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Psychological Experiences and Emotional Impact of Gene Therapy in Genetic Disorder Patients

Didit Fajar Nugroho [1]*, Ninik Nurhidayah [2]
[1]Universitas Islam Sultan Agung, Indonesia
[2]Poltekkes Kemenkes Surakarta, Indonesia
[1]diditfajar.dr@gmail.com*, [2] niniknurhidayah@gmail.com

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ABSTRACT

Gene therapy has emerged as a revolutionary treatment for genetic disorders, offering the potential for long-term cures by addressing the root cause of disease at the genetic level. While clinical studies have focused on the efficacy of gene therapy, there remains limited understanding of the psychological and emotional impacts experienced by patients undergoing such treatments. This study aims to explore the emotional and psychological experiences of patients receiving gene therapy, specifically focusing on how they perceive and adapt to changes in their health status. Using an interpretative phenomenological approach, this research investigates the lived experiences of 12 patients who underwent gene therapy for genetic disorders. Data was collected through semi-structured interviews and analyzed thematically, revealing that while patients initially experience intense hope and relief (reported by 10 out of 12 participants), they also face significant psychological challenges, including heightened anxiety (observed in 8 participants) and profound shifts in personal identity (experienced by 6 participants). These findings suggest that emotional support and holistic care are crucial for enhancing patient well-being during and after gene therapy. However, the study is limited by its relatively small sample size and reliance on self-reported experiences, which may not capture the full diversity of patient perspectives. The study contributes to a deeper understanding of the emotional complexities associated with gene therapy and calls for further research into the psychological effects of medical interventions.

INTRODUCTION

Gene therapy represents a transformative approach in medical treatment, where genetic material is introduced into a patient's cells to correct or mitigate genetic disorders (Mohta & Halder, 2021). This cutting-edge field holds significant promise for treating diseases once deemed untreatable, such as rare genetic disorders, certain cancers, and inherited conditions (Wang et al., 2022). The ability to target the root cause of diseases at the genetic level has revolutionized therapeutic possibilities. However, the impact of gene therapy extends beyond clinical efficacy; it also encompasses complex emotional, psychological, and social dimensions often overlooked in scientific discourse.

Despite growing recognition of its medical success, there is still a limited understanding of how patients psychologically and emotionally experience gene therapy. This gap is particularly critical because such treatments often entail long-term changes to patients' health, identity, and perceptions of future well-being (Dang et al., 2021; Waterman et al., 2019). Exploring these lived experiences is therefore essential for advancing holistic care. The human experience of gene therapy is shaped not only by medical outcomes but also by broader cultural and social contexts, such as values attached to health, stigma surrounding genetic disorders, and family or community dynamics

(Sun et al., 2020). While prior studies have acknowledged these psychosocial aspects, few have provided an in-depth exploration of patients' lived experiences. This study seeks to address that gap.

Phenomenology offers a valuable lens for examining these subjective perspectives, focusing on how individuals make sense of their journey with gene therapy (Chuang et al., 2023; Fennie et al., 2021; van Lier et al., 2022). By delving into patients' narratives, researchers can uncover not only the emotional and psychological impacts but also the broader meanings attached to health, treatment, and identity. Such insights are crucial for designing support systems that complement clinical care.

Research into these experiences has become increasingly relevant in both medical and psychological domains (Aust et al., 2019; Chamsi et al., 2023; Scalia et al., 2024). Understanding patients' perceptions informs clinical outcomes, sheds light on coping mechanisms, and supports long-term adjustment(Fung et al., 2023).

Qualitative approaches, especially phenomenology, have gained traction for their ability to capture the deeper meanings of these personal journeys, providing a more holistic view than clinical results alone.

However, methodological challenges persist. Traditional quantitative approaches, while valuable for measuring efficacy, are insufficient to capture the nuanced emotional and psychological dimensions of patient experience. This underscores the need for qualitative, phenomenological studies that directly address patients' voices and perspectives.

However, exploring such deeply personal and complex phenomena poses significant methodological challenges. Traditional quantitative approaches, with their reliance on statistical data and objective measures, are limited in their ability to capture the nuances of human experience. These methods often fail to address the emotional and psychological dimensions of patient experiences, focusing predominantly on measurable outcomes rather than the subjective meanings that individuals attach to their illness and treatment. Consequently, while many studies have contributed valuable clinical data on the efficacy of gene therapy, they fall short in providing a comprehensive understanding of how individuals experience the treatment, particularly from a psychological and emotional standpoint.

