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Integrating Medical Aid and Philanthropic Support for Rare Diseases: A Global Policy Analysis Based on the Case of Hemophilia

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ABSTRACT

In the global pursuit of health equity, the medical security of the rare disease population is an extremely urgent issue. There are over 7,000 known rare diseases globally, affecting approximately 350 million people. The high treatment costs of these diseases seriously threaten the lives, health, and quality of life of patients, imposing a heavy burden on individuals, families, and society. Against this backdrop, this study comprehensively employs methods such as comparative case studies and policy analysis. Taking hemophilia as an example, it deeply explores the relationship between medical assistance and charity support for rare diseases.By comparing the relevant policies of countries and regions such as China, the United States, the European Union, Japan, and Germany, the study finds that there is a lag in incorporating new gene therapies into medical insurance in China. In contrast, the European Union has reduced the prices of rare - disease drugs by an average of 20% - 30% through its joint procurement mechanism. In City W, China, the charity subsidy coverage rate for rare - disease patients in suburban areas is only 65%, which is much lower than the 90% in urban centers, while Australia has significantly narrowed the regional gap through its national charity resource allocation mechanism. In City W, China, the out - of - pocket expenses of hemophilia patients account for 30% of the total medical costs, while in Japan, the out - of - pocket expense ratio for rare - disease patients has been reduced to less than 10% through multi - party collaboration. In addition, there are problems such as policy fragmentation, low administrative efficiency, and insufficient stakeholder synergy.To address these issues, this study proposes a series of specific solutions. For example, a dynamic adjustment mechanism for the medical insurance catalog should be established to ensure that eligible new therapies are included in medical insurance in a timely manner. A cross - regional equity fund should be set up, including the construction of a special transfer payment fund pool, a regional medical insurance fund mutual assistance mechanism, and a policy coordination mechanism. Digital transformation should be promoted by building a national rare - disease information platform and using blockchain technology to ensure data security, and implementing the "One - Window Acceptance" approval model. The legislative framework should be improved to clarify the rights and responsibilities of all parties, and a tripartite cooperation mechanism among the government, charitable organizations, and pharmaceutical companies should be established. These strategies aim to improve the global rare - disease support system, promote medical fairness, and provide references for the formulation of global rare - disease policies.

Keywords: *Rare diseases; Medical assistance; Charity support; Health equity; Global governance; Hemophilia*

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1. INTRODUCTION

1.1 Research Background

Rare diseases, though individually affecting a small number of patients, collectively pose a significant global health challenge. According to the World Health Organization (WHO), there are over 7,000 known rare diseases, impacting approximately 350 million people worldwide. These diseases are characterized by low prevalence, complex pathophysiology, and often, exorbitant treatment costs. Hemophilia, a hereditary bleeding disorder, serves as a prime example.

Hemophilia is an X - linked recessive genetic disorder, mainly affecting males. Due to a deficiency of specific clotting factors in the body, patients face a lifelong risk of spontaneous bleeding, which can occur in joints, muscles, and internal

organs. This not only causes severe pain but also leads to long - term complications such as joint deformities and chronic disabilities if not properly managed. The standard treatment for hemophilia involves regular replacement of the deficient clotting factor, which is both time - consuming and extremely costly. For instance, in many countries, the annual cost of clotting factor replacement therapy for a severe hemophilia patient can range from hundreds of thousands to over a million yuan.

In the context of global healthcare, the situation varies significantly across different countries. In OECD countries, despite relatively comprehensive medical security systems, hemophilia patients and their families still bear a substantial financial burden. A recent survey in some OECD countries found that, on average, families of hemophilia patients contribute 20 - 30% of the total treatment costs out - of - pocket, which can strain household finances and limit patients' access to optimal care.

In developing countries, the challenges are even more acute. Limited medical resources, underdeveloped healthcare infrastructure, and insufficient insurance coverage mean that many hemophilia patients are unable to receive timely and adequate treatment. In some regions, the lack of access to proper diagnosis and treatment facilities leads to a high rate of undiagnosed cases and untreated bleeding episodes, which can be life - threatening. For example, in certain sub - Saharan African countries, the diagnosis rate of hemophilia is less than 20% of the estimated patient population, and the mortality rate due to untreated bleeding complications is alarmingly high.

In China, with the continuous development of the economy and the increasing emphasis on people's well - being, the rare disease medical security system has gradually been established and improved. In 2018, five ministries and commissions, including the National Health Commission, jointly released the "First Batch of Rare Disease Catalogs," which was a significant milestone in China's rare disease healthcare efforts. However, in the practical implementation process, there are still many problems in the coordination between medical assistance and charity support for rare disease patients, which cannot fully meet the diverse and complex needs of this vulnerable group.

1.2 Research Significance

This research holds both theoretical and practical importance. Theoretically, by applying the theories of welfare pluralism and collaborative governance to the study of rare disease medical assistance and charity support, it enriches the theoretical framework in the field of social security. Welfare pluralism emphasizes the roles of multiple actors, such as the government, market, non - profit organizations, and families, in the supply of social welfare. This provides a comprehensive perspective for analyzing the complex interactions and responsibilities within the rare disease security system. The theory of collaborative governance, on the other hand, offers practical solutions for promoting cooperation among different stakeholders, facilitating the integration of various disciplinary theories in rare disease security practices.

