Journal of Information Systems Engineering and Management
2025, 10(48s)
e-ISSN: 2468-4376
https://www.jisem-journal.com/

Research Article

Towards a National 2030 Strategy to Improve Community Awareness of Gene Therapies in Saudi Arabia

Salma Elhag, Maryam Alzein, Kholoud Owais, Shrooq Alsalmi
Associate Professor Faculty of Computing & IT King Abdulaziz University
Research Students Faculty of Computing & IT King Abdulaziz University
Research Students Faculty of Computing & IT King Abdulaziz University
Research Students Faculty of Computing & IT King Abdulaziz University

ARTICLE INFO

Received: 29 Dec 2024
Revised: 12 Feb 2025
Accepted: 27 Feb 2025

ABSTRACT

The study was carried out to demonstrate the level of gene therapy awareness within Saudi society. This would be in the context of the existing cultural and hereditary genetic disorders. The study was done through an online questionnaire in line with the knowledge, attitude, and practice model (KAP). The participants in the survey were from different areas, and the results obtained were analyzed descriptively. The researchers in addition to the SWOT and PESTEL strategic models and the Balanced Scorecard applied participated in the determination of the outcomes that could be linked to the defined targets of the study. The result of this research was the gap of knowledge that was evidently big, with 56% of the participants who are not aware of 'gene therapy', and the majority of them being the imposition of unreliable media as a source, while the other 68% was for the government implementing the initiatives if the information is accurate. The study promotes the internet-based educational programs that cover the genetics concepts and their applications on several issues, taking into account that it is caused by the absence of a physical example, the constraints of the distance of impact, and self-bias source of information.

Keywords: Gene Therapy, Genetic Medicine, Knowledge Gap, Healthcare Innovation, Health Education

INTRODUCTION

The prevalence of genetic diseases represents a significant contemporary healthcare challenge due to the substantial costs they impose on medical facilities and the long-term social and economic consequences. These diseases primarily arise from inherited mutations that disrupt cellular and tissue functions, with effects ranging from minor disorders to life-threatening conditions. Gene therapy stands as a major medical breakthrough which enables disease treatment through direct modification of faulty genes at their molecular origin [1].

Modern medicine has witnessed the emergence of gene therapy as a revolutionary treatment method which enables doctors to fix faulty genes at the molecular level to treat diseases [1].The innovative approach has created fresh possibilities for treating rare genetic disorders as well as other diseases that lacked effective treatment methods[2]-[1].

The achievement of gene therapy depends on multiple factors beyond scientific advancements. Healthcare systems require public acceptance and understanding to successfully implement such therapies. The prevalence of genetic diseases in Saudi Arabia makes this understanding essential. The local community's perception of gene therapy has become a vital research area because of its importance[3].

The social perception of gene therapy needs to be understood because it determines both present-day knowledge and future health communication approaches. The public acceptance of gene therapies will directly affect their implementation success in regions that have cultural or religious concerns about biomedical innovations. Health authorities need to take into account the social values which determine health practices in Saudi Arabia. The research highlights the need for targeted strategies which address ethical issues and provide education and cultural sensitivity to promote gene therapy awareness. The research helps expand knowledge about how Saudi Vision 2030 national vision frameworks can support health innovation.

BACKGROUND

Studies conducted in Saudi Arabia reveal elevated rates of genetic diseases which scientists link to traditional consanguineous marriage customs. The occurrence of rare diseases in the region primarily stems from recessive genetic mutations which are responsible for these conditions [3]. The inherited medical conditions continue to strain families and healthcare infrastructure which demands culturally sensitive scientific solutions for prevention and treatment.

Healthcare institutions together with academic bodies now implement exome and genome sequencing technology as advanced genetic diagnostic tools. These technological advancements enable better identification of genetic conditions in early stages which leads to enhanced clinical treatment approaches [3]. The hospitals and genetic centers in Saudi Arabia achieve better identification of inherited diseases that enable prompt medical interventions and reproductive guidance. Advanced medical technology has brought progress but the society remains poorly educated about these developments.

Public knowledge about genetic testing together with genetic counseling and gene therapy remains insufficient despite recent technological developments. The research shows that people understand genetic testing to some extent yet they remain uninformed about prevention methods and diagnostic processes and treatment choices [4]. The insufficient knowledge about medical innovations results in delayed diagnoses and incorrect health decisions and diminished medical innovation adoption. Public media together with schools have insufficient resources to teach genetic education which worsens the problem.

The Saudi Human Genome Program alongside other national initiatives demonstrate substantial progress in their field. Public education campaigns together with data privacy and advanced data analytics outreach programs need better enhancement according to [5]. The current level of community participation remains low because many Saudi citizens remain unaware of the benefits their families and descendants could receive through program participation.

