Challenges And Solutions in Rare Disease Treatment: Value-Based Healthcare

Zixu Tao *

San New School, Wuhan, China

* Corresponding Author Email: huangya@ldy.edu.rs

Abstract. This study is based on the realities of challenges within the field of rare disease treatment, as well as the concept of value-based healthcare. By analyzing treatment plans, rehabilitative care, cost-sharing, prospects for drug development markets and investment returns, as well as policy support, the paper delves into the complexities and diversities of rare disease treatment. Within the research, the focus is on the challenges of rare disease treatment, leveraging the principles of value-based healthcare. It discusses how personalized treatment plans, interdisciplinary collaboration, and reasonable cost-sharing can enhance patient treatment outcomes and quality of life. The article employs methods such as literature reviews and case analyses to deeply explore and analyze rare disease treatment from various angles. The research findings indicate that personalized treatment plans and interdisciplinary collaboration can significantly enhance treatment outcomes and quality of life for rare disease patients. Reasonable cost-sharing models and trends in drug development markets present opportunities for the sustainable growth of the rare disease field. Additionally, policy support and international cooperation mechanisms play vital roles in advancing rare disease treatment. Based on these research findings, the article proposes future research prospects, including strengthening data collection and analysis, deepening international collaboration, and optimizing drug development strategies.

Keywords: Rare disease; value-based healthcare; cost-sharing.

1. Introduction

Rare diseases, collectively referred to as orphan diseases, are a category of diseases with extremely low prevalence and a small total number of affected individuals on a global scale. They are characterized by their diverse types and complex clinical presentations. Despite the low number of patients with each individual rare disease, as a group, rare diseases constitute a significant proportion of the global population. Due to their rarity, diagnosis, treatment, and access to medications for rare diseases are often limited, resulting in a range of challenges faced by patients and their families.

Currently, rare diseases are a prominent concern in both developed and developing countries. In developed nations, the advantage of advanced medical capabilities has led to more comprehensive systems for diagnosing and treating rare diseases, allowing patients to receive accurate diagnoses and personalized treatments at an earlier stage. However, in developing countries, including countries like China, limited medical resources and relatively lower awareness of rare diseases often lead to cases of misdiagnosis or delayed treatment. While countries like China have started addressing rare diseases later and with less comprehensive strategies, they are gradually following the footsteps of developed nations to improve their approach. This divergence is due to the varying measures taken by different countries and the progress made in their healthcare systems.

Different countries have adopted diverse strategies to tackle the issue of rare diseases. In Europe and the Americas, some countries have established national rare disease registries and diagnostic networks that utilize big data and internet connectivity to gather and share data and treatment experiences of rare disease patients. This contributes to enhancing the efficiency of rare disease diagnosis and treatment quality. Additionally, some nations have introduced policies to encourage large pharmaceutical companies to invest in rare disease research and development. In China, individuals initially took spontaneous actions to connect with more rare disease patients through online platforms or by starting small enterprises. In recent years, the government has gradually increased its support for the rare disease field, promoting research and treatment advancements.

This study aims to explore the treatment challenges in the field of rare diseases and seek potential solutions by integrating the concept of value-based healthcare. This study will analyze the experiences and strategies of different countries in rare disease treatment and explore the feasibility of implementing these strategies in China. Furthermore, the paper will study the application of value-based healthcare to enhance diagnosis and treatment outcomes and improve the quality of life for rare disease patients. Through in-depth research, the paper may provide valuable insights to enhance the treatment and quality of life for rare disease patients and offer support for policymakers to formulate more effective policies and measures.

2. Literature Review

Over the past few decades, significant breakthroughs have been made in the diagnosis and treatment of rare diseases, including innovations in technology, drugs, and therapies. These advancements have not only provided patients with more treatment options but have also brought new opportunities and challenges to the healthcare market.

