



An Interpretative Exploration of CRISPR-Cas9 Gene Therapy Experiences in Rare Genetic Disorders

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ABSTRACT

to correct genetic defects in rare disorders. However, limited attention has been given to how patients perceive and adapt to such interventions. This study aims to explore patients' lived experiences and the meanings they ascribe to CRISPR-Cas9 therapy. An interpretative phenomenological approach (IPA) was employed, using semi-structured interviews with twelve participants who underwent CRISPR-Cas9 treatment. Analysis revealed four key themes: confronting uncertainty, redefining hope, negotiating identity, and navigating social acceptance. Findings show that the impacts of CRISPR therapy extend beyond clinical outcomes, encompassing psychological, emotional, and social dimensions. These insights emphasize the need for patient-centered frameworks in genomic medicine and provide ethical and research implications for integrating technological innovation with human experience..



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INTRODUCTION

Advancements in gene therapy, particularly CRISPR-Cas9, have transformed modern medicine by enabling precise correction of genetic defects and offering new hope for rare disorders (Mukhlis, Maryam, et al., 2023; Mukhlis et al., 2024). Among the most groundbreaking innovations, CRISPR-Cas9 gene-editing technology has enabled precise and efficient modification of defective genes, significantly improving therapeutic outcomes for patients with rare and complex conditions. Globally, an increasing number of clinical trials have demonstrated promising results, positioning CRISPR-based interventions as a transformative tool in precision medicine and personalized healthcare.

Despite these biomedical breakthroughs, the introduction of CRISPR therapies has also created profound psychosocial and ethical implications for patients and their families. The decision to undergo gene-editing therapy extends beyond medical considerations, encompassing personal identity, family dynamics, cultural values, and societal perceptions of "normalcy." Individuals undergoing treatment often confront emotional challenges related to uncertainty, hope, and self-perception, as they navigate the transformative potential of altering their genetic makeup (Jaganathan et al., 2019; Meng et al., 2022). Such experiences are deeply rooted in broader social and cultural contexts, where diverse attitudes toward biotechnology influence how individuals interpret and integrate these medical advancements into their lives.

While scientific literature has extensively addressed the clinical safety, efficacy, and technical feasibility of CRISPR-Cas9 applications (Ahmed, 2020; Satterstrom et al., 2020), less attention has been given to the subjective experiences of patients who undergo these therapies. Understanding how individuals make sense of their therapeutic journey, reconstruct personal identity, and adapt to

changing perceptions within their communities is critical for shaping patient-centered care and informing ethical frameworks in genomic medicine.

Given these complexities, there is a growing need to explore the lived experiences of patients receiving CRISPR-based therapies. A phenomenological approach offers a powerful framework to uncover the meanings, emotions, and personal interpretations that are often overlooked in quantitative studies. By focusing on the essence of patients' subjective realities, phenomenology provides a nuanced understanding of how individuals experience and internalize the profound impacts of gene therapy, thereby bridging the gap between biomedical advancements and human-centered care.

Research on the subjective experiences of patients undergoing innovative treatments, such as CRISPR-Cas9 gene therapy, has increasingly become a critical area within the field of molecular medicine and psychosocial health. While biomedical studies have established the clinical safety, efficacy, and therapeutic potential of CRISPR technologies (Mukhlis, Janwari, et al., 2023; Mukhlis & Abdullah, 2025), significantly less is known about how patients personally interpret, adapt to, and make sense of these profound medical interventions. Understanding these lived experiences is essential because such treatments do not merely affect biological outcomes but also reshape personal identity, self-perception, and social relationships.

However, exploring these complex experiences presents substantial methodological challenges. Much of the existing literature relies heavily on quantitative research designs, emphasizing biomedical parameters such as gene-editing efficiency, immune responses, or adverse effects. While these findings are valuable, quantitative frameworks are insufficient to capture the depth of patients' emotional, psychological, and existential realities following genetic modification. For instance, prior surveys and clinical trials often reduce patient responses to numerical measures, neglecting the nuanced meanings embedded in their personal narratives and social contexts [Ahmed et al., 2023].