The implementation of gene therapy remains impractical because of expensive costs alongside intricate manufacturing methods and restricted distribution networks. The existing healthcare frameworks face challenges to implement gene therapy because of these obstacles [6]-[2]-[7]. The implementation of gene therapy requires solutions for three critical logistical challenges which involve laboratory resources and skilled personnel and uniform treatment procedures. The assessment of local people's understanding about gene therapy will guide upcoming healthcare policies and create specific educational materials for the population needs.

RESEARCH QUESTIONS:

- RQ1: What is the level of public awareness about gene therapies?
- RQ2: What are the factors that influence the individuals perceptions and acceptance of these treatments?

LITERATURE REVIEW

A. Public Awareness and Perception in Saudi Arabia:

Studies conducted in Saudi Arabia highlight a moderate to limited awareness of genetic disorders and gene therapy among the public. As emphasized by these papers [3][4] that while many participants are aware of genetic diseases, they lack understanding of the importance of genetic counseling and premarital screening. Similarly, this study [5] found that despite positive attitudes toward the Saudi Human Genome Program, knowledge about data privacy and genetic technology remains poor. At the same time, these papers [8][9] further emphasize the societal and regional disparities in awareness, particularly concerning consanguinity and rare disorders. And study [10] reinforced the need for public education to enhance the understanding of DNA variants and their link to common diseases in Saudi Arabia.

B. Technological Advancements and AI in Gene Therapy Awareness:

Artificial intelligence and next-generation sequencing technologies are rapidly transforming the landscape of genetic diagnostics and public awareness. These studies [11][12] illustrated how WGS is improving clinical diagnostics, while study [13] explored machine learning's role in detecting rare disorders. Other papers, like [14][15], discussed how AI tools like Face2Gene and personalized diagnostic systems help bridge the awareness gap through better diagnostics and patientspecific insights. A study [16] emphasized AIs integration with big data and its role in improving accessibility and accuracy in healthcare communication and awareness.

C. Policy, Equity, and Access to Gene Therapies:

Several papers focus on policy-related challenges and the need for equitable access to gene therapy. This study [17] introduced a national multi-criteria framework to support decision- making. While [6]-[7] identified key barriers such as high costs, regulatory complexities, and limited insurance coverage. Also, [18][19] stressed the ethical and legal challenges in ensuring equitable access. As well as paper [20] highlighted India's experience and called for integrated government-industry-patient collaborations to improve accessibility.

D. Clinical Trials, Therapeutic Models, and Future Perspectives:

Research on clinical applications and therapeutic strategies underlines both the progress and limitations in gene therapy for rare diseases. These studies [1][21] presented successful gene therapy applications using viral vectors. Also, these studies [22][23][24] emphasized diagnostic efficiency and future integration into healthcare. Global studies like [25][26][27] explored diagnostic delays and under-recognition of rare diseases. But [28][29] argued for cost-effective models and national planning to scale gene therapy. At the same time, this paper [30] focused on drug repurposing as an accessible and affordable alternative.

TABLE LITERATURE REVIEW

Reference	Focus Area	Main Weaknesses	Key Findings
[3][8]-[10]	Diagnosis & Genetic Testing in Saudi Arabia	Limited sample diversity, focus on specific regions or disorders, and reliance on exome sequencing without broader genomic integration.	Demonstrated the utility of genetic testing (especially WES) in identifying mutations and the impact of consanguinity in Saudi populations.
[4]	Public Awareness & Perceptions	Regional bias and reliance on online self-reported data; limited generalizability to all Saudi regions.	High awareness of genetic diseases but limited understanding of genetic testing practices.
[17][19][20]	Policy & Framework Development	Lack of real-world application data; conceptual and expert-driven approaches without empirical validation.	Outlined initial frameworks for policy prioritization and multi-criteria decision-making in genetic therapy implementation.
[5][13][14][15]	AI & Data Sharing	Challenges in data quality, privacy concerns, and lack of standardized integration across platforms.	Positive attitudes toward AI and data sharing; studies showed improved diagnostic efficiency using AI models and machine learning.
[6]-[7][30]	Access & Affordability of Therapies	High cost of treatment, complex regulatory pathways, and limited reimbursement strategies.	Identified key systemic barriers and proposed conceptual models to improve patient access to gene therapies.