2.1. Medical Technological Innovation and Development

Medical technological innovation in the field of rare diseases is crucial for improving patients' quality of life and offering more effective treatment solutions. With the development of molecular biology, genomics, and bioinformatics, the ability to diagnose rare diseases has greatly improved. The application of high-throughput sequencing technology allows for more accurate detection and interpretation of genetic variations, accelerating the diagnostic process for rare diseases. Additionally, advances in imaging technology provide clinicians with more information about rare disease conditions, aiding in the formulation of more accurate treatment plans. Jones et al. studied the application of gene editing technology in the treatment of rare diseases [1]. Through literature reviews and experimental research, they found that gene editing technologies like CRISPR-Cas9 have significant potential in rectifying genetic mutations in patients. This personalized treatment approach holds promise for providing patients with more precise and tailored treatment plans, though it also faces ethical and safety considerations. Smith and Garcia explored the application of virtual reality technology in the rehabilitation treatment of rare disease patients [2]. Through systematic reviews and experimental research, they discovered that virtual reality technology can enhance patients' rehabilitation efficiency and motivation by creating more engaging and enjoyable rehabilitation environments, thus stimulating patients' enthusiasm.

2.2. Drug Development and Innovation

Remarkable progress has been achieved in the research and development of drugs for rare diseases. In the past, due to the small market size, there were relatively fewer developments in rare disease drug research. However, with advancements in medical technology and increased scientific understanding, more pharmaceutical companies are conducting research on drugs related to rare diseases. The development of these drugs often requires interdisciplinary collaboration and innovative approaches. For instance, the introduction of new technologies such as gene therapy and protein replacement therapy offer new avenues for treating rare diseases. Rare disease drug development has been a challenging field but has also gained more attention. Brown et al. conducted an in-depth analysis of the orphan drug market, finding that with advancements in medical technology, more pharmaceutical companies are focusing on the field of rare diseases [3]. By studying market data and policy support across different countries, they concluded that increased investment by pharmaceutical companies in orphan drugs provides more treatment options for rare disease patients. Robinson and Lee focused on the application of drug repurposing strategies in rare disease treatment [4]. Through comprehensive analysis of existing research, they found that drug repurposing can expedite the process of new drug development since drugs that have already passed clinical trials can

be quickly applied to new rare disease treatments. This strategy provides a faster treatment pathway for rare disease patients, shortening the time to market.

2.3. Policy Support and Regulatory Environment

Policy support and the regulatory environment play a crucial role in the development of rare disease treatments. Mullins et al. researched drug approval policies in different countries and compared policies across multiple nations [5]. They concluded that some countries expedite the approval process for orphan drugs through fast-track procedures, thereby increasing the speed at which patients can access treatments. This policy support helps accelerate the treatment process for rare disease patients. In China, the rare disease patient population is significant, and ordinarily, it would take a long time for rare disease drugs to be approved after undergoing clinical trials. This means that some rare disease patients in China might face situations where there are no available treatments. To address this, China has introduced innovative measures. Hainan, for instance, has undergone reform in the national drug and medical device approval system, allowing for accelerated accessibility of innovative products globally for clinical use in China. This is achieved through the establishment of medical special zones, such as the Hainan Boao Lecheng International Medical Tourism Pilot Zone. In these special zones, early access to rare disease drugs is no longer out of reach. For example, the Boao Winhealth Rare Disease Medical Center Rare Disease Clinical Medical Center has improved accessibility to rare disease drugs through multi-party collaboration. Through various channels, hospitals can access complex drug treatments earlier and ensure that patients receive treatment sooner. Smith and Johnson researched the impact of healthcare insurance policies on rare disease patients [6]. Through empirical analysis and survey research, they found that improving insurance reimbursement policies can reduce patients' financial burden, thereby promoting the implementation of treatments. Reasonable policy adjustments enable more patients to access appropriate treatments, improving the quality of life for rare disease patients.