Consequently, previous approaches have struggled to uncover the essence of the phenomenon, particularly the profound ways in which individuals interpret their identities, relationships, and roles after undergoing CRISPR-based interventions (Burunkaya et al., 2024; Subasri et al., 2021). This gap highlights the limitations of traditional methodologies in providing a holistic understanding of the therapeutic journey. A qualitative approach rooted in phenomenology offers a more effective framework to explore these dimensions, as it prioritizes participants' subjective perspectives and seeks to uncover the meanings and transformations embedded within their lived experiences.

By focusing on the interpretative dimension of patients' realities, phenomenology facilitates a deeper understanding of the psychological and social consequences of CRISPR therapy that quantitative metrics alone cannot provide. This emphasis on meaning-making forms the foundation for addressing the central objectives of the current study, which seeks to bridge the gap between biomedical advancements and patient-centered perspectives.

Despite remarkable advancements in CRISPR-Cas9 gene-editing technologies, current research has largely prioritized biomedical outcomes, focusing on treatment efficacy, genetic correction rates, and potential adverse effects (Alejo-Armijo et al., 2022; Pearce et al., 2024). These studies have provided valuable insights into clinical effectiveness; however, they have primarily relied on quantitative frameworks and structured assessments that fail to capture the depth of patients' subjective experiences. As a result, critical questions remain unanswered regarding how individuals perceive, interpret, and internalize the transformative nature of undergoing gene-editing therapy.

Existing approaches, such as clinical surveys and standardized patient-reported outcome measures, offer only limited insights into the personal meanings, identity shifts, and emotional challenges faced by patients (Tran et al., 2019). While these tools are effective for tracking treatment success, they lack the capacity to reveal the nuanced psychological, social, and existential dimensions embedded in patients' lived experiences. Consequently, the current understanding of how CRISPR therapy impacts self-perception, family relationships, and social integration remains fragmented and incomplete.

To address this gap, a phenomenological approach offers a rigorous and systematic framework for exploring the essence of patients' experiences. By prioritizing individual narratives and personal meaning-making, this method enables researchers to uncover the emotional, cognitive, and social transformations that emerge during and after CRISPR-based interventions. Such an inquiry not only complements existing biomedical knowledge but also contributes to the development of patient-centered care strategies and ethically informed clinical practices.

Recent studies have explored the psychosocial implications of gene therapy, but most have focused primarily on clinical outcomes rather than patients' lived experiences. For instance, Ivanets et al. (2021) examined emotional adaptation following gene-editing interventions, highlighting the psychological complexity of identity reconstruction. Similarly, Leung et al. (2019) discussed the ethical and societal dimensions of CRISPR therapies but did not capture the subjective meaning-making processes of patients. These findings underscore the growing need for studies that go beyond biomedical success rates to explore personal interpretations and social contexts. By integrating insights from these studies, the present research positions itself to provide a deeper and more holistic understanding of the phenomenon.

To address this need, the study employs an Interpretative Phenomenological Analysis (IPA) approach, which focuses on uncovering the personal meanings embedded in patients' experiences. IPA was selected because it allows for an in-depth exploration of participants' emotional, cognitive, and social realities beyond what structured surveys or clinical assessments can provide. This approach directly responds to the gap identified earlier, seeking to understand how patients perceive, interpret, and adapt to the transformative impacts of CRISPR-Cas9 therapy. Through rich narrative accounts, this method captures the essence of subjective experiences often overlooked in quantitative research. In doing so, it bridges the divide between clinical advancements and human-centered perspectives.

This article is structured into several interconnected sections to guide readers through the study (Mukhlis, 2025a; Mukhlis & Saidah, 2025). The Introduction provides the contextual background and rationale for the research, followed by a detailed discussion of the phenomenon and its significance. The Method section describes the phenomenological approach, data collection, and analytical procedures. The Results present the thematic findings, supported by participants' direct quotations to preserve authenticity. Finally, the Discussion synthesizes the findings, explores theoretical and practical implications, and concludes with recommendations for future research.

RESEARCH METHODS

Study Design

A phenomenological research design was employed to explore the lived experiences of patients who underwent CRISPR-Cas9 gene therapy for rare genetic disorders (Daly, 2007; Murphy & Dingwall, 2017). The phenomenological approach was selected due to its focus on uncovering subjective meanings and understanding the essence of participants' personal experiences. Within this study, an Interpretative Phenomenological Analysis (IPA) framework was applied, allowing for an in-depth exploration of how participants interpreted, assigned meaning to, and reflected upon their therapeutic journey. IPA is particularly suitable when the objective is to capture not only descriptive accounts of lived experiences but also their deeper psychological, emotional, and social interpretations.

