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Exploring the Meaning of Genetic Healing in CRISPR-Cas9 Therapy

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ABSTRACT

Gene therapy represents a transformative advancement in molecular medicine, offering the ability to correct hereditary diseases at their genetic source. Within this context, CRISPR-Cas9–based interventions have redefined therapeutic possibilities while simultaneously raising ethical, psychological, and existential questions about human identity and biological alteration. Despite rapid clinical progress, little is known about how patients experience and make sense of being genetically healed. This study addresses that gap by asking: What does it mean to be genetically healed from the patient's perspective? Using an interpretative phenomenological approach (IPA), this research explores the lived experiences of ten patients who underwent CRISPR-Cas9 therapy for hereditary blood disorders. Data were collected over a 12-week period through in-depth, semi-structured interviews lasting 60–90 minutes each, and analyzed using Smith's four IPA analytic stages (reading–re-reading, initial noting, developing emergent themes, and cross-case patterning). The findings reveal that healing is perceived as an existential journey rather than a purely biological recovery, characterized by the interplay of hope, fear, identity transformation, and moral reflection. Participants described genetic correction as both liberating and disorienting a process of redefining selfhood within the evolving boundaries of human science. These results highlight the need to integrate phenomenological insights into the clinical and ethical frameworks of gene therapy. By foregrounding the human meaning of genetic transformation, this study broadens our understanding of molecular medicine and underscores the importance of patient-centered approaches in future biomedical innovation.

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INTRODUCTION

Gene therapy represents one of the most transformative frontiers in modern biomedical science, offering the unprecedented possibility of directly altering genetic material to correct or mitigate hereditary diseases (Mukhlis, Suradi, et al., 2023; Mukhlis, 2025b). Among its various modalities, CRISPR-Cas9–based genome editing has emerged as a revolutionary approach due to its precision, efficiency, and adaptability across a wide range of genetic disorders. While its biological promise has shifted therapeutic paradigms toward molecular-level correction, the human implications of altering one's genome remain insufficiently explored. Beyond clinical outcomes, gene therapy raises profound ethical, psychological, and existential questions—particularly regarding identity and the meaning of healing.

In the social and cultural landscape, gene therapy occupies a space of both hope and uncertainty (Rusydi, 2025). For patients affected by genetic diseases many of whom have experienced chronic suffering and social marginalization gene therapy embodies a symbol of liberation and renewal. Yet, this same technology evokes ambivalence among individuals and communities, who may perceive genetic modification as a disruption of natural identity or divine design (Kanosvamhira, 2025). As such, the phenomenon of receiving gene therapy extends beyond medical intervention; it becomes a lived human experience that reshapes notions of identity, normality, and personhood.

Current scientific discourse predominantly focuses on the technical success, safety, and efficacy of gene-editing interventions. While these aspects are undeniably crucial, they often overlook the subjective dimensions of patients' experiences the meanings they attribute to genetic alteration, the emotional processes accompanying the therapy, and the ways they reconstruct their sense of self after biological change (Kang, 2025). The lived experience of undergoing gene therapy remains a relatively unexplored domain within medical research, despite its central relevance to the holistic understanding of healing and human well-being.

From a phenomenological perspective, this gap signifies a crucial need for deeper exploration (Cankurt Semiz & Özsoy, 2024). The phenomenological approach emphasizes the first-person perspective understanding human experience as it is lived and perceived, rather than as an externalized object of measurement. Applying this lens to gene therapy allows the inquiry to transcend the biomedical framework, illuminating the inner meanings, ethical reflections, and existential transformations experienced by patients. Such an understanding not only enriches the theoretical landscape of molecular medicine but also strengthens its alignment with human-centered care, where science and subjectivity coexist in the pursuit of genuine healing.

