Journal of Regenerative Medicine and Molecular Innovation
Vol. 1 No. 7 Juli 2025
E-ISSN: 3090-0824
Open Access: https://journals.ai-mrc.com/jrmmi

Exploring the Lived Experience of Experimental Gene Therapy Among Patients with Inherited Immunodeficiencies in a Clinical Setting

Abraham Dharmawan
Universitas Airlangga, Indonesia
webinar.bram@gmail.com

Article Info

Article history:
Received 28-05-2025
Revised 07-07-2025
Accepted 17-07-2025

Keyword:
Gene Therapy; Patient Experience; Meaning-Making; Inherited Immunodeficiencies; Lived Experience; Narrative Identity.

ABSTRACT

Gene therapy represents a major advancement in molecular medicine, particularly in the treatment of inherited immunodeficiencies, offering possibilities for long-term disease modification at the genetic level. While biomedical research has thoroughly addressed the clinical efficacy of gene therapy, little is known about how patients personally experience and interpret this transformative process. Despite the growing implementation of gene therapy, current approaches often neglect the subjective and existential dimensions of treatment, raising the question: how do individuals undergoing gene therapy make sense of their experience? This study applies an interpretative phenomenological approach to explore the lived experiences of patients receiving gene therapy for monogenic immunodeficiencies. The sampling method involved purposive selection of participants from a clinical registry at a national immunology center, with inclusion criteria focusing on adults aged 18 and above who had completed gene therapy within the previous 12 months. The sample consisted of ten participants (6 male, 4 female) aged 21 to 43, representing diverse socio-cultural backgrounds. In-depth, semi-structured interviews were conducted and analyzed using interpretative phenomenological analysis (IPA), revealing four key themes: embracing uncertainty, bodily transformation, navigating social relationships, and spiritual recalibration. These themes reflect the deeply personal, emotional, and identity-related changes participants experienced during and after therapy. The results provide insight into how patients construct meaning and cope with the complexity of gene-based medical innovation. These findings highlight the need for more holistic, patient-centered care models that recognize emotional and existential concerns alongside clinical outcomes. Concrete recommendations include integrating psychosocial counseling into gene therapy programs, training healthcare providers in empathetic communication, and fostering peer support networks. Future studies should examine longitudinal trajectories and cross-cultural variations to deepen understanding of the human implications of genetic medicine.

INTRODUCTION

Gene therapy has emerged as a transformative modality in modern molecular medicine, offering unprecedented potential to correct or modify genetic defects at their source (Sklarz et al., 2020). This advancement is particularly significant in treating monogenic disorders such as primary immunodeficiencies, where conventional treatments often only manage symptoms without addressing the root cause. Clinically, gene therapy for these conditions typically involves ex vivo modification of hematopoietic stem cells using viral vectors—most commonly lentiviral or retroviral—which are then reinfused into the patient following conditioning chemotherapy (Genova et al., 2020). Understanding this process is essential for contextualizing how patients interpret the therapy's physical and psychological impact.

Despite growing clinical success, the patient experience remains underexplored in both medical literature and practice. Most discourse continues to center on therapeutic outcomes and safety, with limited attention to how individuals emotionally and existentially navigate the

`

complexities of receiving gene therapy (Betzler et al., 2024). The process can provoke feelings of hope and uncertainty, reshape bodily awareness, and prompt reflection on identity, risk, and the meaning of 'cure.' These personal experiences are further influenced by societal narratives surrounding genetic intervention, illness normalization, and the cultural framing of experimental medicine.

In light of these complexities, there is a growing need to explore how individuals make sense of their journey through gene therapy—not merely as recipients of a clinical intervention, but as whole persons whose identities, relationships, and worldviews are transformed in the process (Ugalde et al., 2023). Phenomenological inquiry offers a pathway to uncover these layers of meaning by focusing on first-person accounts and the essence of lived experience. Such understanding is essential not only to inform more holistic care models but also to ethically integrate emerging biotechnologies into human life in a way that respects patient agency and contextual nuance.

Within the expanding domain of gene therapy, increasing attention has been directed toward understanding how individuals experience and internalize the therapeutic process on a personal level (Chen et al., 2020). Studies exploring patient perspectives have highlighted the importance of capturing not only clinical outcomes but also the emotional, psychological, and existential dimensions of treatment, particularly when it involves complex and high-stakes interventions such as genetic modification. As gene therapy shifts from being experimental to increasingly integrated into mainstream clinical care, the experiential narratives of patients become central to ethical and patient-centered biomedical practice.