Participants

Participants consisted of individuals diagnosed with rare genetic disorders who had completed CRISPR-Cas9 therapy within the past two years (Clair, 2003; Fenton & Baxter, 2016). A purposive sampling strategy was utilized to ensure the inclusion of participants with relevant and rich experiences of the phenomenon. The inclusion criteria were:

- A confirmed diagnosis of a rare genetic disorder.
- Completion of CRISPR-Cas9 therapy at least six months prior to data collection.
- Age between 18 and 50 years.

- Ability to communicate experiences clearly in English.

Participants with cognitive impairments that could limit their ability to recall or articulate experiences were excluded. A total of 12 participants (7 females and 5 males) were included, with an average age of 32.4 years. The demographic diversity of the participants allowed for a wide range of perspectives while maintaining relevance to the phenomenon under investigation.

Data Collection

Data were collected through semi-structured, in-depth interviews to capture the richness and depth of participants' personal experiences (Fife, 2020; Kawamura, 2020). An interview guide was developed based on the research question and included open-ended prompts to encourage participants to describe their thoughts, emotions, and interpretations freely. Examples of guiding prompts included:

“Can you describe your initial thoughts and feelings when you were introduced to CRISPR therapy?”

“How has your perception of yourself changed since undergoing the treatment?”

Interviews were conducted face-to-face in a private clinical consultation room to ensure confidentiality and comfort, while virtual interviews were arranged when participants were unable to attend in person. Each interview lasted between 45 and 75 minutes and was audio-recorded with participants' consent. Field notes were also maintained to capture non-verbal expressions and contextual observations. All recordings were transcribed verbatim, and pseudonyms were assigned to protect participants' identities.

Data Analysis

Data were analyzed using the Interpretative Phenomenological Analysis (IPA) framework (Mueller et al., 2024; Perry, 2023). This method was chosen for its capacity to reveal nuanced layers of meaning embedded within personal narratives. The analysis followed a systematic, iterative process consisting of the following steps:

1. Immersion in the Data – Transcripts were read multiple times to achieve a deep familiarity with participants' accounts.
2. Identification of Meaning Units – Relevant statements reflecting key experiences and emotions were extracted and coded.
3. Development of Emergent Themes – Codes were clustered into themes that captured shared patterns across participants.
4. Interpretative Layering – Deeper meanings were derived by linking themes to participants' contextual realities and psychosocial perspectives.
5. Validation of Findings – Themes were reviewed against the original transcripts to ensure that interpretations accurately reflected participants' lived experiences.

NVivo 14 software was used to support data organization and facilitate the coding process, but the interpretation remained grounded in the phenomenological framework rather than being driven by software outputs.

Ethical Considerations

Ethical approval was obtained from the Institutional Research Ethics Committee before data collection commenced (Hammersley, 2003; McMahon & McGannon, 2024). Informed consent was secured from all participants through written documentation, which explained the purpose of the study, the voluntary nature of participation, and their right to withdraw at any stage without consequence. Anonymity and confidentiality were strictly maintained by assigning pseudonyms and securely storing all digital files in encrypted formats. This study adhered to the ethical principles

outlined in the Declaration of Helsinki and complied with relevant institutional and national research ethics standards.

RESULTS

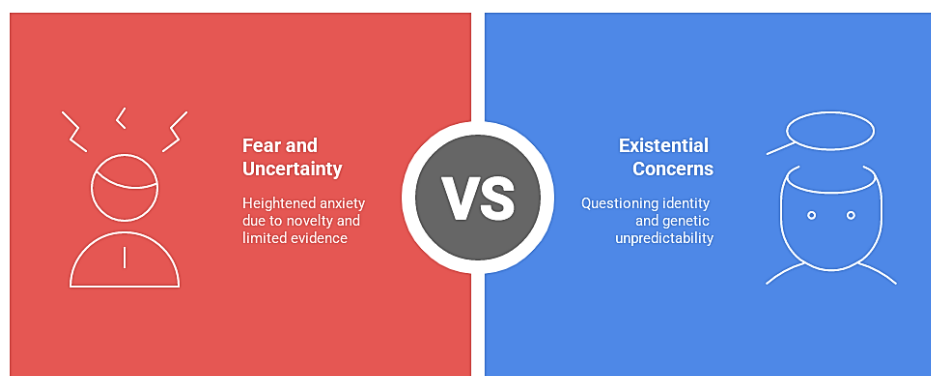
Confronting Uncertainty and Fear

Participants consistently described profound feelings of uncertainty and fear when initially considering CRISPR-based gene therapy. The novelty of the treatment, combined with limited clinical evidence, created heightened anxiety regarding safety, long-term outcomes, and potential ethical implications.