The limitations of these traditional methods highlight the need for an approach that can engage with the lived experiences of patients in a more meaningful and nuanced way (Clifton et al., 2022). Phenomenology, with its emphasis on the subjective interpretation of lived experience, offers a more effective approach to understanding the essence of phenomena such as gene therapy (Pharasi & Patra, 2020; wube et al., 2025). By focusing on the personal meaning that individuals attach to their experiences, phenomenology provides deeper insights into the psychological impacts and social dynamics of undergoing such transformative treatments. This approach allows for a more complete understanding of the human experience of gene therapy, which is essential for improving patient care and informing future therapeutic practices.

In the context of gene therapy, most research has traditionally relied on quantitative methodologies that focus on measurable clinical outcomes, such as the success rates of the therapy or its physical impacts on the patients (Aratti & Zampini, 2024; Ibrahim et al., 2024; Mohammadi et al., 2023). These practical approaches, while valuable in assessing the efficacy of gene therapy, fail to capture the profound emotional and psychological experiences of patients undergoing this transformative treatment. Such methods often overlook the subjective meanings that individuals attribute to their treatment journey, reducing the complexity of their experiences to numbers and statistics (Baker et al., 2021). This results in a partial understanding of the phenomenon, neglecting the depth of personal significance that accompanies medical interventions like gene therapy.

The existing approaches are limited in their ability to explore the essence of patient experiences, particularly when it comes to understanding the emotional and psychological dimensions that play a critical role in the healing process. Patients' perceptions of their health status, their coping mechanisms, and their social interactions throughout and after the treatment are central to their overall well-being. These nuanced experiences are difficult to capture through conventional quantitative methods. Therefore, while the clinical effectiveness of gene therapy has been extensively studied, the personal meanings, emotional responses, and psychological effects remain underexplored.

To address this gap, it is essential to adopt a phenomenological approach that can delve deeper into the lived experiences of individuals (Zhan et al., 2021). Phenomenology, with its focus on subjective experience and the meanings that people attach to their lived reality, offers a more holistic and insightful lens for examining the impact of gene therapy. This approach allows for a richer exploration of how patients make sense of their condition, their treatment, and the subsequent changes in their lives. By adopting this methodology, it is possible to gain a comprehensive understanding of the emotional and psychological landscape surrounding gene therapy, thereby informing more empathetic and effective patient care strategies.

Previous research has explored the clinical outcomes of gene therapy, focusing primarily on physical and medical improvements. However, few studies have delved into the psychological and emotional experiences of patients undergoing this treatment. Several studies have examined patient experiences with other forms of medical treatments or chronic illness, using phenomenological approaches to explore the deeper meanings behind their experiences. These studies have highlighted how health interventions, particularly those involving major changes to one's physical state, can evoke complex emotional responses that require a more nuanced understanding. For instance, phenomenological research on cancer treatments has uncovered themes of fear, hope, and identity transformation, all of which are highly relevant to the context of gene therapy.

Given the existing knowledge gap, this study employs a phenomenological approach to explore the lived experiences of patients undergoing gene therapy for genetic disorders (Kim et al., 2020). Phenomenology is particularly well-suited to this inquiry because it focuses on uncovering the subjective meanings individuals attach to their experiences, providing a rich and in-depth understanding of their emotional and psychological states. By using interpretative phenomenological analysis (IPA), the study aims to go beyond clinical measures and explore how patients emotionally process their treatment, perceive changes in their health, and navigate the challenges and transformations brought about by gene therapy.

The structure of this article is organized to provide a clear and comprehensive exploration of the research. Following the introduction, the paper explains the context of the phenomenon under investigation, outlining the importance of examining the psychological impacts of gene therapy. The methodological approach, including the use of phenomenology and IPA, is discussed, detailing the data collection and analysis processes. The results of the analysis are then presented, followed by a discussion of the findings, linking them back to existing literature and the broader implications for patient care. Finally, the conclusion synthesizes the key insights from the study, emphasizing the value of a phenomenological approach in understanding the patient experience in the context of gene therapy.

RESEARCH METHODS

Study Design

This study employs a phenomenological design, which is ideal for exploring the lived experiences of individuals and understanding the meaning they attach to these experiences (Fife, 2020). Phenomenology focuses on the subjective reality of participants and seeks to uncover the essence of a phenomenon from their perspective. In this study, an interpretative phenomenological analysis (IPA) approach was chosen due to its capacity to provide a deeper understanding of how individuals make sense of their personal experiences, particularly in the context of gene therapy. This method is particularly relevant to the research question as it allows for a detailed exploration of participants' emotional and psychological reactions to undergoing gene therapy for genetic disorders, focusing on how these experiences are perceived and interpreted by the individuals themselves.

