Nufarcal Medical and Health Sciences

e-ISSN xxxx-xxxx (online)

Volume 1, Issue 1 2024
Page 1-10

https://doi.org/xxxxxx

Optimizing Healthcare Delivery for Patients with Rare Genetic Disorders in Low-Resource Settings

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ARTICLE INFO

Received: 06 Nov 2024
Revised: 20 Nov 2024
Accepted: 18 Dec 2024
Available online: 27 Dec 2024

Keywords:
Rare Genetic Disorders
Healthcare Delivery

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ABSTRACT

***Purpose**: This study examines the factors influencing healthcare delivery for patients with rare genetic disorders in low-resource settings, focusing on the availability of diagnostic tools, treatment options, specialized care, and healthcare provider experience.*

Subjects and Methods: Utilizing a quantitative research design, the study analyzed data collected from healthcare providers and patients in resource-constrained areas.

***Results:** The findings reveal that greater availability of diagnostic tools ($r = 0.55$), treatment options ($r = 0.47$), and specialized care ($r = 0.62$) are positively associated with improved patient outcomes. Healthcare provider experience also showed a positive correlation ($r = 0.36$) with patient outcomes, although weaker. Furthermore, barriers such as financial constraints ($\chi^2 = 14.23$, $p < 0.01$), lack of training and knowledge ($\chi^2 = 13.85$, $p < 0.01$), and cultural factors ($\chi^2 = 10.67$, $p < 0.05$) significantly affect healthcare delivery effectiveness. Regression analysis confirmed that the availability of diagnostic tools ($\beta = 0.28$), specialized care ($\beta = 0.31$), and healthcare provider experience ($\beta = 0.18$) are strong predictors of healthcare delivery effectiveness. The study addresses a gap in the literature by offering evidence on how resource availability and provider experience influence outcomes for rare genetic disorder patients in low-resource environments.*

***Conclusions:** These findings underscore the need for targeted interventions to improve healthcare infrastructure and provider training to enhance care for this vulnerable patient population.*

INTRODUCTION

Health care delivery in resource constrained environments is complex especially for individuals with rare genetic diseases. Genetic diseases, by their nature, can be considered as occurring in a small number of patients but the burden caused to these patients and their families can be really high Since these are chronic diseases and require medical attention for a long time, it has a great influence on genetic patients. These conditions are relatively uncommon but bear a significant share of costs, emotional distress and require lifelong monitoring. low-resource health care systems are limited with systemic failure, few resources, and insufficient health care infrastructure to support such patients adequately (Okpechi et al., 2021). Consequently, increasing healthcare efficiency for patients with rare genetic disorders in such settings does not just cherry-really means patients with rare genetic disorders are important for the improvement of patients' health and general health equity.

They include: significance, severity, unpredictability, complexity, and chronicity and when overwhelmed by these factors, management becomes even more arduous in low-resource

countries which struggles with financial hurdles, lack of medical facilities and more so, lack of specialists. Reports have pointed to the fact that these patients are still denied adequate care because of high cost of diagnosis, poor availability of treatment and lack of appropriate health care workers (Au et al., 2022). High-income nations' healthcare systems have improved to address these issues: the establishment of genetically focused centers and early diagnostics strategies, while low-resource settings persist in facing obstacles to effective healthcare provision.

One of the main challenges with regard to enhancing healthcare effectiveness for patients with rare genetic disorders in these contexts is a knowledge deficit among practitioners. According to Kruse et al. (2022), in most of the low resource settings, health care workers do not receive orientation on detecting or even diagnosing such rare genetic disorders and therefore diagnoses are delayed and treatment inadequate. In addition, these areas usually cannot afford diagnostic facilities including genetic testing laboratories which assist in diagnosing and confirming rare genetic disorders. These diagnostics challenges when compounded by the lack of specialized care can lead to missed diagnosis, inadequate management and overall worse outcomes for the patients (Hus & Segal, 2021).

A third challenge to healthcare delivery in low resource settings and especially for people with Rare genetic disorder is the availability of treatment. Some of the more uncommon genetic disorders bring lifelong responsibilities in terms of medical regimes, physiotherapies and other treatments including surgeries. These treatments are however not available to clients who come to health care facilities in low-resource setting due to either cost or lack of specialists to administer the interventions. enzyme replacement therapies for the treatment of certain genetic diseases such as Gaucher disease, are hard to come by and very costly, for most families within the low-income countries. These settings may also not be well equipped for infusing or offering sustained care and supportive and rehabilitative services which makes intervention attenuated in these aspects (Doucet et al., 2021).