Practically, the findings of this study can provide valuable references for improving the global rare disease medical security system. Taking hemophilia as a case study, analyzing the existing problems in its medical assistance and charity support can reveal common challenges faced by rare disease groups in medical security. This can offer practical lessons for policy - making and practices related to other rare diseases. Moreover, the proposed multidimensional collaborative strategies can help reduce patients' economic burdens, enhance the accessibility and fairness of medical services, and ultimately contribute to the global goal of health equity.

1.3 Research Methods and Data

In this study, to deeply explore the relationship between medical assistance and charity support for rare diseases, a variety of research methods were comprehensively employed, and data from multiple channels were extensively collected for systematic analysis.

The research methods mainly include the comparative case - study method and the policy - analysis method. In the comparative case - study, representative countries and regions were carefully selected for in - depth analysis. The selection of countries and regions such as China, the United States, the European Union, Japan, and Germany is based on multiple considerations. As the largest developing country in the world, China is in a stage of rapid development. During the construction of the rare - disease medical security system, it faces many typical challenges. For example, there is an imbalance in the distribution of medical resources between urban and rural areas and among different regions, and the medical insurance policy lags behind in adapting to the rapid development of medical technology. These problems are common in developing countries, and China's case can provide valuable lessons for other developing countries.

The United States, as a highly developed economy, has leading - edge medical technology and a relatively complete medical security system. Its mechanism for incorporating new drugs and therapies into medical insurance is efficient. Through accelerated approval channels and negotiations with pharmaceutical companies, innovative therapies can be included in insurance coverage relatively quickly. This model provides an important reference for other countries to optimize their

medical insurance policies to facilitate patients' access to advanced treatments.

The European Union, as an alliance composed of multiple developed countries, has adopted a unique joint - procurement mechanism to improve the accessibility of rare - disease drugs. By negotiating drug prices collectively, it can reduce drug prices, making new treatment options more affordable for patients. This regional - cooperation model is innovative and exemplary in solving cross - border rare - disease problems.

When dealing with rare - disease challenges, Japan has formed a close - cooperation model among the government, enterprises, and charitable organizations. They jointly established a special rescue fund and implemented a diversified payment system, which significantly reduces the economic burden on patients. Its successful experience provides useful references for other countries in integrating resources from various sectors of society.

Germany excels in stakeholder synergy. A good cooperation mechanism has been established among the government, charitable organizations, and pharmaceutical companies. Through sound policy support, active charity assistance, and the fulfillment of corporate social responsibility, the stable and efficient operation of the rare - disease support system is guaranteed, providing an example for other countries to promote cooperation among all parties.

City W in China was selected as a regional case because the problems exposed in its medical assistance and charity support are typical and representative. For example, in City W, the update of the medical insurance catalog cannot keep up with the development of medical technology, resulting in new treatment methods being difficult to be quickly included in medical insurance. This problem is widespread in many regions of China. At the same time, there is an uneven distribution of charity resources between urban and rural areas. The charity subsidy coverage rate for patients in suburban areas is much lower than that in urban centers, reflecting the impact of regional development imbalance on the assistance of rare - disease patients. Through the study of City W, we can deeply understand the dilemmas and challenges faced by specific regions in practice.

In terms of policy analysis, a comprehensive and in - depth analysis was carried out on the relevant policies of various countries and regions, including the formulation background, goal setting, specific measures, and implementation - effect evaluation of the policies. By systematically combing and analyzing these policies, the influencing factors behind the policies, such as economic development level, political system, and social and cultural background, were explored.

2. THEORETICAL APPLICATION

2.1 Welfare Pluralism

Welfare pluralism posits that the supply of social welfare requires the joint participation of multiple actors, including the government, market, non - profit organizations, and families, to build a multi - level and multi - dimensional welfare supply system. In the field of rare - disease medical assistance and charity support, each actor has a distinct role. The government, as the main provider of public welfare, formulates policies, provides financial support, and ensures regulatory compliance. For example, some countries have introduced legislation to protect the treatment rights of rare - disease patients. The market provides supplementary welfare through mechanisms such as commercial insurance and private medical services. Charitable organizations can respond flexibly to the emerging needs of patients, and families assume the responsibilities of daily care and emotional support. From the perspective of the "Health System Strengthening" theory, each actor acts on different levels of the health system. Government policies influence the institutional construction and resource allocation of the health system, providing a macro - framework for rare - disease management. Market mechanisms promote more efficient resource allocation to meet the diverse needs of patients. The participation of charitable organizations and families enriches the service content of the health system and improves the quality of patient care, jointly contributing to the strengthening of the health system. For instance, patient education activities carried out by charitable organizations can help improve patients' self - management ability and enhance the resilience of the health system in dealing with rare diseases.