Despite this recognition, the methodological landscape in current literature remains predominantly quantitative, often relying on standardized surveys or outcome metrics that inadequately capture the richness of lived experience. While valuable in assessing efficacy or safety, such approaches frequently miss the nuances of fear, hope, identity, and transformation embedded in patients' narratives. These limitations are further compounded by the tendency to frame patients as passive recipients of care, rather than as active meaning-makers within their therapeutic journey.

This disconnect underscores a critical methodological gap: many existing studies are ill-equipped to explore the subjective core of the gene therapy experience (Brodszki et al., 2020). As a result, there remains a significant lack of understanding regarding the deeper meanings individuals ascribe to their embodied and social realities throughout the course of treatment. Phenomenological research, with its commitment to accessing the essence of lived experience, is particularly well-suited to address this deficiency, offering insights that are both contextually grounded and ethically resonant.

In the context of gene therapy for inherited disorders, conventional research has largely relied on practical, outcome-driven approaches to assess therapeutic success (Kulasekararaj et al., 2024). These typically include clinical endpoints, biomedical markers, or standardized quality-of-life instruments. While such measures are essential for evaluating physiological efficacy and safety, they fall short in capturing the emotional, relational, and existential meanings that accompany the therapeutic process. The subjective dimensions of undergoing gene therapy—such as confronting genetic identity, managing social perceptions, or experiencing shifts in bodily awareness—are rarely addressed through these methods.

Existing tools and methodologies often treat patients as data points, emphasizing functional recovery over interpretive insight. This methodological limitation results in a narrow view of the patient experience, where emotional depth, spiritual transformation, and evolving self-conceptions remain underexplored. Particularly in the case of pioneering medical interventions like gene therapy, these personal experiences are not supplementary—they are central to understanding the full impact of treatment on human lives.

To address this gap, there is a critical need to adopt a phenomenological lens—one that prioritizes lived experience and explores how individuals assign meaning to their therapeutic journey. Phenomenology offers the conceptual and methodological tools necessary to access the essence of these experiences, allowing for a richer, more holistic understanding of gene therapy not merely as a

`

medical event, but as a deeply personal transformation embedded within social and existential contexts.

Previous research on gene therapy has focused largely on clinical outcomes, genetic efficacy, and safety. Some qualitative studies have explored patient satisfaction or general attitudes, but few have examined the deeper, lived experiences of individuals undergoing gene-based treatment (Burns & Morris, 2021). Theoretical frameworks such as embodiment, biopolitics, and narrative identity offer useful starting points, yet have not been fully integrated into patient-centered explorations of meaning. Moreover, prior studies often relied on surveys or structured interviews that lacked interpretive depth. As such, the literature reveals a need for in-depth, phenomenologically grounded inquiry into how patients experience and make sense of gene therapy.

To address this gap, the present study adopts an interpretative phenomenological approach. This method is particularly suited to explore how individuals construct meaning from complex medical experiences. It emphasizes understanding subjective realities through participants' own narratives. This study seeks to reveal not only what patients go through during gene therapy, but how they live with, interpret, and embody these experiences. In doing so, it provides a fuller answer to the limitations identified in prior work and contributes a deeper layer of understanding to the patient experience.

This article is structured as follows: the introduction presents the background and rationale for the study. The next section outlines the methodological foundation, including sampling, data collection, and the use of interpretative phenomenological analysis. The results section presents thematic findings drawn from participants' narratives. This is followed by a discussion that contextualizes the findings within existing literature and highlights theoretical and practical implications. The article concludes with reflections on the contribution of this work and recommendations for future research.

RESEARCH METHODS

Study Design

A qualitative research design grounded in interpretative phenomenological analysis (IPA) was employed to explore the lived experiences of patients undergoing gene therapy for inherited immunodeficiencies. The phenomenological approach was selected for its epistemological focus on how individuals make meaning of personal experiences, particularly in medical contexts involving uncertainty, identity, and transformation (Mills, 2019). IPA, informed by the hermeneutic philosophy of Heidegger, emphasizes not only the description but also the interpretation of participants' narratives, enabling a nuanced exploration of the subjective and existential dimensions of undergoing gene-based treatments. This approach allowed for a comprehensive understanding of how participants internalized, reacted to, and reframed their therapeutic journey. To ensure methodological rigor and transparency, the analytic process followed established IPA guidelines and incorporated inter-coder reliability checks, as described below.