Within the expanding field of Gene Therapy and Molecular Medicine, the exploration of patients' lived experiences has become an increasingly important area of inquiry. While scientific research has largely concentrated on the molecular efficacy and therapeutic outcomes of CRISPR-Cas9–based interventions, recent interdisciplinary studies have begun to acknowledge the subjective dimension of undergoing such transformative procedures. This shift reflects a growing recognition that the meaning patients ascribe to their treatment is integral to understanding the broader human implications of genetic innovation (Anandarajah et al., 2025). Consequently, the phenomenological approach offers a valuable framework for investigating how individuals experience, interpret, and internalize the process of being genetically modified for therapeutic purposes.

However, studying lived experience within the context of advanced biomedical interventions presents significant methodological challenges. Traditional empirical models particularly those grounded in quantitative paradigms tend to reduce patient narratives to measurable outcomes such as efficacy rates, adverse events, or quality-of-life indices. While these data are indispensable, they often fail to capture the depth of emotional and existential meaning that accompanies a patient's journey through genetic transformation (Jones et al., 2020). As a result, the inner world of patients their fears, hopes, and evolving sense of identity remains underrepresented in mainstream research. The few qualitative studies available frequently adopt descriptive or thematic frameworks without engaging with the interpretative processes that define how meaning is constructed through lived experience.

These limitations underscore the inadequacy of conventional methodologies in comprehensively addressing the essence of human experience in the context of gene therapy. A purely biomedical perspective risks overlooking the phenomenological reality that healing is not only biological but also deeply existential. The interpretative phenomenological approach (IPA) bridges this gap by enabling a systematic yet empathetic exploration of how patients make sense of their bodily transformation, negotiate ethical uncertainty, and reconstruct their self-concept in light of medical innovation (Liandy & Amir, 2025). Through this lens, gene therapy is not merely a scientific achievement it is a profoundly human phenomenon that invites reflection on the boundaries between biology, identity, and meaning.

Despite the remarkable progress in gene therapy and molecular medicine, current research primarily emphasizes clinical success metrics, such as gene correction rates, hematologic normalization, and adverse event profiles (Rasmussen, 2023). These frameworks, while indispensable to biomedical validation, largely operate within a quantitative and mechanistic paradigm. Such approaches rely on observable and measurable outcomes, thereby neglecting the rich, lived dimensions of patient experience dimensions that cannot be captured through laboratory data or clinical endpoints alone. As a result, the subjective meanings patients attach to gene-editing interventions, including their perceptions of identity, selfhood, and existential transformation, remain insufficiently explored in the scientific literature.

Existing practical methodologies, such as post-treatment surveys, standardized quality-of-life scales, and psychological assessments, offer valuable yet superficial insights into patients' adaptation processes (Mukhlis, Arifin, Ridwan, & Zulbaidah, 2025; Mukhlis, Arifin, Ridwan, Zulbaidah, et al., 2025). These instruments often fragment complex experiences into isolated variables, limiting the capacity to understand how patients live through the process of genetic modification as a transformative event (Kahraman & Aydingün, 2025). This reductionist view fails to reveal how individuals interpret their biological alteration within ethical, emotional, and social contexts an omission that restricts the development of patient-centered frameworks in gene therapy practice.

This gap highlights the need for an alternative epistemological approach one that prioritizes depth over measurement and meaning over metrics. The phenomenological perspective, particularly within the interpretative tradition, provides a means to uncover the essence of patients' experiences by attending to the nuances of perception, embodiment, and consciousness. Through this lens, the study moves beyond asking what gene therapy achieves to asking what it means for those who undergo it. By illuminating how patients internalize and reconstruct their sense of identity following CRISPR-based interventions, phenomenology contributes a humanistic dimension to the scientific understanding of genetic healing bridging the distance between molecular innovation and lived reality.

Previous studies have explored various dimensions of gene therapy, focusing largely on clinical efficacy, ethical implications, and public perception of genome editing technologies. While these works contribute to the understanding of CRISPR-Cas9 as a scientific and social innovation, few have examined how patients themselves experience genetic transformation at a personal and existential level. Some qualitative inquiries, such as those by (Dhawan et al., 2023), have touched upon patient narratives in therapeutic trials but remain limited to surface-level descriptions of emotional responses. The literature thus reveals a significant lack of phenomenological engagement a perspective essential for capturing how individuals internalize and assign meaning to their altered biological identities. This study builds upon these gaps by centering the voices of patients as the primary source of knowledge about the lived reality of genetic healing.