Health care systems in low resource setting is another important system that dictates the delivery of care to individual with rare genetic disorder. There is a general lack of health facilities and human resource to address the difficulties of diagnosing and treating rare genetic disorders (Abdallah et al., 2023). There are common difficulties of deficit and maldistribution of health care workers, especially professionals with specialized knowledge of complicated cases of inherited diseases; general practitioners are often in short supply in the rural areas after being trained in the urban centres. there is a lack of well-developed HLIS, which hampers the identification of patient's outcome, assessment of the treatment's effectiveness, or integration of care from different healthcare actors.

It is important to note that patients with rare genetic disorders in low resourced communities that also experience social and cultural barriers that exacerbate their life challenges when affected by these diseases. Preconceptions associated with the genetic disorders lead to prejudice and prejudice, exclusion in society, and tend not to receive timely treatment and medical assistance. For a long time, the communities diagnosed with genetic disorders suffer from poor understandings or flank misrepresentations of their conditions as witchcraft or curses and in search for better cure they turn to other systems of medicine not the conventional medicine system. Major ethnic barriers contribute to delayed diagnosis and treatment, and furthermore an inability to follow health management regimens that were given to the patient to help improve their health situation.

There is increasing awareness, acknowledgment of the imperative to seek for new approaches to enhance the management of PTGIS in low-resource settings. One is the use of telemedicine since this enables patients in hard to reach or inareas where there are no specialists to get consultations from such specialists without actually having to physically go to the specialist. Telemedicine has the potential in maintaining access to healthcare services, decreasing transportation costs, enhancing patient satisfaction (Haleem et al., 2021). there has been collaboration between local healthcare facilities and the international organizations in enhancing healthcare delivery since

they enhance knowledge sharing, supply of needed resources, and development of capacity building programs.

There is another possible solution which identifies with community-based healthcare approaches for delivering care in such settings. These models include equipping CHWs with tools to offer community based basic health care services right from identifying, managing rare hereditary disorder. Models of this type have proved effective in increasing the availability of healthcare to rural and other hard-to-reach populations by using knowledge of the local context and facilities (Haleem et al., 2021). These strategies when used alongside public health education around genetic disorders can greatly enhance early diagnosis and management of individuals with RDs.

To meet the healthcare service demands in low health-care-resource-settings for RDs, there are a number of approaches that need to be taken into consideration. This includes not only investing in health human resource and expanding accessibility to the right diagnostic technologies and therapies but also addressing the philosophy and social determinants of care. There is great potential to improve the quality of care for patients with rare genetic disorders by designing policy strategies for the provision of appropriate services, mobilising communities, and implementing new approaches to delivering health care.

The study aim is to establish the extent to which available healthcare strategies can be implemented to address symptoms and support the management of heterozygous mutations affecting low-resource settings for rare genetic disorders and the challenges likely to be faced in the process. This way, it is expected that this research will make its input into ongoing efforts of enhancing healthcare for patients, particularly in LMICs and ensuring equal opportunities, regardless of patient's class, color, or disease un likeliness.

METHODOLOGY

This study aimed to evaluate the optimization of healthcare delivery for patients with rare genetic disorders in low-resource settings. A quantitative research design was employed to collect and analyze data on healthcare access, diagnostic processes, treatment availability, and patient outcomes. The study used a cross-sectional survey approach to gather data from healthcare providers, patients, and healthcare administrators in selected low-resource settings. The methodology involved several key steps, including participant selection, data collection, and statistical analysis.

Participant Selection

The participants for this study were selected from three low-resource settings, which were identified based on their limited healthcare infrastructure and prevalence of rare genetic disorders. These settings included urban and rural areas within these regions, ensuring a diverse representation of healthcare environments. The inclusion criteria for healthcare providers required them to be involved in the care or diagnosis of patients with rare genetic disorders. A total of 200 healthcare providers participated in the survey, which included general practitioners, specialists, and healthcare administrators. Additionally, 300 patients diagnosed with rare genetic disorders and their families were included in the study. The patient sample was selected based on the following inclusion criteria: patients diagnosed with any rare genetic disorder, patients who had been under medical care for at least six months, and patients who received care in the participating healthcare settings.

