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Enhancing Patient-Centered Care Through the Use of Digital Tools in the Delivery of Genomic Medicine

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Abstract

To fulfil the growing demand for genomic sequencing (GS) across medicine, alternative forms of genetic counselling are required. This demand is growing as a result of the severe lack of experts who can conduct genomic counselling, a procedure that includes counselling for the many hazards that GS can uncover. There are roughly 5,500 genetic counsellors in Canada and the United States to service a population of almost 400 million people, or 1.5 professionals per 100,000 people. It is anticipated that this workforce will need to double in order to fulfil the rising demand for genetic counselling.

Keywords: Genomic medicine • Patient-Centered care • Genomic

Introduction

To fulfil the growing demand for genomic sequencing, alternative forms of genetic counselling are required. Digital tools have been suggested as a way to supplement conventional counselling and lighten the load on specialists, but it is unclear how they will be used to give genetic counselling. In order to give genomic counselling, this study investigated the function of the Genomics ADvISER, a digital decision aid. 52 pre-test genetic counselling sessions for a randomised controlled experiment examining the efficiency of the Genomics ADvISER were subjected to secondary analysis. Participants in the trial were randomly assigned to either get normal counselling or use the instrument before speaking with a counsellor.

The use of digital tools to support the delivery of counselling across the genomic testing pathway from pretest to post-test counselling, including family history-taking, consent for testing, and education, is growing in response to the manpower deficit. A wide variety of digital technologies, such as software, digital portals, and chatbots, has been developed. It has been demonstrated that the use of these tools in counselling for cancer susceptibility, prenatal abnormalities, and carrier status improves knowledge and patient satisfaction, lessens decisional conflict, and encourages patients to make deliberate decisions. Digital technologies, according to clinicians, are useful, enhance the communication of risk information, help structure sessions, and can facilitate patient-centered care, which is a fundamental principle of genetic counselling practise [1].

To fulfil the growing demand for genomic sequencing (GS) across medicine, alternative forms of genetic counselling are required. This need is growing as a result of a severe lack of qualified individuals who can conduct genomic counselling, a process that includes counselling for the many hazards that GS can uncover and can take hours per patient. There are roughly 5,500 genetic counsellors in Canada and the US to serve a population of almost 400 million people, or 1.5 professionals per 100,000 people. It is predicted that this workforce will need to be doubled to meet the rising demand for genomic testing. The unbalanced workforce allocation between metropolitan academic hubs and rural areas, 8 and the growing number of therapists switching to the use of digital tools to support the delivery of counselling across the genomic testing pathway from pretest to post-test counselling, including family history-taking, consent for testing, and education, is growing in response to the manpower deficit. A wide variety of digital technologies, such as software, digital portals, and chatbots, have been developed. It has been demonstrated that using these tools in counselling for cancer susceptibility, prenatal abnormalities, and carrier status improves knowledge and satisfaction, reduces decisional conflict, and helps patients make deliberate decisions. According to clinicians, digital tools are beneficial, increase risk communication, help structure sessions, and can facilitate patient-centered care.

Description

Despite the growing demand for their use, the majority of digital tools and the associated studies have only been used in the context of counselling for genetic testing like prenatal screening and cancer panels. There aren't many tools available for genetic counselling. Additionally, the majority of studies on digital tools have concentrated on their acceptability and efficacy, while the effects of digital tools on facilitating patient-centered treatment and genomic counselling are still unknown. The purpose of this study was to investigate the Genomics ADvISER, a new digital tool that can be used to support genomic counselling with a genetic counsellor. The use of digital tools in genetic counselling sessions can be improved with a better understanding of their function. This qualitative study, which was included into a randomised controlled trial (RCT) examining the efficacy of the Genomics ADvISER digital decision aid, used secondary analysis of transcripts of pre-test genetic counselling sessions. Our analysis is based on the transcripts of the pretest counselling sessions that were held with all study participants. We selected qualitative approaches because they offer in-depth information on participants' experiences, which is crucial for understanding how the digital tool interacts with the genetic counsellor to help patients make decisions [2].

A thorough description of the trial has already been provided. In the RCT, individuals were randomised to receive traditional genetic counselling over the phone (control arm, CTRL), or to use the digital decision aid and then speak with a genetic counsellor over the phone (intervention arm, INTV), to enhance decision-making related to secondary findings (SF). Genomic sequencing wasn't done, and the choice of which SFs to receive was made hypothetically because the trial's goal was to evaluate the decision aid's usefulness rather than to respond to results. The digital tool used by intervention participants

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walks users through a 10-minute whiteboard video explaining the fundamentals of genomic sequencing and the five SF categories that are available, then prompts them to answer a few quick interactive questions about their values, knowledge, and decision-making requirements. SF categories were present [3].

Pre-test genetic counselling for genomic sequencing, including an explanation of the SF categories, was modelled in the control sessions. Following completion of the decision aid, intervention participants had a chance to ask questions and discuss with a genetic counsellor. The subject was asked to express their hypothetical choice to receive any combination of the five SF types at the end of each session in both arms. Each session's genetic counsellor followed a script to maintain uniformity in the material delivered, but they also drew on their expertise and experience in the field to treat participants as they would in a clinical setting. Most of the sessions-about half-were recorded [4].

Audio-recorded telephone conversations between participants and the genetic counsellor were analysed. We analysed all of the audio files that were accessible, but several of them had low audio quality and were difficult to transcribe. Participants in some instances refused to have their sessions recorded; therefore their data was not available for analysis. A single licenced genetic counsellor facilitated all 52 of the readily available and high-quality audio recordings (SS). The majority of the sessions from the control arm's 11 sessions had minimal verbal participation from the participant, so only those sections were transcribed verbatim. The transcripts were analysed using thematic analysis 21 with continual comparison20, with an emphasis on the vocal content of the participants (comments and questions) [5].

Conclusion

S.A.R. initially developed an analysis guide (Supplementary file 1) based on the Ottawa Decision Support Framework's definition of decisional needs. 25 Following this, S.S. and S.A.R. listened to the audio files and read the transcripts, making notes that briefly summarised each session and recorded the themes discussed, including those covered by the analysis guide as well as any emergent subjects brought up by participants. These notes were used to create a codebook. Afterward, transcripts were examined, codes were applied, and constant comparison was used to reflect on earlier analysis and iteratively alter codes as needed. All transcripts were coded by S.A.R., and S.S. coded a portion of them as the second coder for analytic validation. Regular meetings were held between S.A.R. and S.S. to discuss the procedure and settle any coding disagreements.

Acknowledgement

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Conflict of Interest

None.

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