(1) Linkage with Empirical Analysis

As shown in Figure 1 "Comparison of Global Rare Disease Medical Assistance Coverage Rates (2023)", we can observe the practical manifestations of welfare pluralism. The government, through financial investment and policy - making, provides a basic guarantee for rare - disease patients' medical assistance. However, as seen in the case of China, there are still issues in policy implementation, such as the slow inclusion of new gene therapies in medical insurance. The market, represented by commercial insurance, offers additional support, but it also faces challenges like market failure in rare - disease drug development due to high costs and small patient populations. Charitable organizations contribute to the assistance, but in some regions, like City W in China, there are problems with unequal distribution of charity resources.

(2) Limitations

Welfare pluralism has its limitations in the rare - disease context. The goals of different actors may conflict. The

government aims for public welfare, the market pursues profit, and non - profit organizations focus on social influence. This can lead to difficulties in coordinating resource allocation and policy implementation. For example, in rare - disease drug pricing, the government's pursuit of affordability and the market's need for profit can create tensions. Also, the boundaries of responsibilities among different actors are not always clear, which may result in a situation where no one takes full responsibility for certain aspects of patient care.

2.2 Collaborative Governance

Collaborative governance advocates for equal participation, resource sharing, and joint decision - making among multiple stakeholders in public affairs governance. In the field of rare diseases, all parties need to share patient medical information, treatment needs, etc., to achieve precise assistance and policy adjustment. At the same time, clarifying responsibilities and coordinating interests are crucial for the sustainable development of the support system. Combining with the "Health System Strengthening" theory, this collaborative governance model is essential for strengthening the health system. The collaboration among all parties can optimize the resource integration of the health system and avoid resource waste and duplicate investment. For example, the cooperation between medical institutions and charitable organizations can accurately match charitable resources with patient needs, improve resource utilization efficiency, and enhance the service - providing ability of the health system. Moreover, through collaborative policy - making and planning, the consistency of goals in rare - disease prevention and treatment within the health system can be ensured, and the overall effectiveness of the system can be improved.

(1) Linkage with Empirical Analysis

In practice, as demonstrated by the situations in City W and Germany, collaborative governance shows different results. In City W, the lack of synergy among the government, charitable organizations, and pharmaceutical companies leads to a fragmented support system, with inefficiencies and gaps in patient care. In contrast, Germany has a high - level of collaboration, where the government, charitable organizations, and pharmaceutical companies work together effectively. The government creates a favorable policy environment, charitable organizations provide personalized assistance, and pharmaceutical companies invest in R&D and ensure drug accessibility.

(2) Limitations

However, collaborative governance also faces challenges. In reality, power imbalances exist among stakeholders. The government often has more power in decision - making, while non - profit organizations and enterprises may have less influence, which can undermine the principle of equal participation. Additionally, establishing an effective information - sharing mechanism is difficult. Information systems of different organizations may be incompatible, and data security concerns can prevent seamless information flow, reducing the efficiency of collaborative governance.3 Global Challenges and Policy Comparisons

3. SYSTEMIC BARRIER

3.1.1 Policy Fragmentation

In City W, China, and on a broader scale in China, the update of the medical insurance catalog fails to keep pace with the rapid development of medical technology. Taking hemophilia as an example, new gene therapies, which hold great promise for long - term treatment by correcting the underlying genetic defects of hemophilia, are mostly not covered by medical insurance. The exorbitant cost of these therapies, often exceeding 1 million yuan per treatment, places them far beyond the reach of the vast majority of hemophilia patients. This situation not only restricts patients' access to advanced medical treatments but also exacerbates their long - term health risks and economic burdens.

In contrast, the United States has established a relatively efficient mechanism for incorporating new drugs and therapies into medical insurance. Through accelerated approval channels and negotiations with pharmaceutical companies, innovative therapies can be included in insurance coverage in a relatively short time. For instance, for a recently approved gene therapy for a rare form of hemophilia, it took less than two years from its initial approval by the Food and Drug Administration (FDA) to being covered by major insurance providers. According to relevant reports of the Organization for Economic Co - operation and Development (OECD), under the accelerated approval process in the United States, the average time for rare - disease innovative therapies to be included in medical insurance is about 1.5 - 2 years. In China, the process of including similar therapies in medical insurance is much slower, with a significant time lag. This delay means that Chinese patients have to wait much longer to benefit from these advanced medical technologies, which can have a profound impact on their quality of life and long - term health outcomes.