“When my doctor first mentioned CRISPR, I was terrified. I felt like they were about to change my entire DNA, and I wasn’t sure who I would become after that.” — Participant 3

Several participants expressed that this fear was not only biomedical but also existential, questioning their sense of identity and the unpredictability of altering their genetic makeup. This emotional struggle became a recurring point of tension prior to undergoing treatment.

How to manage emotional responses to CRISPR gene therapy?



Redefining Hope Through Scientific Breakthroughs

Despite initial fears, participants reported experiencing a renewed sense of hope after receiving detailed explanations from clinicians about CRISPR’s mechanisms and potential outcomes. This sense of optimism emerged as patients developed trust in the scientific process and envisioned the possibility of reclaiming control over their health.

“For the first time in years, I felt like I had a chance to live without constant hospital visits. It gave me a hope I didn’t know I still had.” — Participant 5

The therapy symbolized a scientific turning point, shifting participants’ perspectives from enduring a chronic genetic condition to actively participating in a transformative medical journey. This reorientation toward hope was often linked to increased resilience and motivation to complete the therapy process.

Negotiating Identity and Self-Perception

Following treatment, participants reported complex emotional responses regarding changes in their self-perception. Some described a sense of becoming “genetically different” from who they were before, leading to reflections on identity and belonging.

“It’s strange... knowing that a part of me has literally changed. I feel healthier, but I also wonder if I’m still the same person I used to be.” — Participant 7

For many, this negotiation of identity extended beyond biology and encompassed psychological and social dimensions, such as shifting family dynamics, stigma, and the meaning of “normalcy” in the context of living with altered genetics.

Navigating Social Acceptance and Ethical Dilemmas

Participants highlighted challenges related to social acceptance and societal perceptions of genetic modification. While some reported supportive family and community responses, others encountered skepticism, stigma, and ethical debates surrounding CRISPR's implications.

“Some relatives celebrated my treatment, but others said I was ‘unnatural’ now. It’s hard to explain what this means when society doesn’t understand the science.” — Participant 2

This theme underscores the broader social and cultural contexts influencing participants’ post-treatment experiences, demonstrating that the impacts of CRISPR therapy extend beyond individual biology and into collective narratives about ethics and identity.

Essential Synthesis of Findings

Across these themes, the findings reveal that CRISPR-Cas9 therapy is more than a biomedical intervention; it is an existential journey intertwined with fear, hope, identity reconstruction, and social negotiation. Participants’ lived experiences highlight the importance of considering psychosocial dimensions alongside clinical outcomes in developing patient-centered gene therapy practices.

DISCUSSION

Contribution of Findings to the Research Question

The study provides an in-depth understanding of how patients perceive and internalize their experiences before, during, and after CRISPR-Cas9 therapy. The emergence of fear and uncertainty highlights the existential dimensions of confronting irreversible changes to one’s genetic identity, an aspect rarely captured in biomedical literature. At the same time, the findings reveal a parallel narrative of hope and empowerment, where participants reconstruct personal meaning and agency through engaging with cutting-edge medical science. Importantly, the study contributes a unique phenomenological perspective by showing that patients’ journeys are not defined solely by clinical success but also by their interpretive processes, emotional negotiations, and social realities. These insights extend beyond answering the research question; they enrich our understanding of how technological innovations intersect with human meaning-making, thereby informing the development of more patient-centered therapeutic frameworks.

