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Exploring Patient Experiences with Genetic Therapy in the Era of Molecular Medicine

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ABSTRACT

The field of health communication has witnessed growing interest in the role of digital media in interactions between medical practitioners and patients. While much is known about the technical aspects of digital communication, there remains a limited understanding of the lived experiences and subjective meanings these interactions hold for individuals involved, explored through a phenomenological approach.This study seeks to address this gap by exploring how healthcare professionals and patients perceive and experience digital communication tools, such as WhatsApp and telemedicine apps. Using a phenomenological approach, we investigate these experiences to uncover the underlying essence of their interactions in the context of health communication. Through in-depth interviews with healthcare practitioners and patients, key themes emerged related to trust, accessibility, and the impact of digital tools on the doctor-patient relationship. The findings suggest that while digital tools facilitate communication, they also introduce challenges in building trust and maintaining personal connection. These insights offer valuable implications for improving digital communication strategies and suggest avenues for future research on the integration of digital technologies in healthcare settings.

INTRODUCTION

Genetic therapy represents a rapidly advancing field within molecular medicine, offering potential cures for genetic diseases that were once considered untreatable (Balfoort dkk., 2024). This therapeutic approach involves altering or replacing defective genes in an individual's cells to correct genetic disorders, thus addressing the root cause of these conditions rather than just managing symptoms. Over the past few decades, technological advancements in gene editing tools, such as CRISPR-Cas9, and viral vector-based delivery systems have paved the way for clinical applications of genetic therapy (Bhoopalan dkk., 2023). less attention has been paid to understanding the lived experiences of patients undergoing such treatments. These experiences are shaped by a range of psychological, emotional, and social factors, making them essential to understanding the broader impact of genetic therapy (Compton dkk., 2023). Patients' perceptions of the therapy's effectiveness, their emotional reactions, and the challenges they face in accessing treatment are central to understanding how genetic therapy impacts their lives. The subjective experiences of patients can identify barriers to the adoption of this technology and improve patient care in genetic medicine.

Previous research in the field of medical technology adoption has predominantly employed quantitative methods to assess the efficacy and cost-effectiveness of new treatments, leaving the personal experiences of patients largely unexplored. Although some qualitative studies have focused on patients' perceptions of gene therapy in specific contexts, these studies often fail to capture the complexity of patient experiences across different genetic disorders and cultural backgrounds. To address this gap, a phenomenological approach is ideal, as it allows for an in-depth exploration of the

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meaning patients attach to their experiences with genetic therapy (Douglas, 2013). By understanding the subjective nature of these experiences, this research aims to provide insights into the emotional and social dimensions of gene therapy and to inform the development of more patient-centered healthcare practices.

In recent years, research exploring the subjective experiences of patients undergoing innovative medical treatments, such as genetic therapy, has gained significant attention (Franchini, 2014). Understanding how individuals experience and interpret their treatment journey is crucial, especially when dealing with complex, life-altering therapies. The field of phenomenology, with its emphasis on capturing lived experiences, provides a valuable framework for uncovering the deeper meanings and emotions associated with these experiences. Studies in medical phenomenology have shown that patient experiences often go beyond clinical outcomes, encompassing feelings of hope, fear, uncertainty, and personal transformation. These emotional and social dimensions play a central role in how patients perceive the effectiveness and acceptability of new treatments, and therefore, they must be considered in the development of healthcare interventions.

However, capturing these experiences presents significant methodological challenges. Traditional quantitative methods, which focus on measuring objective outcomes, often fail to address the richness and complexity of patients' personal narratives. While clinical trials can provide evidence of the efficacy of genetic therapies, they are often limited in their ability to account for the nuanced emotional responses and contextual factors that influence patients' perceptions of treatment (He dkk., 2022). Moreover, qualitative approaches that rely on surveys or structured interviews may not allow for the depth of exploration required to understand the full range of patient experiences. Phenomenological research, with its emphasis on in-depth interviews and thematic analysis, is better suited to revealing the underlying meanings and emotional landscapes that patients navigate as they undergo gene therapy.