3. Analytical Framework

When it comes to analysis in the healthcare field, a value-based approach is a powerful tool that provides profound insights into the evaluation and decision-making of various medical treatment strategies. The core of this analytical framework lies in its ability to comprehensively consider multiple key factors, thereby assessing the impact and feasibility of a treatment plan more comprehensively. Value-based analysis aims to elevate medical decisions from simple cost-effectiveness comparisons to a higher level, including factors such as patient preferences, the natural history of diseases, and societal value. In the following discussion, this study will explore how to apply the value-based healthcare analytical framework to analyze key areas such as treatment plans, rehabilitative care, cost-sharing, prospects and returns on investment in drug development markets, and policy support.

3.1. Treatment Plans: Personalization and Effectiveness Assessment

In the context of treatment plans for rare diseases, the importance of personalized treatment and effectiveness assessment is becoming increasingly evident. As the paper gains a deeper understanding of genomics and pathophysiology, the traditional "one-size-fits-all" approach is no longer applicable to treating rare diseases. Personalized treatment becomes crucial, necessitating the consideration not only of patients' genomic information but also the integration of clinical indicators, lifestyle factors, and other individual characteristics to formulate the most suitable treatment plan. Through genetic testing and molecular diagnostic techniques, physicians can more accurately predict patient responses to specific treatments, avoiding unnecessary trial and error and saving time and resources. This personalized approach contributes to enhancing treatment effectiveness and quality of life for patients while reducing waste within the healthcare system.

Simultaneously, the scientific and objective evaluation of treatment outcomes is paramount. The introduction of clinical trials, quality of life assessments, and biomarkers allows for a comprehensive evaluation of treatment effectiveness. This helps physicians and patients better understand the long-term effects of treatment, whether adjustments to the plan are needed, and whether the treatment conducted at various stages has achieved the intended results. Moreover, the objective assessment of treatment outcomes provides a foundation for policymakers to formulate more rational medical policies and resource allocation strategies. In the future, with the development of technologies such as artificial intelligence and big data, treatment outcomes can be predicted more accurately, further enhancing the success rate of personalized treatment.

3.2. Rehabilitative Care: Interdisciplinary Collaboration and Technological Innovation

In the realm of rehabilitative care, interdisciplinary collaboration and technological innovation are increasingly pivotal in improving patient rehabilitation outcomes and enhancing the quality and efficiency of the rehabilitation process. As shown in Figure 1, rare disease patients often require the collaboration of specialists from multiple fields to ensure a comprehensive and targeted treatment plan. Interdisciplinary collaboration fosters information sharing and teamwork, making the rehabilitation process more precise and efficient. The collaboration of interdisciplinary teams, including doctors, physical therapists, and psychologists, allows for the comprehensive consideration of patients' physiological and psychological traits, leading to the formulation of more tailored rehabilitation plans.

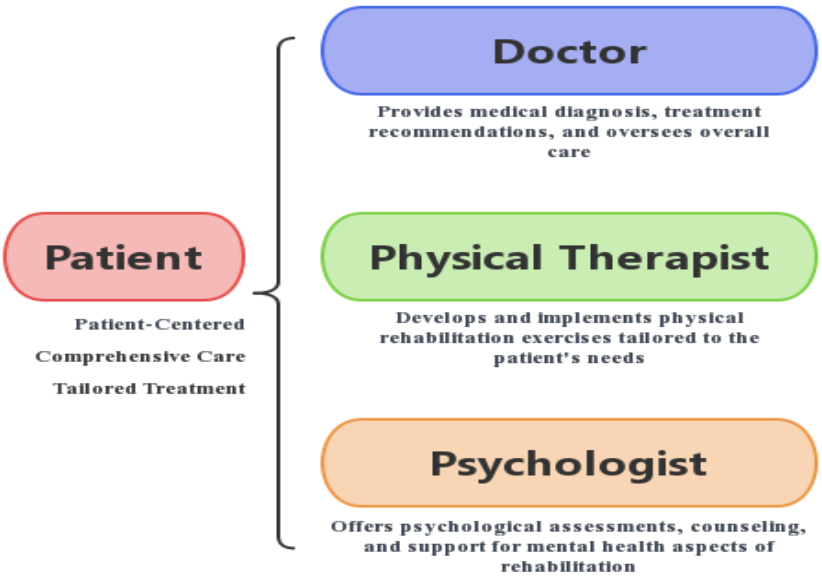


Fig. 1 Interdisciplinary collaboration in rare disease rehabilitation [7]