The European Union has made remarkable progress in enhancing the accessibility of new drugs and therapies through its joint procurement mechanism. By negotiating drug prices collectively, EU member states can secure more favorable pricing, making new treatment options more affordable for patients. This mechanism has been particularly effective in the field of rare - disease drugs, where high prices have long been a major obstacle to treatment. According to public reports

of the World Health Organization (WHO), in the joint procurement of rare - disease drugs in the EU, the drug price can be reduced by an average of 20% - 30%. For example, for a certain special - effect drug for treating rare diseases, before the joint procurement, the average annual treatment cost for European patients was as high as 300,000 euros. After the joint procurement, the cost dropped to between 210,000 and 240,000 euros. This significant price reduction has not only alleviated the financial burden on patients but has also improved the overall treatment rate and prognosis for rare - disease patients in the EU. It also encourages more research and development in the rare - disease drug field by ensuring that pharmaceutical companies can still maintain reasonable profits while making their products more accessible.This is a detailed analysis of the differences in the timing and cost of gene therapy inclusion in each country, and Table 1 can systematically present these data to enhance the persuasiveness of the argument. As shown in Table 1, the average time from approval to inclusion of gene therapy in the United States is 1.5-2 years, while in China there is a significant time lag.

Table 1: Comparison of the timing and cost of gene therapy inclusion in health insurance by country (as of 2023)

Country/Region	Name of Gene Therapy	Approval Time	Time of Inclusion in Medical Insurance	Treatment Cost (per Session)	Patient's Out - of - Pocket Expense Proportion
United States	Hemgenix	November 2022	June 2023	$3.5 million	10%
European Union	Roctavian	August 2022	March 2023	$2.8 million	5%
Japan	Not Approved Yet	Not Approved Yet	Not Included	Not Disclosed	Not Disclosed
China	Under Research	Not Approved Yet	Not Included	Not Disclosed	Not Disclosed

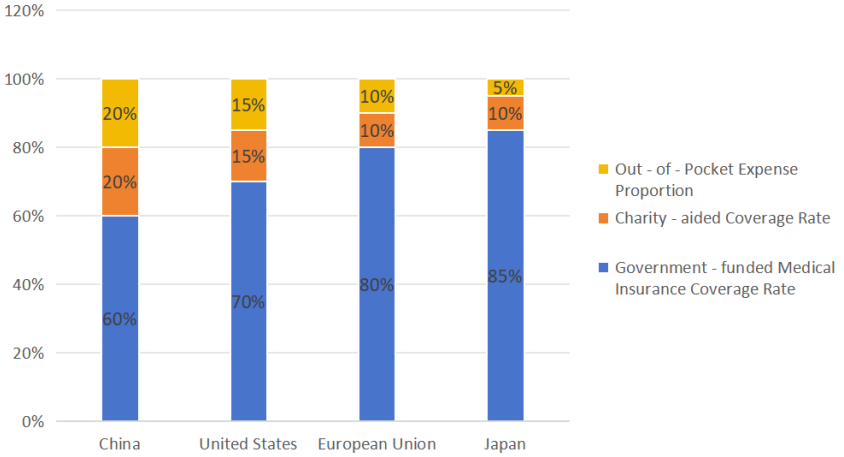
Source: Based on public data from the FDA, EMA, Japan's Ministry of Health, Labor and Welfare, and China's National Health Insurance Administration

3.1.2 Regional Inequity

In City W, China, there is a severe imbalance in the distribution of charity resources. The charity subsidy coverage rate for patients in suburban areas is merely 65%, while that in urban centers reaches as high as 90%. Suburban patients, who generally have fewer economic resources and less convenient access to medical facilities, are at a distinct disadvantage when it comes to obtaining charity assistance. This regional disparity not only leads to unequal access to medical resources but also widens the gap in the quality of life and health outcomes between urban and suburban rare - disease patients. For example, in some suburban areas of City W, patients may have to travel long distances to access specialized medical services, and the lack of sufficient charity support means they often face financial difficulties in covering transportation costs and out - of - pocket medical expenses.

Australia, on the other hand, has established a national charity resource allocation mechanism. As reported by the World Health Organization, a special coordinating agency for rare - disease charity resources in Australia collects comprehensive data on the number of rare - disease patients, their economic status, and the local availability of medical resources in different regions. Based on this information, the agency allocates charity resources in a more equitable manner. Through this mechanism, the gap in access to charity support between remote areas and urban areas has been significantly narrowed. The charity coverage rate for patients in remote areas has increased from about 50% previously to around 75%. This achievement has been crucial in ensuring that all rare - disease patients in Australia, regardless of their geographical location, can receive the necessary support. It also promotes social equality and helps to reduce the overall burden on the healthcare system by ensuring that patients can access timely and appropriate care, reducing the likelihood of more serious health complications and the need for expensive emergency treatments.The differences in medical assistance coverage in different regions of the world are discussed here, and Figure 1 provides a visual representation of comparative data from China, the United States, Europe, and Japan to support the argument. As shown in Figure 1, the coverage rate of medical assistance for rare disease patients in China is significantly lower than that of the United States and the European Union, especially in terms of philanthropic assistance.

Figure 1: Comparison of global medical assistance coverage for rare diseases (2023)



Source: Based on the WHO 2023 Global Report on Rare Diseases and public data from health authorities in various countries

3.2 Economic Burden

3.2.1 Out - of - Pocket Costs

Hemophilia patients in City W, China, bear a heavy financial burden. Out - of - pocket expenses account for up to 30% of the total medical costs, and the annual charity subsidies they can receive are relatively limited, approximately 18,000 yuan. For patients with severe hemophilia who require frequent and expensive treatment, such as regular clotting factor replacement therapy, this financial pressure is overwhelming. The high cost of treatment often forces patients to cut back on necessary medical care, which can lead to more frequent and severe bleeding episodes, long - term joint damage, and other complications. In some cases, patients may even be unable to afford the most basic medical supplies, further deteriorating their health conditions.