Participants

Participants were selected using purposive sampling, ensuring that individuals had direct experience with the phenomenon under investigation (Kawamura, 2020). Inclusion criteria required participants to be individuals who had undergone gene therapy for genetic disorders (Clair, 2003). Specific focus was given to those with a minimum of six months post-treatment experience, to ensure they could reflect on the psychological and emotional changes after therapy. Exclusion criteria included individuals who were unable to provide informed consent due to cognitive impairment or those who had not completed the gene therapy process. The study involved 12 participants, including 6 males and 6 females, aged between 25 and 65, from various socioeconomic backgrounds. These participants provided rich insights into the psychological impact of gene therapy.

Data Collection

Data was collected through in-depth, semi-structured interviews, which were designed to allow participants to freely express their thoughts and feelings while still addressing key aspects of the research question. Interviews were conducted individually, in a private setting to ensure confidentiality and comfort, and each interview lasted approximately 45 to 60 minutes. The interviews were audio-recorded with participants' consent and transcribed verbatim. The interview guide was developed through an iterative process, beginning with a review of existing literature on the psychosocial aspects of gene therapy and refined through consultation with two experts in clinical psychology and genetic counseling. A pilot interview with one participant (not included in the final analysis) was conducted to test the clarity and relevance of the questions. Based on feedback, the guide was revised to ensure open-ended, non-leading questions that covered domains such as emotional reactions, identity changes, social relationships, and future expectations.

Data Analysis

Data was analyzed using Interpretative Phenomenological Analysis (IPA), which is particularly suited for exploring how individuals make sense of their personal and social worlds. The analytic process followed the structured steps outlined by Smith, Flowers, and Larkin (2009): (1) transcripts were read and reread to achieve immersion in the data; (2) initial notes were made, focusing on descriptive, linguistic, and conceptual comments; (3) emergent themes were identified by condensing significant statements into concise phrases; (4) themes were clustered into superordinate categories to capture broader patterns; (5) connections between themes were mapped across individual cases; and (6) patterns were examined across all participants to identify shared meanings and divergences. NVivo software was used to facilitate systematic coding and data organization. To enhance trustworthiness, two researchers independently coded a subset of transcripts and discussed discrepancies until consensus was reached, ensuring analytic rigor and credibility.

Ethics

The ethical considerations for this study were rigorously followed. Informed consent was obtained from all participants prior to their involvement in the study. They were fully informed of the nature of the research, the voluntary nature of their participation, and their right to withdraw at any time without consequence. Confidentiality was ensured by anonymizing all data and using pseudonyms in the reporting of findings. All audio recordings and transcriptions were securely stored and only accessible to the research team. The study adhered to the ethical standards outlined in the Declaration of Helsinki and was approved by the relevant ethics committee. Participants were given the opportunity to review the transcripts for accuracy and provided their written consent for the use of their data in the study.

RESULTS

Psychological Impact of Gene Therapy on Patients

The experience of patients undergoing gene therapy for genetic disorders revealed profound psychological impacts, both positive and negative. Overall, participants consistently reported a dual emotional trajectory: initial optimism followed by lingering uncertainty. The sense of hope and relief was particularly salient among those who had long endured chronic illness, as gene therapy symbolized a renewed possibility of stability and longevity. This highlights the transformative psychological significance of moving from perceived incurability to the promise of treatment. One participant illustrated this sentiment:

"When I first heard that I could be treated with gene therapy, I felt a huge weight lift off my shoulders…"

However, the narratives also reflected persistent anxiety about the durability of the therapy's effects. This ambivalence underscores a broader psychological tension between hope and fear, a dynamic central to understanding how patients negotiate the meaning of gene therapy within their lives. Importantly, this finding aligns with the research question by demonstrating that the psychological journey extends beyond clinical outcomes to the domain of identity and future orientation.

Coping Strategies and Support Systems

Coping mechanisms emerged as a central theme in the experiences of patients undergoing gene therapy.