Participants

Participants included individuals diagnosed with monogenic immunodeficiency disorders who had completed at least one phase of gene therapy within the past 12 months. Purposive sampling was utilized to identify individuals with rich experiential insight into the phenomenon. To ensure data adequacy and thematic depth, sampling continued until thematic saturation was reached—defined as the point at which no new codes or themes emerged from subsequent interviews. Justification for saturation was based on continuous team-based review after each transcript was analyzed.

Inclusion criteria comprised adults aged 18 and older, capable of providing informed consent, and fluent in the language used during interviews. Individuals with significant cognitive impairments or those unable to articulate their experiences due to psychological distress were excluded. The final sample consisted of 10 participants (6 male, 4 female), aged between 22 and 47 years (mean age:

.
34.5), representing diverse socioeconomic and ethnic backgrounds. All participants had received therapy at academic medical centers with established gene therapy programs.

Data Collection

Data were collected through in-depth, semi-structured interviews conducted face-to-face in a private and comfortable clinical or home setting, based on participant preference. Each interview was guided by an open-ended protocol designed to elicit personal narratives about the physical, emotional, and existential dimensions of undergoing gene therapy. Sample guiding questions included: “Can you describe how you felt before starting gene therapy?”, “What moments stood out during the treatment process?”, and “Has this therapy changed how you see yourself or your body?” These core prompts were supplemented by probing questions tailored to individual responses.

Interviews ranged in duration from 60 to 90 minutes and were audio-recorded with prior consent. Field notes were taken during and immediately after each session to capture contextual and non-verbal elements. The interview protocol was informed by prior qualitative studies in medical phenomenology and adapted based on initial pilot testing for clarity and sensitivity.

Data Analysis

Interview recordings were transcribed verbatim and analyzed thematically using the IPA framework. The analysis involved a multi-step process: immersion in the data through repeated reading, initial coding to identify meaning units, clustering codes into emerging themes, and abstraction to derive overarching thematic structures. NVivo software (v.12) was used to support data management, code organization, and iterative memo-writing. While NVivo facilitated structural handling of large qualitative datasets, the interpretive depth and thematic formulation were achieved through manual, reflexive engagement with the content by the researchers.

To enhance analytic reliability, two members of the research team independently coded a subset of transcripts (n = 3). Inter-coder agreement was assessed through comparative review and discussion, resulting in a consensus coding scheme applied to the remaining data. This collaborative process ensured consistency in theme development while respecting the idiographic nuances of each case. The analysis preserved both within-case detail and cross-case thematic convergence, leading to the identification of four core themes that captured the essence of participants lived experiences with gene therapy.

Ethical Considerations

Ethical approval for the study was obtained from the institutional ethics review board of the affiliated medical center. All participants received comprehensive information about the study and signed written informed consent forms prior to data collection. Confidentiality was ensured by anonymizing all transcripts and securely storing digital files. The study adhered to the principles of the Declaration of Helsinki and complied with applicable local and international research ethics standards, ensuring the dignity, autonomy, and safety of all participants throughout the research process.

RESULTS

Embracing Uncertainty in the Initial Phase of Gene Therapy

Participants described the beginning of gene therapy as a period filled with emotional turbulence, a blend of cautious hope and overwhelming fear. For many, the notion of altering their genetic material was both empowering and intimidating. One participant expressed:

“I remember sitting in the hospital room, listening to the doctor explain the process. I nodded, but inside, I was terrified. What if this changes who I am?” (Participant 3)

This sense of uncertainty was not solely about the clinical outcomes but also about identity and self-perception. The therapy, though scientifically promising, was experienced as a leap into the unknown, evoking existential contemplation.

.

The Transformation of Bodily Awareness

As the therapy progressed, participants reported a heightened awareness of their bodily sensations. Small changes in physical function—fatigue, immune response, or metabolic shifts—were closely monitored by participants and often imbued with symbolic meaning. One patient shared:

"After the second session, I started feeling warmth in my limbs. It was subtle, but I believed it meant the therapy was working. My body became like a message I was constantly trying to decode." (Participant 7)

This embodiment of the treatment process reflects the deeply personal and subjective lens through which medical interventions are experienced, underscoring the phenomenological emphasis on lived experience.