[1][22][23][28][29]	Clinical Applications of Gene Therapy	Limited data from clinical trials; focus often on general or theoretical effectiveness rather than long-term outcomes.	Evidence supports the curative potential of gene therapies for rare diseases and calls for further clinical research.
[21][16]	Newborn Screening & Early Detection	Ethical concerns, limited pilot implementation data, and infrastructural challenges.	Emphasized the promise of early diagnosis using genetic newborn screening (gNBS) and advanced predictive models.
[18]	Equity in Genetic Care	Barriers related to healthcare disparities, cost, and limited access to advanced technologies.	Highlighted inequities in access and stressed the need for inclusive and equitable therapeutic development.
[11]-[24][27]	Genomic Strategies & WGS	Resource limitations, lack of standardization, and translation of genomic data into clinical practice.	WGS and related strategies significantly improve diagnostic yield and support precision medicine approaches.
[?][26]	Global Rare Disease Challenges	Limited coordination across countries, insufficient funding, and fragmented data systems.	Called for global collaboration, unified registries, and better integration of rare disease policies and research frameworks.

RESEARCH FRAMEWORK

This research adopts the Knowledge, Attitude, and Practice (KAP) model as its conceptual framework to examine community awareness of gene therapies. The KAP framework is widely used in public health research [31] to assess what people know (knowledge), how they feel (attitudes), and how they behave (practices) toward a particular health issue. In the context of gene therapies, this model facilitates the identification of informational gaps, public perceptions, and behavioral patterns that may influence acceptance and engagement with such innovative treatments. The framework, as illustrated in figure 1, supports the development of targeted awareness interventions by linking knowledge levels with attitudinal trends and health-related behaviors. It also helps explore how sociodemographic factorssuch as age, education, and prior exposure to genetic conditionsmoderate each component of the KAP triad. By structuring the research around this model, the study aims to generate actionable insights to inform education campaigns, policy decisions, and future research on gene therapy acceptance.

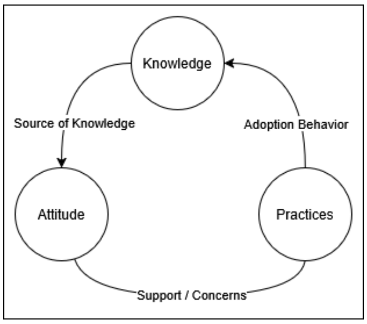


FIGURE 1 Conceptual framework based on the Knowledge, Attitude, and Practice (KAP) model.

METHODOLOGY

This study aims to explore the level of community awareness regarding gene therapies and identify key factors influencing public perceptions and understanding of these advanced treat- ments. In order to prepare for this research, We designed and followed the subsequent work phases shown in figure 2

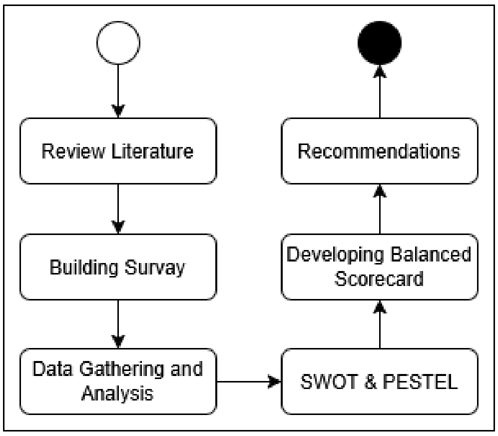


FIGURE 2 Methodology Flowchart

DISCUSSION

A. Measure

We built an online survey that consisted of 14 questions and focused on 7 questions only to evaluate the public awareness in the Saudi Arabian community toward gene therapy and de- fine the main challenges that affect its adoption. Additionally, we measure how supportive the public community is of the government Initiatives related to gene therapies. For the anal- ysis used in the survey, we analyzed the data with Excel and strategic analysis tools such as SWOT, PESTEL, and BSC.

B. Data Collection and Samples

The online survey was distributed among different regions in Saudi Arabia, where 113 diverse participants responded, including patients, caregivers, researchers, and interested indi- viduals from the general public. The most common age group that responded was from 25 to 35 years (58%), and the high- est percentage of responses came from the southern region (65.49%), which reflects that the responses were primarily from younger individuals in that area.

C. Data Analysis

As shown in figure 3, 56% of the participants did not have any familiarity with gene therapies, whereas 44% of them knew about them. This result shows there is a clear gap in awareness, which needs some supportive awareness programs spatially in non-medical fields.