Connection with Previous Literature and Theoretical Perspectives

The findings align with existing studies emphasizing the psychological and social dimensions of gene therapy while offering deeper interpretative insights that previous research has not fully captured. Mukhlis, (2025) reported that patients often experience identity shifts after gene-editing interventions, which resonates with the present finding on negotiating self-perception following CRISPR therapy. Similarly, Cannon et al. (2021) highlighted societal stigma and ethical concerns surrounding genome editing, which complement the theme of navigating social acceptance revealed in this study. However, unlike prior studies that primarily relied on surveys or structured interviews, the phenomenological approach adopted here provides richer, context-sensitive interpretations of patients’ subjective realities. The findings also align with interpretative phenomenological frameworks (Hough et al., 2024), which emphasize that meaning emerges through participants’ sense-making rather than objective measurements. By situating individual narratives within broader cultural and ethical contexts, this research advances theoretical understanding of how biotechnology reshapes identity, belonging, and social integration.

Implications of the Findings

The findings of this study hold significant scientific, clinical, and sociocultural implications. From a clinical perspective, the results suggest that understanding patients’ subjective experiences is essential for developing patient-centered approaches in gene therapy (Mukhlis, Suradi, et al., 2023). By uncovering how individuals interpret and adapt to the transformative nature of CRISPR-Cas9 therapy, the study highlights the need for healthcare providers to integrate psychological and social support mechanisms alongside biomedical interventions. On a broader sociocultural level, the themes

of identity reconstruction and social acceptance illustrate how genetic modification intersects with societal norms and cultural perceptions of normalcy, ethics, and belonging. These insights are relevant not only to patients receiving CRISPR-based therapies but also to policymakers, bioethicists, and clinical practitioners seeking to design holistic frameworks for implementing genomic medicine within diverse populations.

Limitations of the Study

While this research offers valuable insights, certain limitations must be acknowledged. First, the study involved a relatively small sample size of twelve participants, which is consistent with phenomenological approaches but restricts the generalizability of findings to broader populations (Mukhlis, Arifin, Ridwan, & Zulbaidah, 2025; Mukhlis, Arifin, Ridwan, Zulbaidah, et al., 2025). Second, all participants were drawn from clinical settings where access to CRISPR therapy was available, potentially excluding diverse perspectives from individuals with limited healthcare resources or cultural backgrounds where gene-editing interventions are less accepted. Additionally, the study relied on self-reported narratives, which, while rich in meaning, may be influenced by memory biases, emotional states, or social desirability during interviews. These limitations do not diminish the validity of the findings but rather emphasize the contextual nature of phenomenological research, providing direction for future studies to examine more diverse samples and cross-cultural perspectives.

Prospective Directions for Future Research

The results of this study open several avenues for future investigation into the psychosocial dimensions of gene-editing therapies (Psihogios et al., 2019; Sivathamboo et al., 2022). Longitudinal studies are recommended to examine how patients' interpretations and identity negotiations evolve over time, particularly as CRISPR technologies become more widespread in clinical practice. Comparative cross-cultural research may also enrich understanding of how societal values and ethical frameworks shape the meaning patients ascribe to their experiences. Furthermore, integrating phenomenological findings with quantitative measures of psychological well-being could provide a more comprehensive picture of treatment outcomes. By combining experiential insights with clinical data, future research can contribute to designing inclusive, ethically grounded, and patient-centered frameworks for advancing gene therapy in diverse healthcare contexts.

CONCLUSION

This study explored the lived experiences of patients undergoing CRISPR-Cas9 gene therapy for rare genetic disorders using an interpretative phenomenological approach. The findings revealed four key themes—confronting uncertainty and fear, redefining hope, negotiating identity, and navigating social acceptance—that illuminate the psychological, emotional, and social dimensions of patients' therapeutic journeys. By focusing on participants' meaning-making processes, the study addresses a critical gap in the literature, offering a deeper understanding of how individuals perceive and adapt to transformative genomic interventions. These insights contribute to advancing patient-centered care by highlighting the importance of integrating psychosocial support within clinical frameworks. While the findings provide significant contributions, future research could expand on these results by exploring cross-cultural perspectives and conducting longitudinal studies to capture evolving patient experiences over time. This study demonstrates the value of phenomenology in bridging biomedical innovation with human-centered understanding, laying the foundation for more inclusive and ethically informed practices in genomic medicine.

CONFLICT OF INTEREST

The authors declare no conflict of interest related to the design, conduct, or reporting of this study. All procedures were conducted independently, and there was no financial or personal relationship that could have influenced the research outcomes.

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