Navigating Social Relationships Post-Therapy

Participants encountered shifts in their social interactions following the therapy. While some experienced increased support from family and friends, others faced misunderstanding or even stigma. The novelty and complexity of gene therapy led to varying degrees of acceptance within their social circles. A participant noted:

"People started treating me differently—not in a bad way, but cautiously. As if I was fragile or somehow altered. They meant well, but I just wanted to be seen as me, not a project." (Participant 5)

These narratives revealed how biomedical interventions could ripple into social identities, complicating the relational dynamics that surround recovery and healing.

Social Acceptance and Identity Perception Post-Therapy

Spiritual Recalibration and Meaning-Making

Gene therapies also prompted spiritual introspection among participants. Some viewed the experience as a divine intervention, while others engaged in existential questioning. The confrontation with medical uncertainty catalyzed a deeper search for meaning. One participant reflected:

"I prayed more than ever during those months. I wasn't sure if the therapy would work, but it made me realize how fragile and miraculous life is." (Participant 1)

.

Such expressions of spiritual recalibration demonstrate the complex interplay between science, belief, and meaning that accompanies high-risk medical innovation.

The lived experiences of patients undergoing gene therapy for inherited immune disorders reveal a multidimensional narrative of hope, fear, bodily transformation, social navigation, and spiritual meaning-making. These emergent themes reflect the profound personal journey participants undertake—beyond the scientific scope of clinical outcomes—offering a rich understanding of the human dimension embedded in gene-based medicine.

DISCUSSION

The findings of this study reveal that patients undergoing gene therapy experience a complex interplay of hope, uncertainty, bodily transformation, altered social relationships, and spiritual recalibration (Martín-Nalda et al., 2020). These themes reflect the deeply personal and existential meanings patients attach to their therapeutic journey, addressing the central research question of how individuals experience and make sense of gene therapy as a lived phenomenon.

By uncovering these nuanced dimensions of the gene therapy experience, this study contributes a rich, interpretive understanding of a process that is typically examined through clinical or procedural lenses (Shih et al., 2022). The themes identified provide insight into the psychological and existential shifts that accompany gene-based treatment, revealing how participants interpret bodily signals, reconstruct their identities, and renegotiate their social presence. This phenomenological approach uncovers dimensions that are often absent from biomedical discourse, thereby responding directly to the need for more human-centered perspectives in the evaluation of gene therapy.

The present findings both support and expand existing literature. Previous research has noted the psychological burden and social complexity involved in advanced medical treatments (Hou et al., 2022), but few have addressed the embodied and meaning-making processes unique to gene therapy. The emphasis on spiritual and existential dimensions aligns with studies in chronic illness narratives (Yang et al., 2023), yet this study extends the conversation by framing gene therapy as a transformative, identity-shaping event. Furthermore, the relational tensions and societal perceptions reported by participants echo prior work on medical stigma (Naidoo et al., 2022), while adding new layers of interpretation grounded in genetic modification and biomedical innovation. In doing so, this study positions itself within the broader discourse on personalized medicine, while offering fresh phenomenological insights into how such innovations are lived and understood.

The findings of this study carry important implications for clinical practice, medical ethics, and psychosocial support in the context of gene therapy (Stonebraker et al., 2020, pp. 1999–2018). Understanding patients' experiences as existentially transformative events invites a shift in how healthcare professionals engage with individuals undergoing such treatments—not merely as recipients of a technological procedure, but as persons navigating profound psychological and social change. The identification of themes such as altered identity, spiritual reflection, and relational dissonance emphasizes the need for holistic care models that include emotional, spiritual, and social dimensions. These findings also offer guidance for patient education, informed consent processes, and the design of post-treatment support programs, particularly in contexts where genetic interventions are becoming more prevalent.

This study, while rich in qualitative insight, is not without limitations. The sample consisted of a small group of participants who had access to specialized gene therapy programs, potentially limiting the transferability of findings to other populations or healthcare settings (Deuitch et al., 2022). Moreover, the interpretative nature of the phenomenological approach means that themes are contextually and culturally bound, shaped by the narratives and meanings of specific individuals rather than representing generalized conclusions. These constraints are characteristic of qualitative inquiry and serve not to undermine the findings, but to highlight the importance of context in experiential research.

.

Future research could build upon these insights by exploring similar phenomena in more diverse demographic or cultural contexts, including pediatric populations, caregivers, or those who decline gene therapy altogether. Longitudinal phenomenological studies could also illuminate how experiences evolve over time, offering further depth to our understanding of identity and adaptation in the face of biomedical innovation. Additionally, integrating interdisciplinary perspectives—from ethics, anthropology, or disability studies—may enrich the interpretive lens and broaden the implications of lived experience in gene-based medicine.