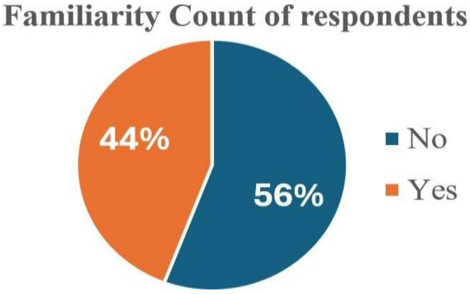
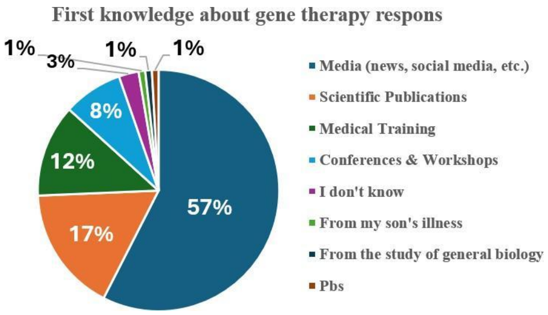


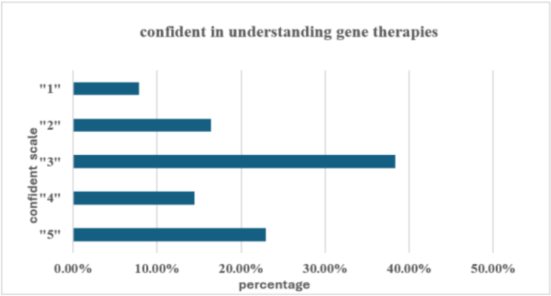
FIGURE 3 Familiarity with gene therapy

figure 4 shows that 57% of participants knowledge about gene therapy through the media, which is the highest percent- age, while 1% of people knowledge about these treatments through three less frequently used sources such as general bi- ology studies, a child's illness in one of the participants in the survey, and the Pbs (Public Broadcasting Service). This result shows that the Saudi Arabian community is based on unreliable and imprecise sources from a scientific view to get information.



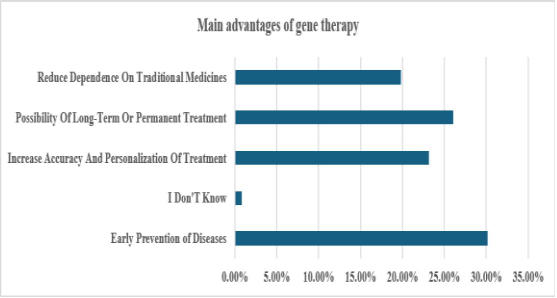
F I G U R E 4 Source of first knowledge

In figure 5 shows confidence levels in a range of 5 to 1 re- garding the understanding of gene therapy among participants. They can choose the 5 scale for high confidence and the 1 scale if they are not sure about their understanding of gene treat- ments. 38.36% refer to the medium confidence scale as 3 out of 5. While the highest confidence scale is only around 22.95%. This shows that most of the participants had a general idea but lacked dedicated knowledge.



F I G U R E 5 confident in understanding gene therapies

It is evident in figure 6 that the highest percentage of partic- ipants, approximately 30.17%, chose early disease prevention as the most prominent benefit of gene therapy, while others, approximately 26.03%, agreed that long-term permanent treat- ment was the most prominent expected benefit of gene therapy. This reflects that Saudi society has a general perception and understanding of the potential benefits, but there is still a need for greater awareness of the precise medical details.



F I G U R E 6 Main advantages of gene therapy

One of the gene therapies that concern participants is the safety and the side effects of using the gene therapy, as well as the ethical concerns, with around 28.5% expressing worries about the safety and side effects, and 21.1% about the ethi- cal concerns. This indicates that some anxiety exists due to a lack of information or misunderstanding about technology. As shown in figure 7

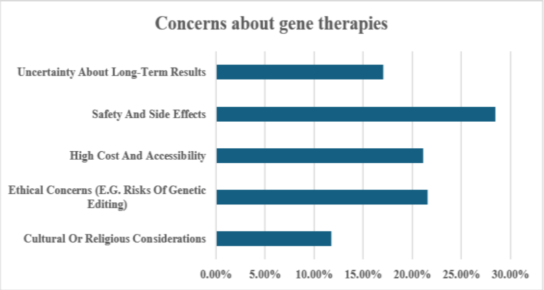


FIGURE 7 Concerns about gene therapies

Around 68.14% of the participants expressed strong support for funding research and scientific initiatives in the field of ge- netic therapy. This high level of public support highlighted the importance of including Saudi community in the futures poli- cies and encouraging investments in this sector. Where these results suggest a good public attitude toward advancing gene medicine in Saudi Arabia. As in figure 8.

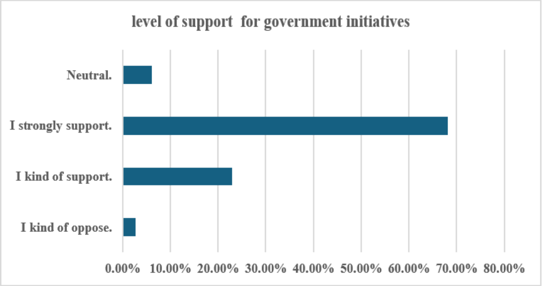


FIGURE 8 level of support for government initiatives

The result in figure 9 demonstrates that 28.3 % of the participants see public awareness as the most important procedure to support the adoption of gene therapy, as well as research studies and government initiatives. In contrast , around 37.1% do not have enough knowledge about what the effective procedures are , and this leads to a need for more education for the community.