In Japan, the government, enterprises, and charitable organizations have joined forces to establish a special rescue fund for rare diseases. Through a combination of government subsidies, corporate donations, and public fundraising, this fund provides substantial financial support for rare - disease patients. Additionally, Japan has implemented a diversified payment model that combines medical insurance, commercial insurance, and charity support. As a result, the out - of - pocket expense ratio for rare - disease patients in Japan has been reduced to less than 10%. According to statistical data from the OECD, after the implementation of this model, the average annual out - of - pocket medical expenses for rare - disease patients in Japan have decreased from about 800,000 yen previously to less than 300,000 yen. This significant reduction in out - of - pocket expenses has not only alleviated the economic pressure on patients and their families but has also improved their overall quality of life. It allows patients to focus more on their treatment and recovery without the constant worry of financial ruin, and it also enables them to access a wider range of medical services and support, leading to better health outcomes.

3.2.2 Administrative Inefficiency

In City W, the application process for charity support is complex. Patients are required to submit multiple documents, such as medical records, income certificates, and application forms. This complex process not only places a heavy burden on patients but also hampers the timely delivery of charity support. The need to gather and submit these documents can be time - consuming and stressful for patients, especially those who are already dealing with the physical and emotional challenges of a rare disease.

Singapore has taken a different approach by establishing a one - stop assistance application platform. Patients can submit all their applications on this single platform, which is connected to various government agencies and charitable organizations. Through this platform, relevant departments can share information and conduct joint approvals, reducing the approval time to less than 7 days. According to reports from the Singaporean health department, after the launch of this platform, the success rate of rare - disease patients applying for charity support has increased by 20%, and the average time from application to receiving assistance has been shortened from the original 12 days to about 5 days. This efficient system has greatly improved the accessibility of charity support for rare - disease patients in Singapore. It has also enhanced the overall efficiency of the charity support system, ensuring that resources are allocated more effectively and that patients receive the help they need in a timely manner. This not only benefits the patients directly but also improves the reputation and credibility of the charity support system, encouraging more people to contribute to rare - disease causes.

3.3 Stakeholder Synergy

In City W, the synergy among stakeholders in the field of rare - disease medical assistance and charity support is insufficient. The government, charitable organizations, and pharmaceutical companies lack effective coordination. The government's policies may not fully consider the capabilities and initiatives of charitable organizations and pharmaceutical companies, while charitable organizations and pharmaceutical companies may not have a clear understanding of the government's long - term goals and priorities. This lack of synergy leads to a fragmented support system, where resources are not integrated effectively, and patients may not receive comprehensive and coordinated care. For example, there may be duplication of efforts in some areas, while other areas may be overlooked, resulting in inefficiencies and gaps in the support provided to patients.

In Germany, there is a high level of collaboration among the government, charitable organizations, and pharmaceutical companies. The government plays a leading role in formulating policies and providing financial support. It creates a favorable policy environment for the development of the rare - disease treatment industry, such as offering tax incentives for pharmaceutical companies engaged in rare - disease drug research. At the same time, it allocates sufficient financial resources to support medical assistance programs. Charitable organizations in Germany are actively involved in fundraising and providing personalized assistance to patients. They conduct in - depth investigations into the specific needs of patients and work closely with medical institutions to ensure that assistance resources are used effectively. Pharmaceutical companies, as key players in the medical industry, are committed to investing in research and development, production, and supply of rare - disease drugs. They also collaborate with the government and charitable organizations to ensure that drugs are accessible and affordable for patients. Germany has established a well - functioning interest - coordination mechanism. This mechanism helps to balance the interests of all parties, ensuring that the rare - disease support system can operate stably in the long term. Through regular communication and negotiation, stakeholders can resolve conflicts and reach consensus on important issues, promoting the continuous improvement of the rare - disease support system.4 Multidimensional Solutions.

4. MULTIDIMENSIONAL SOLUTIONS

4.1 Policy Innovation

4.1.1 Dynamic Insurance Adjustment

A dynamic adjustment mechanism for the medical insurance catalog is crucial. From the perspective of the Health System Strengthening Theory, this aligns with the need to enhance the accessibility of medical products and technologies. Every year, a comprehensive evaluation should be carried out, involving medical experts, medical insurance department staff, and representatives from pharmaceutical companies. During this evaluation, factors such as clinical efficacy, cost - effectiveness, and patient needs should be carefully considered.