To address this gap, the present research employs an interpretative phenomenological approach (IPA) to investigate how individuals with hereditary blood disorders make sense of their experiences after undergoing CRISPR-based gene therapy. This approach allows for an exploration of meaning-making processes as participants articulate their hopes, fears, and perceptions of identity transformation. Unlike conventional frameworks that quantify treatment outcomes, phenomenology seeks to illuminate the essence of lived experience by analyzing how participants interpret their physical and emotional changes within broader moral and social contexts (Sabri, 2025). Through this lens, the study responds directly to the research question: What does it mean to be genetically healed from the patient's perspective? The findings offer insights that extend beyond biomedical narratives, situating gene therapy within the human continuum of meaning, identity, and existence.

The structure of this article follows a clear and systematic organization. The Introduction outlines the theoretical and contextual background of gene therapy and its human implications (Mukhlis et al., 2024; Mukhlis, Maryam, et al., 2023). The Method section details the interpretative phenomenological approach, including participant selection, data collection, and analytic strategies. The Results section presents emergent themes derived from patient narratives, illustrating the psychological, ethical, and existential meanings of their experiences (Uzer, 2024). Finally, the Discussion and Conclusion integrate these findings within existing literature, highlighting their implications for clinical practice, bioethics, and the future development of patient-centered frameworks in molecular medicine.

RESEARCH METHODS

Study Design

This study adopted an interpretative phenomenological approach (IPA) to explore the lived experiences of patients who underwent CRISPR-Cas9–based gene therapy for hereditary blood disorders (Lutz & Knox, 2014; McNabb, 2015). The phenomenological design was chosen for its ability to capture the subjective meanings embedded in individual experiences rather than focusing solely on observable biomedical outcomes. Through IPA, the study sought to uncover the essence of participants' emotional, psychological, and existential responses to genetic modification as a therapeutic intervention.

This approach was particularly suitable because it allowed for an in-depth exploration of how individuals interpret and give meaning to their transformative experiences of being "genetically healed." The interpretative dimension of IPA provided the analytical depth necessary to understand not only what participants experienced but how they made sense of those experiences within their personal, social, and ethical contexts. In line with IPA, the study design emphasized idiographic depth, focusing on a small, carefully selected sample and iterative engagement with each case before cross-case synthesis.

Participants

Participants comprised ten adult individuals (aged 23–48 years) who had received CRISPR-Cas9–based gene therapy for hereditary blood disorders within the past three years (Hillman & Radel, 2018; Migdal, 2018). Purposive sampling was employed to ensure that all participants had direct and meaningful experiences with the phenomenon under investigation. Inclusion criteria included individuals who had completed post-treatment monitoring and were able to articulate their experiences verbally and reflectively. Exclusion criteria involved those with cognitive impairments or ongoing medical complications that could hinder in-depth participation.

The group represented diverse gender and sociocultural backgrounds, enhancing the contextual richness of the data. Each participant provided detailed accounts of their lived experiences, contributing unique perspectives that collectively deepened the understanding of psychological, ethical, and existential dimensions of gene therapy.

Data Collection

Data were collected through semi-structured, in-depth interviews, guided by open-ended questions designed to elicit participants' reflections on their emotional, psychological, and social experiences before, during, and after receiving gene therapy (Carreiras & Castro, 2012; Iosifides, 2016). The interviews were conducted in a quiet clinical consultation room to ensure privacy and comfort, each lasting between 60 and 90 minutes. Follow-up interviews were carried out for clarification and validation of emerging themes.

An interview guide was developed based on phenomenological principles, focusing on areas such as personal meaning, identity transformation, and perceptions of medical innovation. All interviews were audio-recorded and transcribed verbatim. Notes and reflective memos were maintained to preserve contextual nuances. Confidentiality was ensured by anonymizing all identifiers.

