of health care access barriers, such as lack of providers in rural areas and the requirement for rural residents to travel to larger cities for these services. Telehealth technology can reduce these barriers by allowing rural residents to receive genetic counseling through a two-way interactive audio/video secure connection in a local clinic or in their homes” (Rhoads and Rakes, 2020, pg 190)

It seems appropriate that one of the major benefits of telegenetics in the time of COVID-19 is the increased accessibility. The convenience of not having to travel to meet with a genetic counselor in person allows for more patients to be seen in a wider geographic range without having to make a long commute. Looking towards the future once COVID-19 is under control it would be beneficial to still have the option of telegenetics in order to be more accessible to a wider array of patients across a larger geographic area. The ability to conduct telegenetics also decreases cost and expenses needed for visits, so telegenetic also may be more accessible to those in lower socio-economic groups. Further research will need to be studied though in order to understand if there are disparities regarding lower income families and genetic counseling.

Another benefit of telegenetics was the increased productivity. As a result of meetings moving virtual the participants reported they were able to see more patients and spend more time with them. While in previous research there was no direct mention of telegenetics being +more efficient, the respondents reported positive feedback in reaching more people and having impactful conversations. In the future it would be helpful to find out why the virtual meetings are so efficient and how it may help to make in-person meetings more productive.

One barrier addressed in this project was the inability to control the environment. In previous research there was not any mention of issues where the meeting took place. However what other studies did mention quite frequently was that possible barriers of telegenetics included technical issues and equitable access (Pierle and Mahon, 2019) (Meropol et al, 2010). In other research studies there were a few groups that sent webcams and downloadable software to participants in order to in order to maximize hosting the genetic counseling meeting virtually meetings (Meropol et al, 2010) (Hopper et al, 2011). Having this technology available to patients increased the quality of video, and in one study allowed for accurate virtual examinations to be done. In the future it may be worth exploring if webcams and software may be able to improve the quality of virtual meetings and eliminate some technological issues.

A final barrier to telegenetics is the connection to the patient. The participants in this study had mixed feelings about this theme so further research should be conducted in order to discover if there is a negative correlation between telegenetics and the ability to connect with patients. A few studies mentioned how genetic counselors were apprehensive about counseling virtually due to difficulty in understanding patient emotions (Pierle and Mahon, 2019)(Vrečar et al, 2017). In a future study it would be helpful to _study how both genetic counselors and patients viewed a meeting and compare the results. This would allow us to understand if both groups felt connected and comfortable, or if adjustments should be made. If results show difficulty in connecting to patients it may be beneficial to dedicate more time and training into how to successfully connect with patients during counseling sessions. Besides an emotional connection it is important to address the ability to conduct exams using telehealth. Certain features, skin markings, and physical appearances may be indicators to some genetic conditions. From this study there were mixed feelings on the ability to do physical exams using telegenetics. Lindsey felt that she was able to understand features that were sent by photograph, while Jennifer found it more difficult to appreciate and discern features through a screen. Further research should be conducted in order to determine the success of conducting physical exams virtually and if it impacts diagnosis. One study had positive results conducting physical exams which aided in an accurate diagnosis due to the use of a high definition camera and other technology (Hooper et al, 2011). One possible way genetic counselors may be able to perform accurate physical exams is by investing in a high definition camera and other equipment to improve the quality of the video.

Conclusion:

Potential limitations of this study include having all the participants being female and living in the Los Angeles, California area. Additionally the genetic counselors interviewed are primarily involved in pediatric cases, although some adult patients are seen by them as well. In the future it would be helpful to conduct a larger study that is more representative of gender, location, and specialty. Additionally this project is not applicable to counselors outside of genetic counselors. In the future it would be helpful to get a larger and nationally representative sample of participants.

Overall, genetic counselors in this study view telehealth as a success due to its increased accessibility and less time constraints. Implementing telegenetics can be innovative approach to maintaining genetic counseling practices during the COVID-19 pandemic and under normal circumstances this transition would have taken years accomplish. Even though the delivery of the genetic counseling meetings has changed, patients are still being seen and heard. It is extremely likely that telegenetics will still be used frequently even when the pandemic is over so the barriers of controlling the setting, technology requirements, and the ability to complete physical exams need to be addressed in order to get the best results out of doing genetic counseling virtually.

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