Data Collection

Data collection was carried out through a structured questionnaire distributed to both healthcare providers and patients. The healthcare provider survey gathered information on their training, experience with rare genetic disorders, access to diagnostic tools, availability of treatment options, and perceived challenges in managing rare diseases. The patient survey, on the other hand, focused on the patient's demographic information, access to healthcare, satisfaction with care, and the adequacy of treatment received. The surveys included both closed-ended questions,

which allowed for quantifiable data, and Likert-scale items, which measured perceptions and satisfaction levels related to healthcare delivery.

In addition to the surveys, secondary data were collected from health records to assess diagnostic timelines, treatment initiation, and patient outcomes. These records were reviewed to determine the average time taken for a diagnosis, the types of treatments prescribed, and whether patients were able to access specialized care or treatment regimens. Patient outcomes were measured by tracking improvements in health status, frequency of medical visits, and adherence to treatment protocols.

To ensure reliability and validity, pilot testing of the survey instruments was conducted with a small group of healthcare providers and patients prior to the main data collection. Feedback from the pilot test led to minor revisions in the wording of several survey items to improve clarity and understanding.

Variables

The primary variables in this study were: 1) Healthcare access: Measured by the availability of diagnostic tools, treatment options, and specialized care facilities; 2) Healthcare delivery effectiveness: Measured by the timeliness of diagnosis, patient satisfaction, and outcomes; Barriers to care: Measured by challenges reported by healthcare providers and patients, including financial barriers, lack of resources, and cultural factors.

Data Analysis

Data analysis was conducted using statistical software (SPSS version 26). Descriptive statistics were used to summarize the characteristics of the participants, including frequency distributions for categorical variables and measures of central tendency (mean, median) for continuous variables. The relationship between healthcare access, healthcare delivery effectiveness, and patient outcomes was examined using inferential statistics.

Correlation analysis was performed to examine the strength and direction of relationships between key variables such as healthcare access and patient outcomes. Additionally, a multiple regression analysis was conducted to identify the factors that most significantly influenced healthcare delivery effectiveness and patient outcomes. The regression model included variables such as the availability of specialized care, diagnostic tools, and patient satisfaction with care. Chi-square tests were also used to assess the association between categorical variables, such as barriers to care and the effectiveness of healthcare delivery.

The significance level for all statistical tests was set at $p < 0.05$, and all results were interpreted within the context of the study's limitations, such as the potential for response bias and the use of self-reported data.

RESULTS AND DISCUSSION

The results of the study have helped to address knowledge gaps of the crucial factors influencing the healthcare process of patients with rare genetic disorders in LMICs. Through consideration of aspects like access to diagnostic tools, curative interventions, specialisms, and provider expertise in such settings, it is the intent of this study to understand determinants of healthcare outcomes in those settings. The paper also reviews financial and experience constrains, inadequate training, and issues related to cultural background that affect the quality of care offered to the patients. In this section, we will elaborate on how the findings from the analysis add to the understanding of these factors, and how they affirm or defy some of the literature on health care delivery in developing countries.

Table 1. Demographic Characteristics of Healthcare Providers

Variable	Frequency	Percentage
Total Healthcare Providers	200	100%
Gender		
Male	120	60%
Female	80	40%
Professional Role		
General Practitioner	80	40%
Specialist	70	35%
Healthcare Administrator	50	25%
Years of Experience		
0-5 years	50	25%
6-10 years	70	35%
11+ years	80	40%

The healthcare provider sample is predominantly male (60%), with female providers making up 40%. The largest group of healthcare providers is general practitioners (40%), followed by specialists (35%) and healthcare administrators (25%). Most healthcare providers have significant experience in the field, with 40% having 11+ years, 35% having 6-10 years, and 25% having 0-5 years.

Table 2. Demographic Characteristics of Patients

Variable	Frequency	Percentage
Total Patients	300	100%
Gender		
Male	150	50%
Female	150	50%
Age Group		
0-10 years	100	33.3%
11-20 years	80	26.7%
21-40 years	60	20%
41+ years	60	20%
Type of Rare Genetic Disorder		
Metabolic Disorders	120	40%
Neurological Disorders	90	30%
Muscular Disorders	60	20%
Others	30	10%

The patient sample in this study was evenly split between males and females, each representing 50% of the total group. The majority of patients were from the younger age group, with 33.3% falling within the 0-10 years range, followed by 26.7% in the 11-20 years age range. Older age groups were less represented, with 20% of patients in both the 21-40 years and 41+ years age ranges. In terms of the types of rare genetic disorders, metabolic disorders were the most prevalent, affecting 40% of the sample, followed by neurological disorders at 30%, muscular disorders at 20%, and other types of rare genetic disorders making up the remaining 10%. This distribution highlights the diverse range of rare genetic disorders present in the sample and the predominance of metabolic and neurological conditions.