For example, in the case of new gene therapies for hemophilia, once their safety and efficacy have been established through rigorous clinical trials, and a comprehensive cost - effectiveness analysis has been conducted, they should be gradually included in the medical insurance catalog. The government, as emphasized in the Health System Strengthening Theory's leadership and governance aspect, should play a leading role in coordinating this process. In the price negotiation process, the medical insurance department should refer to international market prices and the production costs of domestic pharmaceutical companies. By conducting multiple rounds of negotiations with pharmaceutical companies, a reasonable price can be reached, ensuring both the affordability for patients and the reasonable profits for pharmaceutical companies. This not only enables patients to benefit from advanced medical technology in a timely manner but also promotes the sustainable development of the rare - disease drug industry, in line with the financing requirements of the theory.

4.1.2 Cross - Regional Equity Funds:

To address the regional differences in medical security, a cross - regional assistance cooperation mechanism is essential, which is in line with the resource allocation optimization principle of the Health System Strengthening Theory. The central government can establish a special transfer payment fund pool for rare diseases. Through big data analysis of regional economic development levels, the number of rare disease patients, and the severity of their conditions, funds can be allocated in a scientific and reasonable manner, with a focus on supporting economically underdeveloped regions. This helps to ensure that rare - disease patients in different regions can enjoy fair and effective medical security, promoting equity in the health system.

An inter - regional medical insurance fund mutual assistance mechanism should also be established. This mechanism encourages paired assistance between economically developed regions and underdeveloped regions. When the medical insurance fund for rare disease protection in an underdeveloped region is insufficient, developed regions can provide financial support according to a pre - negotiated proportion. In addition, an inter - regional medical insurance policy coordination mechanism should be put in place. Regular exchanges and cooperation among medical insurance departments

in different regions can help unify some policy standards, promote the connection and coordination of medical insurance policies, and improve the overall use efficiency of medical insurance funds. These measures optimize the resource allocation within the health system, enhancing its overall performance.

4.2 Digital Transformation

4.2.1National Information Platforms

A national rare disease information platform coordinated by the National Health Commission is needed. This is in line with the Health System Strengthening Theory's requirement for a sound health information system. This platform should connect patients, medical institutions, medical insurance departments, and charitable organizations, enabling real - time sharing of diagnosis and treatment data, assistance records, and drug inventory information.

To ensure the security and privacy of patient information, blockchain technology can be employed. Blockchain provides a decentralized and encrypted way of storing and sharing data, making it difficult for unauthorized access or data tampering. A data backup and recovery mechanism should also be established to prevent data loss. For instance, Australia has developed a blockchain - based charity fund tracking system. In this system, every transaction of charity funds is recorded on the blockchain. Donors can use this system to track the entire process of funds from donation to usage in real - time, ensuring that the funds are accurately used for the treatment and rehabilitation of rare - disease patients. This system has effectively enhanced public trust in charitable organizations and improved the efficiency of charity fund utilization (refer to the official reports of Australian charitable organizations). According to relevant research published in Nature Digital Medicine, blockchain technology has a high degree of feasibility in medical data management and fund tracking. Its encryption algorithm can effectively protect patient privacy, ensuring the security of medical data during sharing. Meanwhile, its distributed ledger feature makes the flow of funds transparent and difficult to tamper with, providing a reliable guarantee for the management of charity funds.

By analyzing the data collected on the platform, valuable insights can be obtained. For example, through big data analysis, patterns in patient treatment needs, regional differences in disease prevalence, and the utilization of medical resources can be identified. This information can then be used to support policy - making, resource allocation, and the development of targeted assistance programs, which is crucial for improving the overall performance of the health system as per the Health System Strengthening Theory.

4.2.2 One - Window Approval:

The "One - Window Acceptance" approval model should be implemented. This model integrates the application materials for medical assistance and charity support, allowing patients to submit basic materials at a single location, such as a community health service center or a designated government service window. This is in line with the Health System Strengthening Theory's emphasis on optimizing service delivery.

Subsequent approvals can be coordinated online by relevant departments, including the medical insurance department, civil affairs department, and charitable organizations. Estonia's national health information platform serves as a successful example of a one - stop platform for rare - disease medical assistance and charity support. This platform integrates resources from medical services, medical insurance, and social welfare. During the medical treatment process, patients do not need to run around multiple departments and institutions when applying for medical assistance or charity support. Through the platform, patients' medical information can be shared with relevant departments and charitable organizations in real - time, enabling rapid approval and precise assistance. For example, when rare - disease patients apply for charity medical expense subsidies, the platform automatically matches their medical records, economic status, and other information. Charitable organizations and medical insurance departments can conduct online joint approvals, greatly shortening the approval time (refer to the relevant materials of the Estonian Ministry of Health). Research in Nature Digital Medicine indicates that one - stop platforms break down information silos using digital technology, enabling efficient data circulation and sharing. By establishing unified information standards and interfaces, the systems of different departments and organizations can be seamlessly connected, improving service efficiency and coordination.

To ensure the successful implementation of this model, it can be piloted in selected regions with different economic development levels, population structures, and informatization levels. By summarizing the experiences and lessons from the pilot projects, the model can be refined and gradually rolled out nationwide. A set of supporting mechanisms, such as unified material standards, optimized approval processes, and staff training, should be established. These mechanisms will help shorten the average approval time to 7 working days, improve the assistance efficiency, and provide more convenient services for patients, thus enhancing the overall service - providing ability of the health system.