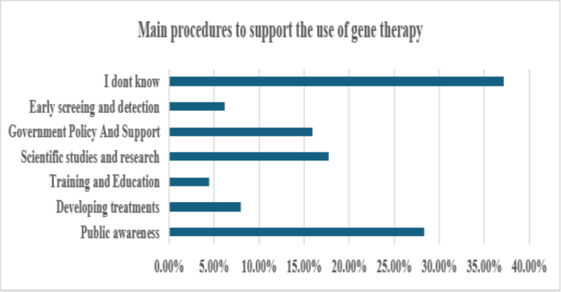
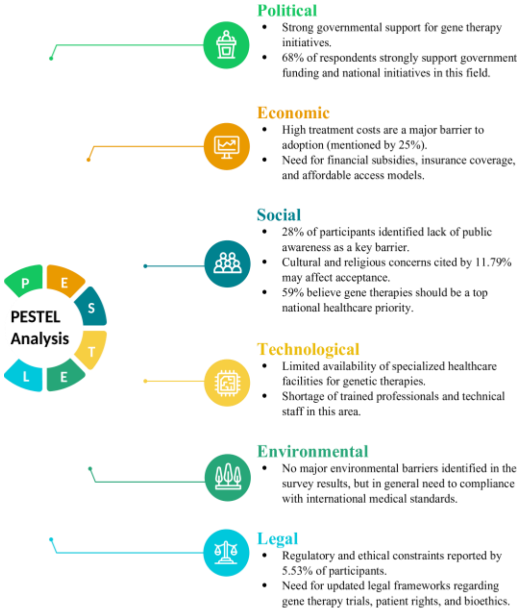


FIGURE 9 Main procedures to support the use of gene therapy

D. Contextual Analysis: PESTEL Framework

To better understand the external factors influencing public awareness and the adoption of gene therapies in Saudi Arabia, a PESTEL analysis was conducted based on the survey findings. This framework provides a structured assessment of the Political, Economic, Social, Technological, Environmental, and Legal factors that may affect the national implementation of genetic therapies for rare diseases. The insights gathered from the survey responses were mapped into each of the six dimensions to reflect the broader healthcare and policy landscape. The following figure 10 illustrates the key elements identified through this analysis.



Political
- Strong governmental support for gene therapy initiatives.
- 68% of respondents strongly support government funding and national initiatives in this field.

Economic
- High treatment costs are a major barrier to adoption (mentioned by 25%).
- Need for financial subsidies, insurance coverage, and affordable access models.

Social
- 28% of participants identified lack of public awareness as a key barrier.
- Cultural and religious concerns cited by 11.79% may affect acceptance.
- 59% believe gene therapies should be a top national healthcare priority.

Technological
- Limited availability of specialized healthcare facilities for genetic therapies.
- Shortage of trained professionals and technical staff in this area.

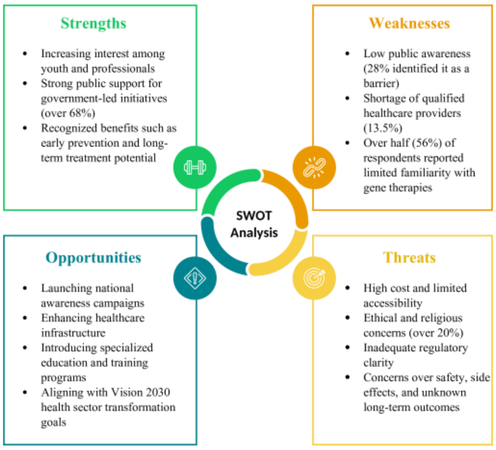
Environmental
- No major environmental barriers identified in the survey results, but in general need to compliance with international medical standards.

Legal
- Regulatory and ethical constraints reported by 5.53% of participants.
- Need for updated legal frameworks regarding gene therapy trials, patient rights, and bioethics.

F I G U R E 10 PESTEL Framework

E. Strategic Assessment: SWOT Analysis

In addition to the external environmental scan, a SWOT analysis was developed to evaluate the strategic position of genetic therapies in the Saudi healthcare context. This analysis highlights the internal strengths and weaknesses, as well as the external opportunities and threats associated with public awareness, policy readiness, and implementation capacity. The categories were derived directly from the survey data to ensure relevance and alignment with current perceptions, barriers, and expectations. The following figure 11 summarizes the key strategic factors identified.