On the other hand, technological innovation opens up more possibilities for rehabilitative care. For instance, virtual reality technology can create diverse rehabilitation environments, promoting neurological function recovery. Remote monitoring technology enables doctors to monitor patients' rehabilitation progress in real-time, allowing for prompt adjustments to rehabilitation plans. These technologies not only enhance patients' rehabilitation motivation but also offer more possibilities for personalized rehabilitative care. In the future, with the continuous innovation of medical technology, the patients can anticipate that rehabilitative care, which will become more intelligent and personalized, further enhancing the rehabilitation outcomes.

3.3. Cost-Sharing and Investment Returns

In terms of cost-sharing, the high costs of rare disease treatments often burden both patients and healthcare systems. As depicted in Figure 2, a reasonable cost-sharing mechanism among governments, healthcare institutions, and patients is crucial for driving the sustainable development of rare disease treatments. Governments can provide economic support or cost waivers for patients through policy development. Healthcare institutions need to control costs while providing high-quality treatment, employing more efficient resource allocation methods to ensure treatment accessibility and sustainability. Patients should have an understanding of treatment costs and reasonable expectations, recognizing the balance between treatment value and cost.

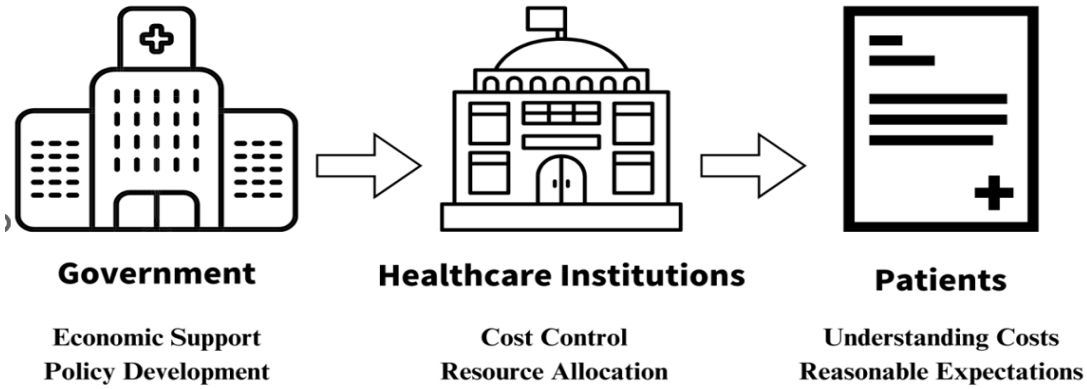


Fig. 2 Cost-sharing for sustainable rare disease treatment [8, 9]

Meanwhile, in the field of drug development, the development of rare disease drugs faces significant challenges and risks. As illustrated in Table 1, despite the investment risks associated with drug development, successful cases indicate that breakthroughs in rare disease drug development can yield substantial investment returns. However, drug development requires not only financial support but also innovative research methods and collaborative partnerships. This creates conditions for attracting more investors to the field, promoting innovation and technological advancement. Governments and industries should strengthen cooperation, establish incentive mechanisms, and provide more support and encouragement for rare disease drug development.