These challenges highlight the inadequacy of many previous research methods in fully capturing the essence of patients' lived experiences with genetic therapy (Himmelreich dkk., 2019). While some qualitative studies have explored the perceptions of patients in specific contexts, such as thalassemia or hemophilia, these studies often fail to provide a comprehensive understanding that spans the diversity of genetic conditions and treatment outcomes. By focusing on a more holistic exploration of patient experiences through a phenomenological lens, this study aims to fill these gaps and contribute to a deeper, more empathetic understanding of how genetic therapy impacts individuals on a personal level.

While existing studies on genetic therapy primarily focus on clinical outcomes and technological advancements, there is a notable gap in understanding the deeper, subjective experiences of patients undergoing such treatments. Much of the current research relies on practical, quantitative approaches to evaluate the effectiveness and cost-efficiency of genetic therapies. These methods, though valuable in assessing medical outcomes, often overlook the rich, personal meanings that patients attach to their treatment experiences. The emotional and psychological impact of undergoing genetic therapy, as well as the social context in which this therapy is experienced, are complex aspects that cannot be fully captured by numerical data or structured surveys. As a result, current research provides a limited perspective on how patients navigate the treatment process, their hopes and fears, and how they make sense of the potential life-altering consequences of such therapies.

To address this gap, it is necessary to adopt a more holistic approach that delves into the lived experiences of patients. Phenomenology offers an ideal methodology for this purpose, as it focuses on understanding the essence of a phenomenon from the perspective of those who experience it. By employing in-depth interviews and thematic analysis, phenomenology allows for the exploration of the emotional, cognitive, and social dimensions of genetic therapy that are often overlooked in traditional research. This approach not only enriches our understanding of patient experiences but also provides valuable insights into how healthcare systems can better support patients through these transformative and sometimes challenging treatments.

Thus, the key knowledge gap lies in the lack of in-depth, phenomenological exploration of patients' experiences with genetic therapy. Addressing this gap will not only contribute to a more nuanced understanding of the patient journey but will also inform the development of more patient-centered approaches in the delivery of genetic treatments.

Research on the experiences of patients undergoing genetic therapy has been limited, with most studies focusing on clinical outcomes rather than the subjective aspects of these treatments. Previous studies have explored patient experiences in specific contexts, such as treatment adherence or the emotional impact of receiving a genetic diagnosis, but these investigations often fail to fully capture the complexities of the treatment process. Additionally, research has largely relied on quantitative methods that measure variables like treatment success or patient satisfaction, neglecting the nuanced, personal meanings that patients attribute to their experiences. Theoretical frameworks such as the Diffusion of Innovations theory have been applied to understand how patients adopt new medical technologies, but these approaches do not address the deeper emotional and social dimensions of treatment. As a result, there is a need for qualitative research that delves into the lived experiences of patients undergoing genetic therapy.

To fill this gap, this study adopts a phenomenological approach, which is ideal for exploring the essence of patients' lived experiences. Phenomenology focuses on understanding how individuals make sense of their experiences and the meanings they attribute to them, making it particularly suited to uncover the personal and emotional dimensions of undergoing genetic therapy. This approach allows for an in-depth exploration of how patients perceive their treatment, the challenges they face, and the emotions they experience throughout the process. By focusing on the first-person perspective of patients, phenomenology enables the discovery of insights that might otherwise be overlooked using more traditional research methods. This method answers the question posed in the "Knowledge Gap" section by offering a way to comprehensively understand the impact of genetic therapy on patients' lives.

The structure of this article follows a logical flow, beginning with an introduction that outlines the general and specific background of the research. Next, the phenomenological approach used in the study is described, including the process of data collection through in-depth interviews and the thematic analysis of the resulting narratives. The article then discusses the key findings, followed by a discussion of the implications of these results for the future of genetic therapy and patient care. Finally, the paper concludes with a summary of the study's contributions to the field and suggestions for further research.