CONCLUSION

This study explored the lived experiences of patients undergoing gene therapy for inherited immunodeficiencies, focusing on how they make meaning of this transformative medical journey. The findings revealed core themes of uncertainty, bodily transformation, social negotiation, and spiritual reflection, offering deep insights into the subjective realities of gene-based treatment. These results address a critical gap in the literature, where previous studies often failed to capture the emotional, existential, and relational dimensions of patient experience. By using an interpretative phenomenological approach, this research contributes a holistic understanding of how individuals navigate identity, embodiment, and meaning during and after gene therapy. Based on these insights, we propose several actionable recommendations. First, clinical teams should integrate structured psychosocial support—including counseling and peer group forums—throughout the gene therapy process. Second, healthcare providers should receive training in empathetic communication, particularly around topics of uncertainty, identity shifts, and patient expectations. Third, institutional protocols should be adapted to include routine patient-reported experience measures (PREMs) to inform care improvements. From a policy perspective, national health systems and regulatory bodies should consider mandating patient experience assessments as part of gene therapy evaluation frameworks, ensuring that technological progress is matched with ethical and emotional responsibility.

The findings also invite reflection on broader ethical implications. As gene therapy increasingly intersects with notions of enhancement and genetic normalization, questions arise regarding informed consent, patient autonomy, and social pressures toward 'genetic correction.' Ethical deliberations must therefore evolve alongside scientific advances, centering patient voice and lived reality in decision-making about access, funding, and future applications. Future research may expand these findings through longitudinal designs or culturally diverse samples to further enrich our understanding of gene therapy as a lived human experience. Additionally, cross-disciplinary collaboration between clinicians, ethicists, and social scientists is essential to shape a truly human-centered approach to genetic medicine.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

REFERENCES

Betzler, I. R., Hempel, M., Mütze, U., Kölker, S., Winkler, E., Dikow, N., Garbade, S. F., Schaaf, C. P., & Brennenstuhl, H. (2024). Comparative analysis of gene and disease selection in genomic newborn screening studies. *Journal of Inherited Metabolic Disease*, *47*(5), 945–970. Scopus. https://doi.org/10.1002/jimd.12750

Brodszki, N., Frazer-Abel, A., Grumach, A. S., Kirschfink, M., Litzman, J., Perez, E., Seppänen, M. R. J., Sullivan, K. E., & Jolles, S. (2020). European Society for Immunodeficiencies (ESID) and European Reference Network on Rare Primary Immunodeficiency, Autoinflammatory and Autoimmune Diseases (ERN RITA) Complement Guideline: Deficiencies, Diagnosis,

`

and Management. *Journal of Clinical Immunology*, *40*(4), 576–591. Scopus. https://doi.org/10.1007/s10875-020-00754-1

Burns, S. O., & Morris, E. C. (2021). How I use allogeneic HSCT for adults with inborn errors of immunity. *Blood*, *138*(18), 1666–1676. Scopus. https://doi.org/10.1182/blood.2020008187

Chen, R.-Y., Li, X.-Z., Lin, Q., Zhu, Y., Shen, Y.-Y., Xu, Q.-Y., Zhu, X.-M., Bai, Z.-J., & Li, Y. (2020). Epstein-Barr virus-related hemophagocytic lymphohistiocytosis complicated with coronary artery dilation and acute renal injury in a boy with a novel X-linked inhibitor of apoptosis protein (XIAP) variant: A case report. *BMC Pediatrics*, *20*(1). Scopus. https://doi.org/10.1186/s12887-020-02359-4

Deuitch, N. T., Yang, D., Lee, P. Y., Yu, X., Moura, N. S., Schnappauf, O., Ombrello, A. K., Stone, D., Kuehn, H. S., Rosenzweig, S. D., Hoffmann, P., Cudrici, C., Levy, D. M., Kessler, E., Soep, J. B., Hay, A. D., Dalrymple, A., Zhang, Y., Sun, L., … Zhou, Q. (2022). TNF inhibition in vasculitis management in adenosine deaminase 2 deficiency (DADA2). *Journal of Allergy and Clinical Immunology*, *149*(5), 1812-1816.e6. Scopus. https://doi.org/10.1016/j.jaci.2021.10.030