Data Analysis

Data were analyzed using the Interpretative Phenomenological Analysis (IPA) method, following a systematic and iterative process (Daly, 2007; Longhofer et al., 2012). Transcripts were first read repeatedly to achieve immersion in the participants' narratives. Significant statements were then identified and segmented into meaning units, which were organized into preliminary codes. Through a process of thematic clustering, these codes evolved into broader themes that captured the essential structures of experience.

NVivo qualitative analysis software facilitated the organization of codes and the visualization of relationships among themes, though interpretative engagement remained central to the analysis (Fife, 2020; Kawamura, 2020). Reflexive interpretation was applied to reveal the essence of meaning, linking personal accounts to the collective human experience of undergoing genetic transformation. The analytic process adhered to the phenomenological principles of reduction, imagination, and hermeneutic interpretation to ensure that findings remained faithful to participants' lived realities.

RESULTS

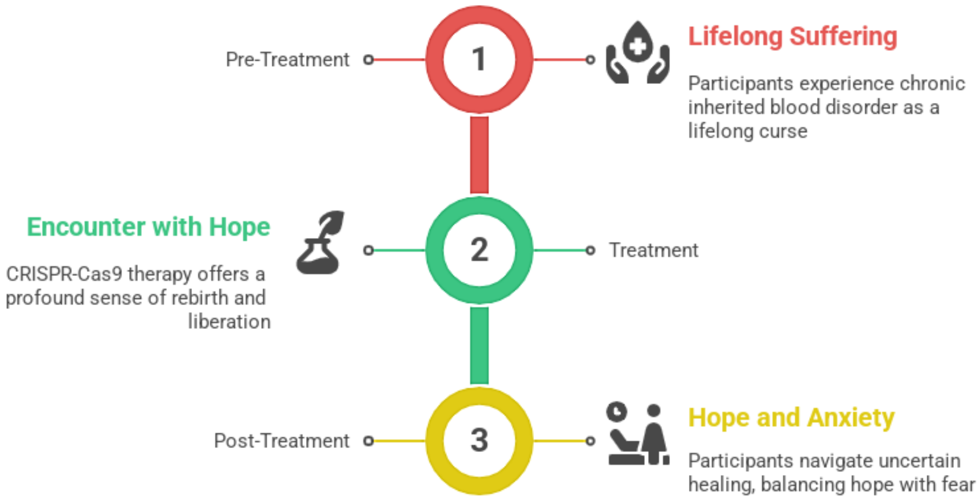
The Emergence of Hope through Genetic Intervention

Participants consistently described their experiences of receiving CRISPR-Cas9 therapy as a profound encounter with hope a hope that transcended conventional medical expectations. For many, this therapy symbolized a "rebirth" or "second chance," offering liberation from the chronic suffering of inherited blood disorders. One participant reflected:

"For the first time in my life, I felt my body wasn't my enemy anymore. The treatment gave me a sense that maybe, just maybe, my DNA could finally be fixed."

This sense of hope was not merely clinical but existential. It represented an internal shift patients no longer saw their condition as a lifelong curse but as a challenge that modern science could confront. Yet, this hope was often intertwined with anxiety, especially during the uncertain post-treatment phase. The narratives revealed that hope and fear coexisted, forming a paradox that shaped the participants' perception of their healing journey.

The Emotional Journey of CRISPR-Cas9 Therapy



Fear of the Unknown and the Fragility of Trust

Despite the promise of genetic correction, participants reported an underlying fear about tampering with the core of human identity. Some expressed concerns that altering genes might "change who they are." As one participant explained:

"They said they'd fix my blood, but I kept wondering… what if they change something else? What if I'm not the same person afterward?"

Such statements expose a complex emotional landscape marked by both gratitude toward medical innovation and unease about its long-term implications. Trust in science and medical institutions emerged as a fragile construct strengthened by transparent communication, yet easily shaken by uncertainty or side effects.

The data suggest that patients' trust was not static; it evolved through continuous negotiation between belief in medical expertise and personal reflections on bodily integrity.