Table 3. Healthcare Access and Availability of Resources

Variable	Frequency	Percentage
Availability of Diagnostic Tools		
Fully Available	120	60%
Partially Available	60	30%
Not Available	20	10%

Availability of Treatment Options		
Fully Available	110	55%
Partially Available	80	40%
Not Available	10	5%
Specialized Care		
Available	140	70%
Not Available	60	30%

The availability of diagnostic tools, treatment options, and specialized care varied among healthcare providers. The majority of providers, 60%, reported that diagnostic tools were fully available, while 30% indicated partial availability, and 10% stated they had no access to these tools. A similar pattern was observed for treatment options, with 55% of providers claiming full availability, 40% stating partial availability, and 5% reporting no access to treatment options. In terms of specialized care, most providers (70%) had access to it, although 30% reported a lack of specialized care in their respective healthcare settings. These findings suggest that while key resources are accessible to many providers, there are still gaps in availability, especially for certain healthcare facilities in low-resource settings.

Table 4. Barriers to Healthcare Delivery as Reported by Healthcare Providers

Barrier Type	Frequency	Percentage
Financial Barriers		
Significant Barrier	100	50%
Moderate Barrier	60	30%
No Barrier	40	20%
Lack of Training/Knowledge		
Significant Barrier	120	60%
Moderate Barrier	40	20%
No Barrier	40	20%
Cultural Barriers		
Significant Barrier	80	40%
Moderate Barrier	60	30%
No Barrier	60	30%

Financial, training, and cultural barriers were identified as significant challenges to healthcare delivery. Half of the healthcare providers reported facing significant financial barriers, with 30% indicating moderate financial constraints and 20% stating that they encountered no financial barriers. The lack of training and knowledge emerged as the most significant barrier, with 60% of providers acknowledging it as a major obstacle to effective care. Additionally, cultural barriers were also a concern, as 40% of providers cited significant cultural challenges, 30% identified moderate barriers, and 30% reported no cultural barriers. These findings highlight the need to address financial constraints, improve training and knowledge, and overcome cultural challenges in order to optimize healthcare delivery for patients with rare genetic disorders.

Table 5. Healthcare Delivery Effectiveness (Provider Perception)

Variable	Mean Score (1-5)	Standard Deviation
Timeliness of Diagnosis	3.7	1.2
Patient Satisfaction with Care	3.9	1.0
Treatment Adherence	4.0	1.1
Overall Care Effectiveness	3.8	1.3

The study revealed varying perceptions regarding the effectiveness of healthcare delivery. In terms of timeliness of diagnosis, providers reported a moderate mean score of 3.7, with a relatively high standard deviation of 1.2, indicating significant variability in how quickly diagnoses were made across different healthcare settings. Patient satisfaction with care was generally high, with a mean score of 3.9 and a lower standard deviation of 1.0, suggesting consistent satisfaction levels among patients across different providers. Treatment adherence was also relatively strong, with a mean score of 4.0 and a standard deviation of 1.1, though some

variability in adherence was observed across the sample. Lastly, the overall care effectiveness was perceived positively, with a mean score of 3.8, but a higher standard deviation of 1.3 indicated that opinions on care effectiveness varied among healthcare providers. These findings highlight both strengths and areas for improvement in healthcare delivery for rare genetic disorders in low-resource settings.

Table 6. Patient Outcomes (Based on Health Record Review)

Variable	Frequency	Percentage
Improvement in Health Status		
Significant Improvement	120	40%
Moderate Improvement	100	33.3%
No Improvement	80	26.7%
Frequency of Medical Visits		
Less than once a month	60	20%
Once a month	120	40%
More than once a month	120	40%
Adherence to Treatment		
Full Adherence	150	50%
Partial Adherence	100	33.3%
No Adherence	50	16.7%

The patient outcomes indicated varying levels of improvement in health status. Approximately 40% of patients experienced significant improvement, 33.3% showed moderate improvement, and 26.7% showed no improvement in their condition. In terms of healthcare engagement, 40% of patients visited healthcare providers more than once a month, suggesting the need for regular monitoring and treatment, while 20% visited less frequently, less than once a month. Adherence to treatment was generally strong, with 50% of patients fully adhering to their prescribed treatment plans. However, 33.3% showed partial adherence, and 16.7% did not adhere to the treatment at all. These results highlight the importance of consistent healthcare visits and treatment adherence in improving patient outcomes for rare genetic disorders.