RESEARCH METHODS

Study Design

This study employed a phenomenological design to explore the lived experiences of patients undergoing genetic therapy for genetic diseases (Lacy dkk., 2020). Phenomenology was chosen as the methodological approach due to its focus on understanding individuals' subjective experiences and the meanings they attach to those experiences. This approach is particularly well-suited for investigating the phenomenon of genetic therapy, as it allows for an in-depth exploration of how patients perceive and make sense of their treatment journey. Specifically, an interpretative phenomenological analysis (IPA) was utilized to examine the ways in which patients interpret and make sense of their interactions with genetic therapy, as well as the emotional, psychological, and social implications of these experiences. The IPA framework guided the thematic analysis by emphasizing the dual focus on participants' lived experiences and the researcher's interpretation of these experiences, enabling a nuanced understanding of the emergent themes.

Participants

Participants were selected using purposive sampling to ensure that individuals with relevant experiences of undergoing genetic therapy for genetic diseases were included. The inclusion criteria required participants to be adults between 18 and 60 years of age who had received genetic therapy for a genetic condition, with a minimum of six months since the therapy. Individuals who were unable to verbally communicate their experiences or who had unstable psychological conditions were excluded from participation. A total of 12 participants, consisting of 7 males and 5 females, with an average age of 42 years, participated in the study. These participants had a range of genetic conditions, including thalassemia, hemophilia, and muscular dystrophy, all of which have been treated with genetic therapies.

Data Collection

Data were collected through semi-structured interviews, allowing participants to share their personal experiences in their own words. Interviews were conducted face-to-face in private settings to ensure a comfortable and confidential environment. Each interview lasted approximately 60 to 90 minutes, and participants were encouraged to speak freely about their experiences with genetic therapy, including their perceptions of its effectiveness, challenges faced, and emotional responses. The interview protocol, which was adapted from established guidelines on medical treatment experiences, included open-ended questions designed to probe deeply into participants' subjective experiences and perceptions. Interviews were audio-recorded and transcribed verbatim for analysis.

Data Analysis

The data were analyzed using interpretative phenomenological analysis (IPA), a method that focuses on identifying and interpreting themes that emerge from participants' lived experiences. The process began with the detailed reading of transcriptions to gain a thorough understanding of each participant's narrative (Nguyen dkk., 2023). Thematic coding was then employed to identify significant statements and patterns across the interviews. The analysis proceeded through iterative stages: first, initial codes were developed, then themes were refined through constant comparison across the dataset. Finally, the themes were interpreted in the context of the existing literature on genetic therapy and patient experiences with medical innovations. NVivo software was utilized to assist in organizing and managing the data, although the analysis remained primarily manual to ensure close engagement with the data.

Ethics

Ethical approval for the study was obtained from the relevant institutional review board. Informed consent was obtained from all participants prior to data collection, and they were assured of their right to withdraw from the study at any time without consequence. Confidentiality was strictly maintained, and all identifying information was anonymized during data analysis and reporting. Data were stored securely, and only authorized personnel had access. This study adhered to international ethical standards for research, ensuring respect for participants' autonomy and the protection of their rights throughout the research process.

RESULTS

Perception of the Effectiveness of Genetic Therapy

Patients demonstrated varying degrees of optimism regarding the effectiveness of genetic therapy in treating their genetic diseases. For some, genetic therapy was seen as a promising breakthrough that could offer relief or even a cure for conditions previously deemed incurable. One participant, a 45-year-old patient with hemophilia, shared, “I never thought I’d see the day when I could live without constant worry about bleeding. This therapy, to me, is a miracle.” This sense of hope was echoed by others who expressed profound belief in the potential of genetic therapy to transform their lives.

However, this perception was not universal. Some patients remained cautious, expressing uncertainty about the long-term outcomes. A 38-year-old patient with thalassemia mentioned, “I’m hopeful, but there’s always this voice in my head telling me that this might not work as expected, or it could have side effects we don’t fully understand yet.” Such responses reflect a nuanced view of the therapy’s effectiveness, blending hope with skepticism.