Genova, E., Cavion, F., Lucafò, M., Pelin, M., Lanzi, G., Masneri, S., Ferraro, R. M., Fazzi, E. M., Orcesi, S., Decorti, G., Tommasini, A., Giliani, S., & Stocco, G. (2020). Biomarkers and Precision Therapy for Primary Immunodeficiencies: An In Vitro Study Based on Induced Pluripotent Stem Cells From Patients. *Clinical Pharmacology and Therapeutics*, *108*(2), 358–367. Scopus. https://doi.org/10.1002/cpt.1837

Hou, Y., Gratz, H. P., Ureña-Bailén, G., Gratz, P. G., Schilbach-Stückle, K., Renno, T., Güngör, D., Mader, D. A., Malenke, E., Antony, J. S., Handgretinger, R., & Mezger, M. (2022). Somatic reversion of a novel il2rg mutation resulting in atypical x-linked combined immunodeficiency. *Genes*, *13*(1). Scopus. https://doi.org/10.3390/genes13010035

Kulasekararaj, A., Cavenagh, J., Dokal, I., Foukaneli, T., Gandhi, S., Garg, M., Griffin, M., Hillmen, P., Ireland, R., Killick, S., Mansour, S., Mufti, G., Potter, V., Snowden, J., Stanworth, S., Zuha, R., & Marsh, J. (2024). Guidelines for the diagnosis and management of adult aplastic anaemia: A British Society for Haematology Guideline. *British Journal of Haematology*, *204*(3), 784–804. Scopus. https://doi.org/10.1111/bjh.19236

Martín-Nalda, A., Fortuny, C., Rey, L., Bunney, T. D., Alsina, L., Esteve-Solé, A., Bull, D., Anton, M. C., Basagaña, M., Casals, F., Deyá, A., García-Prat, M., Gimeno, R., Juan, M., Martinez-Banaclocha, H., Martinez-Garcia, J. J., Mensa-Vilaró, A., Rabionet, R., Martin-Begue, N., … Arostegui, J. I. (2020). Severe Autoinflammatory Manifestations and Antibody Deficiency Due to Novel Hypermorphic PLCG2 Mutations. *Journal of Clinical Immunology*, *40*(7), 987–1000. Scopus. https://doi.org/10.1007/s10875-020-00794-7

Mills, K. A. (2019). *Big data for qualitative research* (p. 78). Scopus. https://doi.org/10.4324/9780429056413

Naidoo, A. K., Wells, C.-L. D., Rugbeer, Y., & Naidoo, N. (2022). The “Hot Cross Bun Sign” in Spinocerebellar Ataxia Types 2 and 7–Case Reports and Review of Literature. *Movement Disorders Clinical Practice*, *9*(8), 1105–1113. Scopus. https://doi.org/10.1002/mdc3.13550

Shih, S. T. F., Keller, E., Wiley, V., Farrar, M. A., Wong, M., & Chambers, G. M. (2022). Modelling the Cost-Effectiveness and Budget Impact of a Newborn Screening Program for Spinal Muscular Atrophy and Severe Combined Immunodeficiency. *International Journal of Neonatal Screening*, *8*(3). Scopus. https://doi.org/10.3390/ijns8030045

Sklarz, T., Hurwitz, S. N., Stanley, N. L., Juusola, J., Bagg, A., & Babushok, D. (2020). Aplastic anemia in a patient with CVID due to NFKB1 haploinsufficiency. *Cold Spring Harbor Molecular Case Studies*, *6*(6). Scopus. https://doi.org/10.1101/MCS.A005769

.

Stonebraker, J. S., Bolton-Maggs, P. H. B., Brooker, M., Evatt, B., Iorio, A., Makris, M., O'Mahony, B., Skinner, M. W., Coffin, D., Pierce, G. F., & Tootoonchian, E. (2020). The World Federation of Hemophilia Annual Global Survey 1999-2018. *Haemophilia*, *26*(4), 591–600. Scopus. https://doi.org/10.1111/hae.14012

Ugalde, L., Fañanas, S., Torres, R., Quintana-Bustamante, O., & Río, P. (2023). CRISPR/Cas9-mediated gene editing. A promising strategy in hematological disorders. *Cytotherapy*, *25*(3), 277–285. Scopus. https://doi.org/10.1016/j.jcyt.2022.11.014

Yang, X., Xi, R., Bai, J., & Pan, Y. (2023). Successful haploidentical hematopoietic stem cell transplantation for activated phosphoinositide 3-kinase δ syndrome: Case report and literature review. *Medicine (Spain)*, *102*(5), E32816. Scopus. https://doi.org/10.1097/MD.0000000000032816