Table 7. Correlation Between Healthcare Access and Patient Outcomes

Variable	Correlation Coefficient (r)	p-value
Diagnostic Tools Availability & Patient Outcomes	0.55	< 0.01
Treatment Availability & Patient Outcomes	0.47	< 0.05
Specialized Care & Patient Outcomes	0.62	< 0.01
Healthcare Provider Experience & Patient Outcomes	0.36	< 0.05

The analysis revealed several important correlations between healthcare factors and patient outcomes. There was a moderate positive correlation between the availability of diagnostic tools and patient outcomes ($r = 0.55$), indicating that greater access to diagnostic tools is associated with better patient outcomes, with the result being statistically significant ($p < 0.01$). Similarly, treatment availability showed a moderate positive correlation ($r = 0.47$) with patient outcomes, suggesting that better access to treatment options is linked to improved health results, and this finding was also statistically significant ($p < 0.05$). Access to specialized care demonstrated the strongest positive correlation ($r = 0.62$) with patient outcomes, highlighting that patient with access to specialized care experienced significantly better outcomes, with the result being highly significant ($p < 0.01$). Lastly, the healthcare provider's experience showed a weaker positive correlation ($r = 0.36$) with patient outcomes, indicating that while provider experience may somewhat contribute to better patient outcomes, the relationship is less pronounced and statistically significant at the 0.05 level. These findings emphasize the importance of diagnostic tools, treatment options, and specialized care in improving patient outcomes, while provider experience plays a more moderate role.

Table 8. Multiple Regression Analysis for Predictors of Healthcare Delivery Effectiveness

Variable	Unstandardized Coefficient (B)	Standardized Coefficient (β)	t-value	p-value
Availability of Diagnostic Tools	0.25	0.28	3.45	< 0.01
Specialized Care Availability	0.35	0.31	4.05	< 0.01
Healthcare Provider Experience	0.15	0.18	2.32	< 0.05

The study revealed that the availability of diagnostic tools, access to specialized care, and healthcare provider experience significantly predict healthcare delivery effectiveness. A positive coefficient (B = 0.25) and a significant standardized coefficient (β = 0.28) indicated that greater availability of diagnostic tools positively predicted healthcare delivery effectiveness ($p < 0.01$). Similarly, access to specialized care showed a strong impact, with a positive coefficient (B = 0.35) and a significant standardized coefficient (β = 0.31), making it a significant predictor of healthcare delivery effectiveness ($p < 0.01$). Additionally, healthcare provider experience was found to be a contributing factor, with a positive coefficient (B = 0.15) and a significant standardized coefficient (β = 0.18), suggesting that more experienced providers are more likely to deliver effective care ($p < 0.05$). These findings underscore the importance of improving diagnostic tools, ensuring access to specialized care, and enhancing provider experience to optimize healthcare delivery for patients with rare genetic disorders.

Table 9. Chi-Square Test for Association Between Barriers to Care and Healthcare Delivery Effectiveness

Barrier	Effective Healthcare Delivery (Yes)	Ineffective Healthcare Delivery (No)	Chi-Square Value	p-value
Financial Barriers	70% (140)	30% (60)	14.23	< 0.01
Lack of Training/Knowledge	60% (120)	40% (80)	13.85	< 0.01
Cultural Barriers	55% (110)	45% (90)	10.67	< 0.05

The study found that financial barriers, lack of training/knowledge, and cultural barriers all significantly impact healthcare delivery effectiveness. A significant chi-square value of 14.23 ($p < 0.01$) indicated a strong association between financial barriers and ineffective healthcare delivery, highlighting the critical role of financial constraints in healthcare access and quality. Similarly, a chi-square value of 13.85 ($p < 0.01$) for lack of training/knowledge suggested a strong link between insufficient provider training and ineffective healthcare delivery. Cultural barriers, while still significant, showed a weaker association, with a moderate chi-square value of 10.67 ($p < 0.05$), indicating that cultural challenges do influence healthcare effectiveness, but not as strongly as financial and training-related issues. These findings emphasize the need to address these barriers to improve healthcare delivery for patients with rare genetic disorders.

This study aimed to examine key factors influencing healthcare delivery for patients with rare genetic disorders in low-resource settings, focusing on diagnostic tools, treatment availability, specialized care, and healthcare provider experience. It also addressed the significant barriers such as financial constraints, lack of training, and cultural challenges. The findings offer a comprehensive understanding of healthcare effectiveness and provide valuable insights into the improvements needed in these settings. By addressing critical gaps in the literature, this study contributes to the growing body of research on healthcare delivery for patients with rare genetic disorders, particularly in low-resource environments.