Strengths
- Increasing interest among youth and professionals
- Strong public support for government-led initiatives (over 68%)
- Recognized benefits such as early prevention and long-term treatment potential

Weaknesses
- Low public awareness (28% identified it as a barrier)
- Shortage of qualified healthcare providers (13.5%)
- Over half (56%) of respondents reported limited familiarity with gene therapies

Opportunities
- Launching national awareness campaigns
- Enhancing healthcare infrastructure
- Introducing specialized education and training programs
- Aligning with Vision 2030 health sector transformation goals

Threats
- High cost and limited accessibility
- Ethical and religious concerns (over 20%)
- Inadequate regulatory clarity
- Concerns over safety, side effects, and unknown long-term outcomes

F I G U R E 11 SWOT Framework

F. Strategic Assessment: BSC Analysis

The Balanced Scorecard (BSC) approach was employed to organize the results of this study, which investigates the level of community awareness of gene therapy in Saudi Arabia. The survey responses were categorized into four main perspectives: Financial, Customer, Internal Processes, and Learning and Growth. Each perspective includes strategic objectives linked to key performance indicators (KPIs), which were either derived from the survey data or suggested for future measurement. Where applicable, actual values were drawn from the survey data. Target values were estimated based on logical assumptions to address any information gaps and enhance public health awareness. This is illustrated in the following table which presents the Balanced Scorecard summarizing the strategic objectives and key performance indicators.

Note: The symbol (-) indicates that there were no current data available for this indicator.

TABLE Balanced Scorecard (BSC)

Perspective	Objective	KPI	Target	Actual	Measurement Method / Unit
Financial	Support research funding	Public support for funding	≥ 80%	68.14%	Percentage of survey respondents supporting funding (%)
Customer	Increase awareness of gene therapy	Awareness of gene therapy	≥ 70%	44%	Percentage of participants correctly identifying the concept (%)
	Improve confidence	Confidence in understanding (15 scale)	≥ 4	3	Average score on 5-point Likert scale
	Use scientific sources	Scientific sources used	≥ 50%	~1%	Percentage of total sources cited that are scientific (%)
Internal Processes	Improve research methods	Research process efficiency (time per study)	≤ 6 months	8 months	Average duration per research cycle (in months)
	Increase collaboration	Number of interdisciplinary collaborations	≥ 10/year	5	Number of collaborations per year
Learning & Growth	Promote education	Student participation	≥ 500/year	–	Number of students enrolled in related activities

RESULTS

The survey results analysis demonstrates that the Saudi population increasingly understands the significance of gene therapies. The survey results demonstrate that public understanding about gene therapy remains restricted while most people learn about it through media channels instead of academic or clinical sources. The survey results demonstrate that people lack confidence in their ability to understand gene therapy concepts thus requiring immediate education and awareness programs. The research shows that people strongly back gene therapy research and government programs despite their limited understanding of the subject. The current situation offers policymakers an important chance to establish awareness programs which match community priorities.

CONCLUSIONS

The research has delivered important information about how the public of Saudi Arabia views gene therapy at present. The country's drive to modernize healthcare and treat genetic disorders has not eliminated the ongoing scientific knowledge deficit among the population. The majority of participants obtained their information from non-academic sources while showing restricted knowledge about gene-based medical treatments. The results show positive attitudes toward research and government involvement which indicates that better access to correct information could increase public acceptance. The research contains specific boundaries that need recognition. The online survey methodology excluded participants who were older than 65 years and those without consistent internet access. The study sample came from a specific location and its small size prevents researchers from drawing conclusions that would apply to the general population. Self-reported assessments of knowledge might contain biases because participants evaluate their own understanding. Future research needs to expand its participant base while implementing qualitative methods to analyze how cultural and social factors affect public opinions. The implementation of educational campaigns that unite healthcare providers with educators and media professionals will enhance public knowledge and acceptance of gene therapy. The implementation of genetic concepts in educational institutions along with strategic planning tools SWOT, PESTEL and Balanced Scorecard will help develop policies and engage communities.