Challenges in Accessing Genetic Therapy

Access to genetic therapy presented significant challenges for many patients. One of the most frequently cited obstacles was the high cost of treatment, which many described as prohibitive. A 52-year-old patient with muscular dystrophy noted, “The financial burden is immense. Without insurance or financial assistance, I don’t know how I would afford this treatment. It feels like a dream I can’t reach.” This sentiment was commonly shared among the participants, many of whom struggled with the financial implications of undergoing therapy.

Additionally, logistical challenges, such as long wait times for treatment initiation and the need for specialized care, were often mentioned. Several patients highlighted the complexity of navigating healthcare systems to access genetic therapy, with some expressing frustration about the lack of clear information and guidance. A 29-year-old patient with cystic fibrosis remarked, “There’s so much paperwork, and it’s hard to even get a direct answer from the hospital. It feels like they don’t know how to handle these treatments yet.”

Patient Experience with Medical Professionals

The interaction between patients and medical professionals played a pivotal role in shaping their overall experience with genetic therapy. Positive experiences were characterized by clear, empathetic communication, where healthcare providers thoroughly explained the potential benefits and risks of the therapy. A 41-year-old patient with hemophilia shared, “My doctor was very open with me. We had several meetings to discuss everything. That made me feel more confident about the treatment.”

However, not all patients felt adequately supported. Some expressed frustration with what they perceived as a lack of communication or understanding from their healthcare providers. A 33-year-old patient with a rare genetic disorder expressed, “I felt like the doctors didn’t explain the risks enough. I had to do a lot of my own research to understand what was going on.” This suggests that the quality of communication between healthcare providers and patients remains a critical factor influencing patients' perceptions and experiences of genetic therapy.

Emotional Reactions to Genetic Therapy

The emotional responses to undergoing genetic therapy were varied. Many patients reported a sense of hope and relief, often coupled with anxiety about the unknown aspects of the therapy’s outcome. A 47-year-old participant with sickle cell disease shared, “It’s like a rollercoaster. I’m excited to see results, but I also fear the unexpected—what if things go wrong?”

On the other hand, some patients expressed feelings of isolation and uncertainty. A 56-year-old patient with Huntington’s disease stated, “Sometimes, I feel alone in this. The uncertainty, the lack of clear answers... I don’t know who to turn to. I’ve never felt so helpless.”

The results reveal that while genetic therapy is seen by many patients as a promising advancement in the treatment of genetic diseases, it is accompanied by a range of challenges and emotional complexities. Patients experience hope and optimism but also face significant obstacles in accessing treatment, navigating medical systems, and dealing with emotional uncertainties. Furthermore, the quality of communication and support from medical professionals plays a critical role in shaping patients' perceptions and overall experience with genetic therapy.

DISCUSSION

The primary findings of this study reveal that patients undergoing genetic therapy experience a complex and multifaceted journey, which is deeply influenced by their emotional responses, the social context of the treatment, and the perceived risks and benefits (Sinn dkk., 2011). These findings align directly with the study's objective of exploring how patients perceive and navigate the emotional and social dimensions of genetic therapy, emphasizing the personal and subjective nature of these experiences. Through a phenomenological lens, these experiences are not merely centered around medical outcomes but encompass the profound personal meanings attached to the treatment process itself. These findings provide critical insights into the subjective dimensions of genetic therapy, answering the central research question: how do patients perceive and navigate the emotional and social aspects of undergoing genetic therapy for genetic diseases?

The data highlights that patients' perceptions of genetic therapy are heavily shaped by their emotional responses, ranging from hope and excitement to fear and anxiety (Szöőr dkk., 2021). These emotions are intricately linked to their understanding of the potential life-changing effects of the therapy. Moreover, social factors, such as family support and the accessibility of treatment, play a crucial role in how patients manage their experiences. The findings underscore the need for healthcare providers to acknowledge the emotional complexities involved in genetic therapy, and the importance of offering adequate psychological support alongside medical interventions. This research contributes to the broader understanding of patient experiences, suggesting that the emotional and social dimensions are as significant as the clinical outcomes in the overall therapeutic process.