The availability of diagnostic tools and its positive correlation with better patient outcomes ($r = 0.55$, $p < 0.01$) confirms the pivotal role of accurate and timely diagnostics in the treatment of rare genetic disorders. This finding is consistent with previous studies, such as those by Nadeau

et al. (2020) and Endo et al. (2021), which highlighted that early diagnosis through accessible and reliable diagnostic tools significantly improves treatment outcomes for patients with rare diseases. the relationship between treatment availability and patient outcomes ($r = 0.47$, $p < 0.05$) further emphasizes the importance of ensuring treatment options are accessible in low-resource settings. These results align with studies by Stranneheim et al. (2021), who found that the availability of appropriate treatments positively influences the health outcomes of rare disorder patients, even in settings with limited resources.

Specialized care emerged as the strongest predictor of healthcare delivery effectiveness, with a correlation of $r = 0.62$ ($p < 0.01$). This finding is in line with previous research, such as by Domaradzki & Walkowiak (2021), who argued that specialized care is critical in managing rare genetic disorders. Access to specialists with expertise in these rare conditions can improve diagnosis, treatment planning, and patient support, leading to better health outcomes. The positive coefficient for specialized care availability ($B = 0.35$, $\beta = 0.31$, $p < 0.01$) further reinforces its significance as a key factor in healthcare delivery effectiveness. These findings support the importance of establishing specialized care networks in low-resource settings, a gap that remains under-addressed in many parts of the world, as noted by Van et al. (2021).

While healthcare provider experience was found to be a moderate predictor of healthcare delivery effectiveness ($B = 0.15$, $\beta = 0.18$, $p < 0.05$), it is important to note that its influence was less pronounced compared to diagnostic tools, treatment, and specialized care. have shown that experienced healthcare providers tend to deliver more effective care; however, this study reveals that other factors, such as the availability of specialized care and diagnostic tools, may play more substantial roles in low-resource settings. This finding addresses a gap in the literature by highlighting that the quality of care provided in these settings cannot rely solely on the experience of healthcare providers but also requires systemic support in terms of resources and training.

Regarding barriers to effective healthcare delivery, this study found significant correlations between financial barriers (chi-square = 14.23, $p < 0.01$), lack of training/knowledge (chi-square = 13.85, $p < 0.01$), and cultural barriers (chi-square = 10.67, $p < 0.05$). These findings are consistent with prior research by Abu-Odah et al. (2020), who identified financial limitations and inadequate training as major obstacles to providing effective healthcare for patients with rare genetic disorders. The impact of financial barriers is particularly relevant in low-resource settings, where limited budgets often hinder access to necessary diagnostic and treatment tools. Similarly, the lack of training or knowledge among healthcare providers is a well-documented issue in these settings, emphasizing the need for comprehensive education and training to improve healthcare outcomes. Cultural barriers, while influential, were found to have a weaker association with healthcare delivery effectiveness compared to financial and training-related barriers, echoing the findings of Alderwick et al. (2021), who suggested that cultural factors play a role in healthcare delivery but often have less impact than systemic issues like financial constraints and lack of provider training.

This study provides a novel contribution by emphasizing the significant role of specialized care in improving healthcare delivery for rare genetic disorders, a factor that has often been underexplored in the existing literature. The strong correlation between specialized care availability and patient outcomes ($r = 0.62$, $p < 0.01$) underscores the need for targeted interventions that establish specialized care pathways in low-resource settings. by identifying both financial and knowledge-related barriers as significant factors influencing healthcare delivery, this study highlights the need for multifaceted approaches that address these barriers in tandem.

CONCLUSION

This study highlights the critical factors influencing healthcare delivery for patients with rare genetic disorders in low-resource settings, emphasizing the importance of accessible diagnostic tools, treatment options, specialized care, and healthcare provider experience. The findings indicate that greater availability of diagnostic tools and specialized care, alongside experienced providers, significantly improve patient outcomes. Additionally, financial, training, and cultural barriers were identified as substantial obstacles to effective healthcare delivery, requiring

multifaceted interventions to address these challenges. By addressing these gaps, this study contributes valuable insights to the existing literature and provides a foundation for policy changes and resource allocation to improve healthcare outcomes for patients with rare genetic disorders in resource-constrained environments.

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