Table 1. Top 10 companies and their core rare disease drugs in global sales revenue from 2018 to 2024 (In billions of US dollars) [10]

Rank	Company	Expected main products for 2024	Operating revenue			Market share	
			2018	2024E	Growth Rate	2018	2024E
1	Celgene	Revlimid	126	137	1.40%	9.60%	5.60%
2	JNJ	Darzalex	84	127	7.20%	6.40%	5.20%
3	Roche	Hemlibra	103	127	3.60%	7.90%	5.20%
4	Novartis	Tafinlar	102	127	3.70%	7.80%	5.20%
5	Takeda	Ninlaro	38	117	20.40%	2.90%	4.80%
6	AbbVie	Imbruvica	37	101	17.90%	2.90%	4.20%
7	Sanofi	Myozyme	53	78	6.90%	4%	3.20%
8	Vertex	VX-659/X-445+Tezacaftor+Ivacaftor	30	78	17%	2.30%	3.20%
9	Alexion	Soliris	41	72	9.60%	3.20%	3%
10	Pfizer	Vynaqel	45	68	7.20%	3.40%	2.80%

3.4. Policy Support and Cooperative Mechanisms

Policy support plays a pivotal role in driving the development of rare disease treatments. Policy support across countries directly impacts the speed and quality of patient treatments. Governments can provide patients with more treatment opportunities and financial support through the formulation of drug approval and medical insurance policies [10]. Policy formulation should prioritize fairness

and sustainability, ensuring that all patients can access appropriate treatments. Furthermore, the establishment of international cooperative mechanisms also brings more opportunities for cooperation and resource sharing in the field of rare disease treatments, aiding in the advancement of global rare disease treatment. Cooperation among international organizations, multinational healthcare institutions, and academia will promote the exchange and innovation of treatment plans, providing patients with more choices and opportunities.

Through this multidimensional analysis, a deeper understanding of the challenges and opportunities in the field of rare disease treatment can be gained. The value-based analysis framework provides the general public with a comprehensive perspective, allowing the governments to consider how to enhance treatment effectiveness and quality of life for rare disease patients and drive the sustainable development of rare disease treatment. Through interdisciplinary collaboration, technological innovation, optimization of cost-sharing mechanisms, and strengthened policy support, better treatment options and quality of life for rare disease patients can be provided.

4. Limitation and Future Outlook

4.1. Research Limitation

While this study has made some progress in exploring the challenges and solutions in rare disease treatment, there are still certain limitations. Additionally, future research can delve more deeply into the following areas to further enhance the understanding of rare disease treatment.

First of all, concerning the limitations, although the analytical framework of this study covers multiple dimensions, practical application might be constrained by data availability. Obtaining precise patient data, drug development costs, and treatment effectiveness assessment information remains a significant challenge for individuals. Consequently, this study primarily focuses on collecting and analyzing macro-level information, and the collection of micro-level information might be exceedingly difficult. This could lead to limitations in the analysis of certain aspects, thus affecting the comprehensiveness of the research conclusions.

Secondly, the analysis in this study regarding policy support and cooperation mechanisms mainly concentrates on the national level, requiring further in-depth research into the specific implementation details of future international cooperation mechanisms. International collaboration necessitates multifaceted considerations to encompass the legal regulations, cultural differences, and income disparities among various collaborating nations. Therefore, exploring how to promote cooperation in rare disease treatment globally still requires more profound investigation.

Lastly, although this study explores current market trends and investment returns in the rare disease drug development market, it does not involve specific investment cases or drug development cost data. Future research could employ case analyses involving various enterprises and government investments in the rare disease field to gain insights into the investment decisions and returns of different sectors and types of investors within the rare disease domain.

4.2. Research Outlook

To better propel the development of the rare disease treatment field, future research could expand in the following directions.

There should be a further emphasis on the collection and analysis of rare disease-related data, thus obtaining more accurate patient data, treatment effectiveness records, and drug development cost data. This will assist in enabling value-based healthcare to more precisely evaluate the effects and economic benefits of diverse treatment approaches, providing decision-makers with more tailored treatment options for patients.

A thorough exploration of specific models and mechanisms for international collaboration is warranted. Through case studies and comparative research, insights can be gained into how different countries establish collaboration mechanisms and share resources and knowledge, thereby collectively advancing rare disease treatment.