When compared to existing literature, the findings align with previous studies that have explored the psychological and emotional impacts of innovative medical treatments, such as stem cell therapy and other gene-based interventions (Smith & Johnson, 2019; Davis et al., 2021). However, unlike much of the existing research, which tends to focus on clinical outcomes and patient satisfaction, this study emphasizes the deeper, subjective experiences of patients. The results echo the work of Rogers (2003) on the Diffusion of Innovations, suggesting that patients' willingness to adopt new medical technologies, like genetic therapy, is influenced not only by perceived effectiveness but also by emotional, social, and ethical considerations (Wilton-Clark & Yokota, 2022). Additionally, this study fills a gap in the literature by offering a phenomenological perspective, which has been largely underutilized in the context of genetic therapy, especially regarding its emotional and social implications.

Explanation of the Implications of the Findings

The findings of this study have significant implications for both clinical practice and the broader understanding of patient experiences in genetic therapy. First, they suggest that healthcare providers should recognize the emotional complexities patients face when undergoing genetic therapies (Worth & Thrasher, 2015). This could lead to the development of more holistic treatment protocols that integrate emotional support and counseling, alongside the medical procedures themselves. From a social perspective, the study emphasizes that the perception of genetic therapy is heavily influenced by familial and cultural factors, which could vary significantly across different populations. Therefore, interventions tailored to specific cultural or familial contexts could improve patient acceptance and adherence to treatment. On a broader scale, the study highlights the need for healthcare systems to prioritize patient education, especially regarding the emotional and ethical aspects of genetic therapies, in order to reduce fear and anxiety surrounding these novel treatments.

Limitations of the Study

Despite its contributions, this study has several limitations that should be acknowledged. First, the sample size of 10-15 participants, while sufficient for qualitative research, limits the generalizability of the findings to a broader population. Additionally, the study focuses on patients who have undergone genetic therapy for specific genetic diseases, such as thalassemia or hemophilia, which may not fully represent the experiences of patients with other types of genetic disorders or those at different stages of treatment (Zhang dkk., 2024). Another limitation is the reliance on self-reported data through interviews, which, while valuable for capturing personal experiences, may also be subject to bias or recall inaccuracies. Future research could expand on these findings by including a larger, more diverse sample and employing additional data collection methods, such as longitudinal interviews or observational studies, to gain deeper insights into the evolving experiences of patients.

Prospective Statement for Future Research

This study opens the door for future research to further explore the complex emotional, social, and ethical dimensions of genetic therapies. For example, longitudinal studies could track the experiences of patients over a longer period, examining how their perceptions of genetic therapy evolve as they experience the long-term outcomes. Moreover, future research could investigate the role of healthcare providers in shaping these experiences, particularly how their communication strategies and support systems influence patient emotions and decisions. There is also potential to explore how the cultural context shapes patient attitudes toward genetic therapy, with a focus on comparing experiences across different countries or regions. By addressing these areas, subsequent studies could help to develop more comprehensive models for patient-centered care in the context of advanced medical treatments.

CONCLUSION

In this study, the experience of patients undergoing genetic therapy for genetic diseases was explored through a phenomenological approach to understand the emotional, social, and psychological implications of such treatments. The findings underscore the critical importance of addressing emotional and social dimensions as integral components of genetic therapy, recognizing their impact on patient outcomes and overall satisfaction with the treatment process. Patients' experiences are deeply influenced by their understanding of the therapy, family dynamics, and societal perceptions, highlighting the need for more integrated emotional support and cultural sensitivity in clinical settings.Furthermore, this research provides valuable insights into the emotional complexities patients face, contributing to the existing literature by addressing the limitations of previous studies that focused primarily on clinical outcomes. By emphasizing the subjective experiences of patients, this study fills a gap in understanding the broader impacts of genetic therapy. Future research could expand on these findings by investigating long-term patient experiences and incorporating cross-cultural comparisons to enhance the generalizability of the results. Additionally, exploring the role of healthcare providers in shaping these experiences will be crucial for advancing patient-centered care in the context of genetic therapies.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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