4.3 Stakeholder Synergy

4.3.1 Legislative Frameworks

The legislation of the "Rare Disease Prevention, Treatment, and Assistance Law" is urgently needed. From the perspective

of the Health System Strengthening Theory, this is an important aspect of leadership and governance. During the legislative process, the rights and responsibilities of different stakeholders, including the government, charitable organizations, pharmaceutical companies, and patient families, should be clearly defined.

The government should assume the leading responsibility in formulating policies, providing financial guarantees, and strengthening supervision. Charitable organizations should be required to fulfill their duties of supplementary assistance and providing personalized services. Pharmaceutical companies should be obligated to invest in rare disease drug research and development, production, and supply. For example, pharmaceutical companies could be mandated to invest 1 - 3% of their annual profits in rare disease drug R&D. This legislation will provide a solid legal foundation for rare disease assistance, standardize the behavior of all parties, and ensure the orderly progress of assistance work, which is essential for the stable operation of the health system.

4.3.2 Tripartite Partnerships

A tripartite cooperation mechanism among the government, charitable organizations, and pharmaceutical companies should be established. Each party has a distinct role to play in improving the rare disease support system, which is also in line with the collaborative governance concept within the Health System Strengthening Theory.

The government takes the lead in policy - making. It formulates favorable policies to encourage the development of rare disease - related industries, such as providing tax incentives for pharmaceutical companies engaged in rare disease drug research. It also allocates financial resources to support medical assistance programs and monitors the implementation of policies to ensure fairness and effectiveness. For instance, the government can set up special funds for rare disease research and treatment, which are used to subsidize clinical trials and the development of new therapies. This promotes the development of medical products and technologies and ensures the financial sustainability of the rare - disease support system.

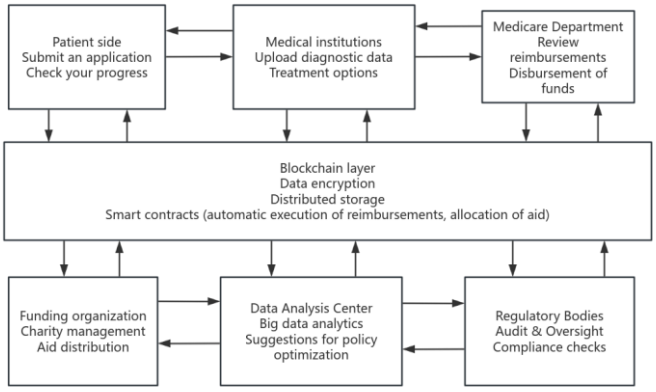
Charitable organizations play a crucial supplementary role. They can conduct in - depth investigations into the special needs of patients and the gaps in medical insurance reimbursement. Based on these findings, they can provide targeted assistance, such as covering the remaining medical expenses that are not reimbursed by insurance. In addition, they offer personalized services like psychological counseling and home - based care support. Some charitable organizations also play an active role in raising public awareness of rare diseases through various public welfare activities, which helps to attract more social resources for rare disease assistance. This enriches the service content of the health system and improves the quality of patient care.

Pharmaceutical companies, as key players in the medical industry, have a responsibility to contribute to the improvement of rare disease treatment. They can reduce drug prices through price negotiations with the government and insurance agencies, making medications more affordable for patients. Pharmaceutical companies can also provide free drugs or discounted drugs to patients in need, especially those from low - income families. Moreover, they should increase investment in research and development to promote the innovation of rare disease drugs and therapies. For example, a pharmaceutical company could collaborate with research institutions to develop more effective and less expensive gene therapies for hemophilia. This promotes the progress of medical products and technologies in the rare - disease field.

To ensure the smooth operation of this tripartite cooperation, a joint meeting system should be established. Regular negotiation meetings are held to discuss and coordinate differences in goals and strategies among the three parties. Joint working groups are formed to be responsible for the implementation of specific cooperation projects. For example, a joint working group can be established to oversee the distribution of free drugs provided by pharmaceutical companies to patients through charitable organizations, with the government ensuring compliance and fairness in the process.

An information - sharing mechanism is also essential. The three parties should share data on patient needs, drug development progress, and available resources. This helps to avoid duplication of efforts and optimize resource allocation. For example, pharmaceutical companies can share information about new drug research plans with the government and charitable organizations, enabling the government to adjust its R&D support policies and charitable organizations to plan relevant assistance programs in advance. This improves the overall efficiency and effectiveness of the rare - disease support system, in line with the requirements of the Health System Strengthening Theory.As shown in Figure 2-1, blockchain technology ensures the data security and transparency of the rare disease information platform through decentralization, encrypted storage and smart contracts, and can also visually display the platform architecture to help understand the implementation of the technology.