Transformation of Identity and Self-Perception

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Post-treatment, several participants described a process of identity reconfiguration. The therapy not only modified their physiology but also reshaped how they perceived themselves in relation to their illness. One participant articulated:

"For years, my disease defined me. Now I have to figure out who I am without it. It's strange… I'm healthy, but I still feel like a patient."

This theme highlights a central phenomenological insight: healing does not always equate to wholeness. The lived experience of genetic transformation extends beyond biological correction, touching dimensions of memory, self-recognition, and belonging. Some patients reported a period of psychological disorientation as they adjusted to a "new normal," emphasizing the need for psychosocial support after gene therapy.

Ethical Awareness and the Moral Weight of Participation

Many participants displayed an acute awareness of the ethical and social dimensions of their involvement in CRISPR-based clinical trials. They did not view themselves merely as recipients of care but as active contributors to the advancement of science. One participant noted:

"I joined this study because someone has to take the risk. If it helps the next generation, then it's worth it."

However, this altruistic stance often coexisted with moral tension particularly when participants reflected on the unknown consequences of genome editing. This ethical reflexivity underscores the dual identity of patients as both beneficiaries and pioneers in an evolving medical frontier.

Social Reintegration and Acceptance

After therapy, participants faced new challenges in redefining their social relationships. Family members oscillated between pride and apprehension, while some communities viewed gene therapy as "unnatural interference." As one participant described:

"My mother was proud, but my uncle said I was part of an experiment. It hurt, because I didn't feel like an experiment I felt like a survivor."

These accounts illustrate that the success of gene therapy is not solely biomedical; it also depends on social acceptance and narrative reconstruction within patients' cultural environments. The therapy thus becomes a social experience as much as a medical one.

DISCUSSION

The findings of this study reveal that patients who underwent CRISPR-Cas9–based gene therapy experienced a profound transformation in how they perceived their identity, body, and sense of hope (Mukhlis, Janwari, et al., 2023; Mukhlis & Abdullah, 2025). The essence of their lived experience centered on the coexistence of healing and uncertainty, illustrating that genetic correction is not merely a biological recovery but an existential redefinition of selfhood. These insights directly respond to the research question posed in the introduction What does it mean to be genetically healed from the patient's perspective? by showing that healing involves a renegotiation of identity and belonging rather than the mere restoration of physiological normalcy.

Contribution of Findings to the Research Question

The results provide a nuanced understanding of the meaning of genetic healing as lived and interpreted by patients. Rather than viewing CRISPR-based therapy as a purely medical success, participants described it as an intimate journey of becoming one that intertwined scientific innovation with deeply personal emotions of fear, gratitude, and moral reflection. The themes of hope, fear of the unknown, transformation of identity, and ethical awareness together depict a multidimensional process where patients oscillated between empowerment and vulnerability. These findings suggest that gene therapy constitutes a form of existential medicine where molecular intervention intersects with human meaning-making (Du Toit, 2025). By uncovering this interpretative depth, the study contributes a human-centered epistemology to molecular medicine, emphasizing that understanding therapeutic outcomes requires engaging with the subjective realities of those who experience them.

Relationship to Previous Literature and Theoretical Perspectives

The findings align with and extend prior phenomenological inquiries into medical transformation and identity reconstruction (Lee, 2023; Morales, 2024). Consistent with these studies, participants in the present research reported feelings of ontological instability a sense of being both cured and changed, which resonates with Merleau-Ponty's notion of embodiment as the intertwining of the physical and the existential. However, this study advances existing discourse by situating such transformations within the context of genomic alteration, a domain where identity is literally rewritten at the molecular level (Allagui & Ibahrine, 2025). The coexistence of hope and anxiety observed here echoes Doudna's (2021) reflections on the ethical ambivalence of CRISPR technologies, yet the phenomenological data enrich these debates by grounding them in lived human narratives rather than abstract ethical reasoning.