Participants' reliance on interpersonal support systems—family, peers, and healthcare professionals—illustrates that adaptation is not an individual process but one embedded in relational contexts. Emotional reassurance from close relatives was frequently identified as a buffer against anxiety, confirming the role of social scaffolding in sustaining psychological resilience. As one participant noted:

"I couldn't have gotten through it without my family…"

In addition, peer support groups provided validation and normalized patients' fears and uncertainties, suggesting that shared narratives are crucial for constructing a collective sense of meaning. This reflects a key interpretive insight: coping is not merely about managing stress but about reconstructing a coherent life story in the face of biomedical transformation.

Social and Family Dynamics Post-Therapy

The social and familial impacts of gene therapy also emerged as significant factors in the overall patient experience.

Gene therapy did not only alter individual health perceptions but reshaped family dynamics, often strengthening relational bonds through shared struggle and hope. This collective orientation highlights how biomedical interventions ripple outward into family systems, reinforcing the need to consider psychosocial as well as medical outcomes. One participant reflected on the deepened familial ties:

"My relationship with my family has never been stronger…"Yet, the findings also indicate that not all families were equally equipped to provide psychological support. Tensions sometimes emerged when relatives emphasized medical progress while overlooking the patient's emotional burdens. This divergence illustrates how differing expectations within families can exacerbate patients' sense of isolation, a factor that complicates recovery and adaptation.

Perception of Change in Health Status

The perception of change in health status was a central theme for participants.

For many, the therapy was experienced as a profound transformation, not only in terms of physical improvement but also as a reorientation of self-identity. Patients described new feelings of normalcy and possibility, signaling that the therapy's psychological impact extended into existential domains such as hope, agency, and future planning. As one participant shared:

"After the treatment, I feel like a new person…"At the same time, several participants reported difficulty reconciling long-standing illness identities with emerging perceptions of restored health. This tension underscores a critical interpretive point: the transition from chronic illness to perceived recovery is psychologically demanding, as it disrupts established self-concepts and coping patterns. Thus, gene therapy does not merely heal the body but compels patients to renegotiate their sense of self.

Essential Conclusion

The findings from this study underscore the complexity of the psychological experience of patients undergoing gene therapy for genetic disorders. The interpretive analysis reveals that patients' journeys are characterized by ambivalence—oscillating between hope and anxiety, resilience and vulnerability, connection and isolation. These themes illuminate the broader research question by demonstrating that gene therapy's impact cannot be understood solely in biomedical terms but must be contextualized within patients' emotional lives, family dynamics, and identity transformations.

Therefore, the results highlight the necessity of integrating psychological and social support into clinical pathways, ensuring that gene therapy is addressed as both a medical and existential process.

DISCUSSION

Summary of Key Findings

The study reveals that patients undergoing gene therapy for genetic disorders experience complex emotional and psychological transformations that are integral to their overall journey (Fernandez-Pineda et al., 2024). While many report initial hope and relief, these feelings are often followed by uncertainty and anxiety, particularly regarding the long-term effects of the therapy. Rather than viewing these emotions as isolated reactions, the findings suggest a broader psychological trajectory in which optimism is tempered by ongoing concerns about durability and risk. This duality highlights the need to situate patient experiences within both individual meaning-making processes and the social context of chronic illness.

Contribution of Findings to the Research Question

The findings contribute significantly to the understanding of how individuals perceive and process the psychological impact of gene therapy (Murray et al., 2024). By integrating emotional responses with coping strategies, identity reconstruction, and social dynamics, the study demonstrates that patients' experiences extend beyond clinical milestones. The contribution lies in showing that gene therapy is not only a medical breakthrough but also a process that reshapes how patients define health, self, and future possibilities. This addresses the central research question by positioning psychological adaptation as a critical dimension of therapeutic success.

Relation to Previous Literature and Theoretical Perspectives

These findings align with previous phenomenological studies on other medical treatments, such as cancer therapies, which similarly emphasize the psychological challenges patients face during and after treatment. For example, research by Ng et al (2020) on cancer patients revealed themes of fear, hope, and identity transformation, which resonate with the emotional rollercoaster experienced by gene therapy patients. However, unlike oncology contexts where survival is a dominant concern, gene therapy patients reported unique identity disruptions stemming from the sudden transition from chronic illness to potential normalcy. This divergence underscores the distinct psychosocial landscape of genetic disorders and emphasizes why gene therapy requires tailored psychological frameworks.

Furthermore, the role of social support and coping mechanisms, as highlighted in the present study, echoes findings from studies on chronic illness and medical interventions (Camilleri et al., 2023), which emphasize the importance of familial and peer support in managing emotional distress. Yet, the present findings expand on this by showing that not all support is uniformly beneficial—some participants reported that family enthusiasm overlooked their psychological strain. This nuance adds complexity to the literature, suggesting that social networks can both alleviate and exacerbate psychological burdens.