REFERENCES

[1] Papaioannou, I., Owen, J. S., & Yáñez-Muñoz, R. J. (2023). Clinical applications of gene therapy for rare diseases: A review. *International Journal of Experimental Pathology*, *104*(4), 154–176. https://doi.org/10.1111/iep.12478

[2] Booth, C., & Aiuti, A. (2023). Realizing the Potential of Gene Therapies for Rare and Ultra-Rare Inherited Diseases. *Human Gene Therapy*, *34*(17-18), 776–781. https://doi.org/10.1089/hum.2023.127

[3] Alqahtani, A. S., Alotibi, R. S., Taghrid Aloraini, Fahad Almsned, Yara Alassali, Alfares, A., … Al, M. M. (2023). Prospect of genetic disorders in Saudi Arabia. *Frontiers in Genetics*, *14*. https://doi.org/10.3389/fgene.2023.1243518

[4] Al, M. M., Fahad Almsned, Alkharji, R. R., Aldossary, Y. M., Raghad AlQurashi, Hawsa, E. A., … Assiri, A. M. (2024). The perception of genetic diseases and premarital screening tests in the central region of Saudi Arabia. *BMC Public Health*, *24*(1). https://doi.org/10.1186/s12889-024-19029-0

[5] Alrefaei, A. F., Hawsawi, Y. M., Almaleki, D., Alafif, T., Alzahrani, F. A., & Bakhrebah, M. A. (2022). Genetic data sharing and artificial intelligence in the era of personalized medicine based on a cross-sectional analysis of the Saudi human genome program. *Scientific Reports*, *12*(1), 1405. https://doi.org/10.1038/s41598-022-05296-7

[6] Berry, D., Hickey, C., Kahlman, L., Long, J., Markus, C., & McCombs, C. K. (2025). Ensuring patient access to gene therapies for rare diseases: Navigating reimbursement and coverage challenges. *Molecular Therapy — Methods & Clinical Development*, *33*(1), 101403–101403. https://doi.org/10.1016/j.omtm.2024.101403

[7] Fox, T. A., & Booth, C. (2024). Improving access to gene therapy for rare diseases. *Disease Models & Mechanisms*, *17*(6), dmm050623. https://doi.org/10.1242/dmm.050623

[8] Abdallah, S., Sharifa, M., I.KH. ALMADHOUN, M. K., Khawar, M. M., Shaikh, U., Balabel, K. M., … Oyelaja, O. T. (2023). The Impact of Artificial Intelligence on Optimizing Diagnosis and Treatment Plans for Rare Genetic Disorders. *Cureus*, *15*(10). https://doi.org/10.7759/cureus.46860

[9] Abuzenadah, A., Alganmi, N., AlQurashi, R., Hawsa, E., AlOtibi, A., Hummadi, A., … Al Eissa, M. M. (2024). Familial Screening for the Prevention of Rare Diseases: A Focus on Lipodystrophy in Southern Saudi Arabia. *Journal of Epidemiology and Global Health*, *14*(1), 162–168. https://doi.org/10.1007/s44197-023-00182-5

[10] Adachi, T., El-Hattab, A. W., Jain, R., Nogales Crespo, K. A., Quirland Lazo, C. I., Scarpa, M., … Wattanasirichaigoon, D. (2023). Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges. *International Journal of Environmental Research and Public Health*, *20*(6), 4732. https://doi.org/10.3390/ijerph20064732

[11] Al-Jedai, A., Hajer Almudaiheem, Yazeed Alruthia, Abdullah Althemery, Alabdulkarim, H., Ojeil, R., … Zuhair Hasnan. (2024). A Step Toward the Development of the First National Multi-Criteria Decision Analysis Framework to Support Healthcare Decision Making in Saudi Arabia. *Value in Health Regional Issues*, *41*, 100–107. https://doi.org/10.1016/j.vhri.2023.12.005

[12] Aleissa, M., Aloraini, T., Alsubaie, L. F., Hassoun, M., Abdulrahman, G., Swaid, A., … Alfares, A. (2022). Common disease-associated gene variants in a Saudi Arabian population. *Annals of Saudi Medicine*, *42*(1), 29–35. https://doi.org/10.5144/0256-4947.2022.29

[13] Alzahrani, A. A., & Alharithi, F. S. (2024). Machine learning approaches for advanced detection of rare genetic disorders in whole-genome sequencing. *Alexandria Engineering Journal*, *106*, 582–593. https://doi.org/10.1016/j.aej.2024.08.056

[14] Brlek, P., Bulić, L., Bračić, M., Projić, P., Škaro, V., Shah, N., … Primorac, D. (2024). Implementing Whole Genome Sequencing (WGS) in Clinical Practice: Advantages, Challenges, and Future Perspectives. *Cells*, *13*(6), 504. https://doi.org/10.3390/cells13060504

[15] Chirmule, N., Feng, H., Cyril, E., Ghalsasi, V. V., & Choudhury, M. C. (2024). Orphan drug development: Challenges, regulation, and success stories. *Journal of Biosciences*, *49*(1). https://doi.org/10.1007/s12038-024-00425-y

[16] Gaviglio, A. M., Skinner, M. W., Lou, L. J., Finkel, R. S., Augustine, E. F., & Goldenberg, A. J. (2023). Gene-targeted therapies: Towards equitable development, diagnosis, and access. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. https://doi.org/10.1002/ajmg.c.32032