Furthermore, the findings challenge reductionist biomedical paradigms by demonstrating that subjective meaning cannot be inferred from clinical outcomes alone. The participants' narratives reveal that healing entails a dynamic interplay between biological correction and existential adaptation, extending beyond what traditional psychosocial or bioethical frameworks have previously articulated (Andras & Balázs, 2025). In this way, the study complements and deepens prior theoretical discussions by illuminating how patients reconstruct coherence and agency amid the profound disruption of genetic transformation.

Implications of the Findings

The findings of this study carry important social, cultural, and clinical implications for the evolving field of gene therapy (Xiang et al., 2025). On a social level, they highlight that patients' engagement with CRISPR-based interventions cannot be understood solely through biomedical or ethical lenses; rather, these experiences are embedded within broader narratives of identity, belonging, and human self-understanding. Culturally, the narratives reveal how genetic healing is interpreted through personal and collective meanings reflecting tensions between scientific progress and moral responsibility. Clinically, the insights underscore the need for integrative patient care models that address not only physiological outcomes but also the psychological and existential adaptation of individuals undergoing genetic modification. These findings invite healthcare professionals, ethicists, and policymakers to approach gene therapy as a deeply human experience that transforms both body and being.

Limitations of the Study

This study's interpretative phenomenological design, while offering rich and nuanced insights, inherently limits generalizability (Mukhlis, 2025a; Mukhlis & Saidah, 2025). The sample comprised a small number of participants with similar therapeutic experiences, which constrains the diversity of perspectives across different genetic conditions or cultural settings. Additionally, the data relied primarily on self-reported narratives, which may be shaped by recall bias or personal interpretation (Eren & Salgamcioglu, 2025). The research context centered around patients in controlled clinical environments may not fully reflect broader societal experiences of gene therapy, particularly in regions where access or perception of genetic technology differs (Landolfi, 2025). These limitations, however, are consistent with the phenomenological aim to achieve depth over breadth, prioritizing lived meaning rather than representativeness. Future studies may overcome these constraints by incorporating longitudinal or cross-cultural perspectives to explore evolving interpretations of genetic transformation.

Prospective Directions for Future Research

The interpretative insights generated by this study open several promising avenues for further investigation. Future research could explore how familial, religious, or sociocultural contexts influence patients' meaning-making processes during and after gene therapy (Kushwah et al., 2025).

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Comparative studies between recipients of different genetic interventions may also reveal variations in the construction of identity and moral perception across therapeutic modalities. Moreover, interdisciplinary collaborations between molecular scientists, phenomenologists, and clinical ethicists could deepen understanding of the psychosocial dynamics of biotechnological innovation, integrating narrative and ethical reflection into therapeutic design (Yang & Lee, 2025). Expanding this work into larger qualitative or mixed-method frameworks may further illuminate how gene therapy redefines the relationship between humanity, science, and the meaning of health in the twenty-first century.

CONCLUSION

This study explored the lived experiences of patients who underwent CRISPR-Cas9–based gene therapy for hereditary blood disorders, focusing on the meaning of being “genetically healed” from a phenomenological perspective. The findings revealed that healing extends beyond biological restoration, encompassing a deep existential transformation involving hope, uncertainty, identity reconstruction, and moral reflection. By illuminating these dimensions, the study addresses the existing gap in gene therapy research, which has often overlooked the subjective and experiential aspects of patients’ journeys. The interpretative phenomenological approach enabled a nuanced understanding of how individuals make sense of their altered genetic realities and integrate them into their broader life narratives. These insights provide a foundation for developing more compassionate, patient-centered practices in molecular medicine. Future studies could expand this exploration across different cultural and clinical contexts to examine how social and ethical environments further shape the meaning of genetic healing.

CONFLICT OF INTEREST

The authors declare no conflict of interest related to the design, execution, or publication of this research. The funding organization, Global Biomedical Research Consortium (GBRC), provided financial support for research implementation but had no role in data analysis, interpretation of results, or the preparation of the manuscript. All authors affirm that the study was conducted independently and adheres to the principles of scientific integrity and transparency.

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