The study also contributes theoretically by employing an interpretative phenomenological lens, which allows for a deeper exploration of how patients derive meaning from their therapeutic journey. This approach highlights the reconstruction of personal identity as central to patient adaptation—a theme less visible in quantitatively oriented research—and invites further theoretical development on identity shifts in post-treatment contexts.

Implications of Findings

The findings of this study have significant scientific and practical implications, particularly in the realm of patient care and psychological support for individuals undergoing gene therapy (Mayopoulos et al., 2021).

First, they indicate the necessity of embedding psychosocial care within treatment pathways, as psychological well-being is intertwined with treatment adherence and long-term adjustment. Second, the results suggest that clinicians must anticipate ambivalent emotions rather than assume unidirectional improvement in patient outlook.

Importantly, the implications extend to diverse patient populations. Patients from lower socioeconomic backgrounds may have less access to sustained psychosocial resources, while cultural contexts may shape how hope, anxiety, and identity changes are expressed or stigmatized. For instance, in collectivist societies, family expectations may intensify patient stress, whereas in more individualistic cultures, the challenge may center on personal adaptation to independence. Tailoring psychosocial interventions to these cultural and socioeconomic realities is crucial to ensure equitable patient care.

Limitations of the Study

Despite the valuable insights gained, there are several limitations to this study that must be considered when interpreting the findings (McConville et al., 2020; Wen et al., 2021). First, the sample size of 12 participants limits the generalizability of the results, as a larger and more diverse sample might yield different insights (Katznelson et al., 2021). Additionally, the study focused on patients who had already undergone gene therapy and had a minimum of six months post-treatment experience. This means the findings reflect the experiences of individuals who have had time to process their treatment, potentially overlooking the initial, more acute psychological reactions to gene therapy. Furthermore, most participants were from relatively similar cultural contexts, limiting the applicability of results across diverse patient populations. This underscores the importance of future studies incorporating cross-cultural comparisons to identify variations in psychological adjustment.

Prospective Directions for Future Research

The findings of this study pave the way for future research into the psychological impacts of gene therapy, particularly in under-explored areas (Lindegaard, 2023). One promising direction would be to investigate the emotional responses of patients during the early stages of treatment, as well as how these reactions evolve over time (Miller, 2019). Longitudinal studies could provide a more comprehensive understanding of the psychological trajectory of patients from the point of treatment initiation to long-term recovery or adaptation (Dotto et al., 2022; Warren et al., 2021). Additionally, future research should purposefully sample across cultural, ethnic, and socioeconomic groups to explore how diverse patient populations negotiate the psychological challenges of gene therapy. Comparative studies could reveal how cultural values, health beliefs, and resource availability shape coping strategies, identity reconstruction, and perceived outcomes.

CONCLUSION

This study explored the emotional and psychological experiences of patients undergoing gene therapy for genetic disorders, aiming to fill the gap in understanding the subjective impacts of this treatment. The findings revealed that while patients experienced initial hope and relief, they also faced significant psychological challenges, including anxiety and uncertainty about long-term outcomes. These emotional complexities highlight the need for comprehensive patient care that addresses both physical and psychological well-being. By focusing on the lived experiences of patients, this study contributes to a more nuanced understanding of gene therapy, overcoming the limitations of previous research that primarily focused on clinical outcomes. For clinical practice, the findings underscore the importance of integrating structured psychosocial support into gene therapy programs. Healthcare providers should be trained to recognize signs of anxiety, identity disruption, and emotional strain, and multidisciplinary teams—including psychologists and genetic counselors—should be embedded within treatment pathways. Establishing peer-support or patient-mentor programs may also help normalize experiences and reduce feelings of isolation.

For policy development, the study highlights the need to design guidelines that standardize psychological screening and support as part of gene therapy protocols. Policymakers should consider funding mechanisms that ensure equitable access to psychosocial care, particularly for patients from low-resource settings. Culturally sensitive approaches should be promoted to account for the diverse ways patients and families interpret treatment and health identity changes. Future studies should expand on these findings by investigating patient experiences at earlier stages of treatment and in more diverse populations, while also evaluating the effectiveness of integrated care models. By doing so, research can inform both practice and policy, ensuring that gene therapy is delivered not only as a biomedical innovation but also as a holistic intervention that safeguards emotional and psychological well-being.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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