[17] Hurvitz, N., Azmanov, H., Kesler, A., & Ilan, Y. (2021). Establishing a second-generation artificial intelligence-based system for improving diagnosis, treatment, and monitoring of patients with rare diseases. *European Journal of Human Genetics*, *29*(10), 1485–1490. https://doi.org/10.1038/s41431-021-00928-4

[18] Lyroi, E., Olusegun, J., Amizu, T., & Tayo, O. (2025). *AI in Rare Disease Diagnosis: Solving the Puzzle of Complex Cases 6 PUBLICATIONS 0 CITATIONS SEE PROFILE AI in Rare Disease Diagnosis: Solving the Puzzle of Complex Cases*.

[19] Méndez-Vidal, C., Bravo-Gil, N., Pérez-Florido, J., Marcos-Luque, I., Fernández, R. M., José Luis Fernández-Rueda, … Antiñolo, G. (2025). A genomic strategy for precision medicine in rare diseases: integrating customized algorithms into clinical practice. *Journal of Translational Medicine*, *23*(1). https://doi.org/10.1186/s12967-025-06069-2

[20] Yousef, N. A., ElHarouni, A. A., Shaik, N. A., Babajan Banaganapalli, Ghamdi, A., Galal, A. H., … Ramu Elango. (2024). Nationwide survey on awareness of consanguinity and genetic diseases in Saudi Arabia: challenges and potential solutions to reduce the national healthcare burden. *Human Genomics*, *18*(1). https://doi.org/10.1186/s40246-024-00700-x

[21] Baldridge, D., Wangler, M. F., Bowman, A. N., Yamamoto, S., Schedl, T., Pak, S. C., … Westerfield, M. (2021). Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. *Orphanet Journal of Rare Diseases*, *16*(1). https://doi.org/10.1186/s13023-021-01839-9

[22] El Naofal, M., Ramaswamy, S., Alsarhan, A., Nugud, A., Sarfraz, F., Janbaz, H., … Rizk, R. (2023). The genomic landscape of rare disorders in the Middle East. *Genome Medicine*, *15*(1). https://doi.org/10.1186/s13073-023-01157-8

[23] Ferlini, A., Edith Sky Gross, Garnier, N., Berghout, J., Zygmunt, A., Singh, D., … Michela Zuccolo. (2023). Rare diseases' genetic newborn screening as the gateway to future genomic medicine: the Screen4Care EU-IMI project. *Orphanet Journal of Rare Diseases*, *18*(1). https://doi.org/10.1186/s13023-023-02916-x

[24] Groft, S. C., Posada, M., & Taruscio, D. (2021). Progress, challenges and global approaches to rare diseases. *Acta Paediatrica*, *110*(10). https://doi.org/10.1111/apa.15974

[25] Henderson, M. L., Zieba, J. K., Li, X., Campbell, D. B., Williams, M. R., Vogt, D. L., … Krueger, J. M. (2024). Gene Therapy for Genetic Syndromes: Understanding the Current State to Guide Future Care. *BioTech*, *13*(1), 1. https://doi.org/10.3390/biotech13010001

[26] Hwu, W.-L. (2024). Gene therapy for ultrarare diseases: a geneticist's perspective. *Journal of Biomedical Science*, *31*(1). https://doi.org/10.1186/s12929-024-01070-1

[27] Khare, V., & Cherqui, S. (2024). Targeted gene therapy for rare genetic kidney diseases. *Kidney International*, *106*(6), 1051–1061. https://doi.org/10.1016/j.kint.2024.07.034

[28] Knowles, J. K., Helbig, I., Metcalf, C. S., Lubbers, Laura S., Isom, L. L., Demarest, S., … Lowenstein, D. H. (2022). Precision medicine for genetic epilepsy on the horizon: Recent advances, present challenges, and suggestions for continued progress. *Epilepsia*, *63*(10), 2461–2475. https://doi.org/10.1111/epi.17332

[29] Roessler, H. I., Knoers, N. V. A. M., van Haelst, M. M., & van Haaften, G. (2021). Drug Repurposing for Rare Diseases. *Trends in Pharmacological Sciences*, *42*(4), 255–267. https://doi.org/10.1016/j.tips.2021.01.003

[30] Tisdale, A., Cutillo, C. M., Nathan, R., Russo, P., Laraway, B., Haendel, M., … Pariser, A. R. (2021). The IDeaS initiative: pilot study to assess the impact of rare diseases on patients and healthcare systems. *Orphanet Journal of Rare Diseases*, *16*(1). https://doi.org/10.1186/s13023-021-02061-3