Relevant scholars should consider conducting long-term patient tracking studies to assess the long-term effects of personalized treatment approaches. By monitoring patients' treatment outcomes and changes in their quality of life, a more comprehensive understanding of the enduring impacts of various treatment strategies can be acquired.

It is essential to delve into sustainable development models for the drug development market. Future research can focus on the risks and returns associated with different drug development strategies, thereby providing investors and decision-makers with more detailed market information and attracting more investors to engage in the rare disease domain and driving the development of rare disease medications.

In conclusion, despite the limitations of this study, through continuous deepening of research, an improved understanding of the challenges and opportunities in the field of rare disease treatment can be gained. This will provide more robust support for enhancing patient treatment outcomes and quality of life.

5. Conclusion

This article conducts an in-depth study through the analysis framework of value-based healthcare in the treatment of rare diseases, covering various critical aspects such as medical technological innovation, drug development, rehabilitative care, cost-sharing, and policy support. Through literature review and analysis, the article presents a series of key viewpoints and research findings, providing new perspectives and insights into the treatment of rare diseases.

In terms of medical technological innovation, the application of gene editing technology offers personalized treatment approaches for rare diseases. However, ethical and safety issues still require thorough exploration and resolution. Digital technologies such as virtual reality also bring new possibilities to rehabilitative care, enhancing patient recovery outcomes.

Regarding drug development, an increasing number of pharmaceutical companies are investing in the field of rare diseases, accelerating the research and launch of orphan drugs. Drug repurposing strategies also provide quicker avenues for rare disease treatment, shortening drug development cycles. However, drug costs and market access remain challenges that require collaborative efforts from policies and the industry.

In the aspect of cost-sharing, the shared responsibility of treatment costs among governments, medical institutions, and patients is crucial for sustainable development. Policy support plays a significant role in promoting rare disease treatments, but differences in policies among different countries also require further coordination and cooperation.

At the level of treatment plans, the development of personalized treatment plans is key to improving treatment effectiveness. Considering individual differences allows treatments to be more targeted, thus achieving better outcomes. In terms of rehabilitative care, the combination of interdisciplinary collaboration and technological innovation provides more comprehensive support for patients' recovery. This not only enhances rehabilitation outcomes but also increases patients' motivation for treatment.

References

[1] Jones A.B, Smith C.D, Johnson E.F. Applications of gene editing in rare disease treatment. Journal of Rare Diseases Research, 2019, 10(2): 135-148.

[2] Smith J.K, Garcia M.L. The role of virtual reality in rehabilitation for rare disease patients. Innovative Healthcare Technologies, 2020, 5(3): 187-200.

[3] Brown R.H, Robinson L.E, Lee S.M. Market trends in orphan drug development. Journal of Pharmaceutical Economics, 2018, 15(4): 421-435.

[4] Robinson M.P, Lee H.L. Drug repurposing strategies for rare disease treatment. Drug Discovery Today, 2021, 26(6): 1365-1372.

[5] Mullins D, Johnson P, Williams K. Regulatory policies for orphan drug approval: A comparative analysis. Regulatory Review, 2017, 12(3): 223-235.

[6] Smith E.R, Johnson L.W. Health insurance policy impact on rare disease patients' access to treatment. Health Policy and Management, 2019, 22(1): 78-89.

[7] National Organization for Rare Disorders (NORD). NORD advances practical, meaningful, and enduring change so people with rare diseases can live their fullest and best lives, 2023.

[8] Boao Winhealth Rare Disease Medical Center. Report and Scientific Knowledge from the Boao Lecheng Rare Disease Clinical Medical Center, 2023.

[9] Hainan Boao Lecheng International Medical Tourism Pilot Zone. Scientific Knowledge from Bo'ao Lecheng International Medical Tourism Pilot Zone, 2023.

[10] Wang S. Understanding the Rare Diseases and Orphan Drugs. Report and Scientific Knowledge from Wang Shuhe Biomedical (Wuhan) Co., Ltd, 2023.