Figure 2-1 Schematic diagram of the architecture of the blockchain information platform



Source: Based on the author's own work, with reference to the Estonian Health Information Platform and IBM Blockchain Medical Solutions

4.3 Stakeholder Synergy

Legislative Frameworks: The legislation of the "Rare Disease Prevention, Treatment, and Assistance Law" is urgently needed. During the legislative process, the rights and responsibilities of different stakeholders, including the government, charitable organizations, pharmaceutical companies, and patient families, should be clearly defined.

The government should assume the leading responsibility in formulating policies, providing financial guarantees, and strengthening supervision. Charitable organizations should be required to fulfill their duties of supplementary assistance and providing personalized services. Pharmaceutical companies should be obligated to invest in rare disease drug research and development, production, and supply. For example, pharmaceutical companies could be mandated to invest 1 - 3% of their annual profits in rare disease drug R&D. This legislation will provide a solid legal foundation for rare disease assistance, standardize the behavior of all parties, and ensure the orderly progress of assistance work.

Tripartite Partnerships: A tripartite cooperation mechanism among the government, charitable organizations, and pharmaceutical companies should be established. Each party has a distinct role to play in improving the rare disease support system.

The government takes the lead in policy - making. It formulates favorable policies to encourage the development of rare disease - related industries, such as providing tax incentives for pharmaceutical companies engaged in rare disease drug research. It also allocates financial resources to support medical assistance programs and monitors the implementation of policies to ensure fairness and effectiveness. For instance, the government can set up special funds for rare disease research and treatment, which are used to subsidize clinical trials and the development of new therapies.

Charitable organizations play a crucial supplementary role. They can conduct in - depth investigations into the special needs of patients and the gaps in medical insurance reimbursement. Based on these findings, they can provide targeted assistance, such as covering the remaining medical expenses that are not reimbursed by insurance. In addition, they offer personalized services like psychological counseling and home - based care support. Some charitable organizations also play an active role in raising public awareness of rare diseases through various public welfare activities, which helps to attract more social resources for rare disease assistance.

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through charitable organizations, with the government ensuring compliance and fairness in the process.
An information - sharing mechanism is also essential. The three parties should share data on patient needs, drug development progress, and available resources. This helps to avoid duplication of efforts and optimize resource allocation. For example, pharmaceutical companies can share information about new drug research plans with the government and charitable organizations, enabling the government to adjust its R&D support policies and charitable organizations to plan relevant assistance programs in advance.

5. CONCLUSION

5.1 Research Summary

The relationship between medical assistance and charity support for rare diseases is a complex and multi - faceted issue, involving various aspects such as systems, resources, information, and stakeholders. Through a comprehensive analysis of global policies and practices, this study has identified several common problems. These include policy lags, which prevent patients from accessing the latest medical treatments in a timely manner; resource allocation inequalities, resulting in some patients receiving inadequate support while others enjoy more favorable conditions; and administrative inefficiencies, which impede the smooth delivery of assistance.

To address these issues, this study proposes multidimensional strategies such as system optimization, resource integration, information sharing, and stakeholder collaboration, aiming to build a more comprehensive and effective rare disease support system. These strategies are designed to enhance the coordination and cooperation among different actors, improve the accessibility and fairness of medical services, and ultimately reduce the economic and psychological burdens on rare disease patients and their families.

5.2 Research Limitations

This study has certain limitations. Firstly, the research mainly focuses on hemophilia as a single rare disease case. Although hemophilia is representative, different rare diseases vary significantly in terms of pathology, treatment methods, and social impacts. As a result, the research findings may not be fully applicable to other types of rare diseases. Secondly, when analyzing the situation in China, the data from some regions (such as City W) are mainly used as examples. These regional data are difficult to comprehensively represent the diversity and complexity of China as a whole, and may not accurately reflect the differences and characteristics of rare - disease medical assistance and charity support in different regions. In addition, although this study proposes various improvement strategies, it does not conduct a quantitative analysis of the effects after the implementation of these strategies. It is difficult to precisely evaluate the actual impact of each strategy on aspects such as reducing patients' burdens, improving the accessibility and fairness of medical services.

5.3 Future Improvement Directions

Future research can be improved in multiple aspects. First, expand the scope of research to cover more types of rare diseases and patient groups from different regions around the world. By comparing and analyzing the differences in medical assistance and charity support for different rare diseases, in - depth understanding of the unique problems and challenges faced by different patient groups can be achieved, providing a basis for formulating more targeted policies. Second, improve the data collection and analysis methods. Gather more extensive regional data in China, taking into account factors such as the economic development level, distribution of medical resources, and cultural background of different regions to ensure that the research results can more accurately reflect the actual situation in China. Third, strengthen the quantitative research on the implementation effects of strategies. Use scientific evaluation methods, such as establishing an evaluation index system and conducting empirical research, to track and evaluate the proposed multidimensional strategies, quantitatively analyze their effectiveness in improving patients' economic situations and enhancing the quality of medical services, and provide more persuasive data support for policy - making and optimization, thus promoting the development of the global rare - disease assistance cause.

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Conflicts of Interest

None declared.

Data Availability

Data are available upon reasonable request.

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