Proceedings of the 5th International Conference on Interdisciplinary Humanities and Communication Studies
DOI: 10.54254/2753-7064/2026.38116

From Cost Burden to Systemic Resilience: Structural Transformation of Healthcare Economies in the Era of Rare Disease

Hanguo Wang

Haidian Kaiwen Academy, Beijing, China
wanghanguo080915@163.com

Abstract. Rare diseases have changed from a pure medical problem into a system-wide economic challenge. The wide use of genetic sequencing, progress in drug development, and longer survival of patients have turned rare disease spending from high one-time costs into lifelong continuous consumption, placing great pressure on the healthcare system. This paper analyzes the economic burden of rare diseases from three dimensions: individuals, medical insurance, and society. It points out that direct medical costs are only one part, while hidden costs such as productivity loss and caregiving costs are even heavier. Current drug cost-effectiveness evaluation standards and annual budget payment models cannot accommodate orphan drugs, which are costly and require long-term treatment. This paper proposes a shift from passively bearing costs to enhancing health system resilience. By setting up a national rare disease registration and monitoring system, building multi-level risk-sharing funds, and using payment methods based on treatment results and payment in installments, the healthcare system can better deal with long-term pressure. These measures not only help rare disease patients but also prepare the whole society better for future public health challenges.

Keywords: Rare diseases, economic burden, orphan drugs, health system resilience, healthcare financing

1. Introduction

In the past, rare diseases were often considered not common in daily life and had no relation to the majority of people. Thus, most institutions and people considered that rare diseases would not have any impact on the economy. However, this perception is inconsistent with the facts.

To be specific, the following three changes are occurring in the field of rare diseases. First, with the spread of genetic sequencing technology, more people with rare diseases have been identified. Second, more and more drugs for rare diseases have been developed, and now patients can live longer with continuous treatment. Third, as a result, the pattern of medical expenses has shifted from short-term, large-scale payments to lifelong, continuous consumption [1].

Together, the three changes have turned rare diseases from a medical problem into an economic system problem. Yang and others have estimated that the total economic burden of the 379 rare

© 2026 The Authors. This is an open access article distributed under the terms of the Creative Commons Attribution License 4.0 (https://creativecommons.org/licenses/by/4.0/).

DOI: 10.54254/2753-7064/2026.38116

diseases in the United States in 2022 was nearly 1 trillion US dollars [2]. Less than half of this was direct medical expenses; most of the cost came from hidden factors such as lost productivity and care expenses. Although rare disease drugs are used by less than 1% of the population, they already account for 10% of the total drug expenditure in some countries.

The original design of the healthcare system, drug-pricing methods, and medical-resource allocation focused on common illnesses and short-term care. Now that there are many more long-term, high-cost, and rare diseases, a different approach is needed. This paper is concerned with. This paper discusses these issues.

2. Rare diseases' impact on the medical economy

2.1. Individual dimension: very high costs

The cost of treating rare diseases is relatively high compared with the cost of medicine alone [1]. Spinal Muscular Atrophy is a typical representative. This relatively well-known rare disease can cost the patients tens of thousands of euros in direct medical expenses each year. Gene therapy may cost more than a million yuan per injection and is expected to be a one-time treatment; therefore, the families and health insurance companies will face considerable cash flow pressure due to the large initial payment.

The economic burden of many rare diseases also includes non-drug expenses. In a 2026 study on X-linked retinitis pigmentosa, societal costs such as lost productivity and caregiving accounted for as high as 79% of the total lifetime cost [3]. Therefore, if only the cost of drugs is considered, the actual economic burden of rare diseases will be understated.

2.2. Healthcare dimension: pressure on the healthcare system

Every citizen pays for medical insurance in China. In short, healthy people help the sick by taking on some of the risks. This way, it works well for common illnesses; the cost per person is relatively low, and thus the risk is spread out.

However, the situation of rare diseases is the opposite of that for common diseases: there are fewer patients and higher costs, so when a few patients with rare diseases take very expensive drugs, it can consume a large portion of the entire medical insurance fund. In 2011, Schey and his colleagues predicted that the proportion of spending on orphan drugs would reach a peak of about 4.6% of total pharmaceutical expenditure before declining, and the overall growth of the pharmaceutical market would offset this increase [4]. However, this structure in which a few people use most of the resources is still a basic problem for the healthcare system under the law of large numbers [4].

A 2024 scoping review of economic analyses of rare kidney diseases also found that the evidence base was limited and that conventional health-economic techniques may systematically underestimate the economic burden of rare kidney diseases [5]. In short, the above results indicate that some of the instruments for assessing healthcare expenditure may not fully reflect the particular problems in rare diseases.

2.3. Social dimension: the hidden costs

In addition to the above treatment fees, there are other costs for rare diseases. According to the EveryLife Foundation's study on the national economic burden of rare diseases in the United States,

DOI: 10.54254/2753-7064/2026.38116

indirect costs (mainly productivity losses) were about $437 billion in 2019 [6]. Early retirement for some older people, loss of income for their families, and extended care needs all incur costs that are not covered by health insurance; these costs must ultimately be borne by society as a whole [6].

As a result, the economic impact of rare diseases is not only due to high drug prices but also a systemic problem affecting individuals and families, as well as health insurance and society at large.

3. Deficiencies in the current rules for existing problems

3.1. An inappropriate evaluation standard

Most of the past pharmacoeconomic studies have used the cost-effectiveness ratio to decide whether a new drug will be covered by insurance. This ratio indicates how much money is required to help a patient live for an extra year, and there is a generally accepted limit for this amount.

Orphan drugs (drugs for rare diseases) have cost-effectiveness ratios several times higher than those of conventional drugs. Chambers et al. systematically compared orphan and non-orphan drugs approved by the US Food and Drug Administration from 1999 to 2015. They found that the median incremental cost-effectiveness ratio for orphan drugs was $276,288 per quality-adjusted life year (QALY), and it was only $100,360 per QALY for non-orphan drugs [7]. The research also found that the yearly treatment cost for orphan drugs was about five times that for other drugs ($150,854 vs. $33,654) [7]. If the health insurance companies still use the old criteria, most orphan drugs will not be covered. However, if they are not covered, patients cannot afford them, and thus the development of these drugs will have little practical effect.

Given this contradiction, health insurance authorities in countries such as the United Kingdom and the United States have been compelled to relax the rules for rare disease drugs by raising the reimbursement threshold. However, this is in fact a temporary measure after determining that the system is not applicable, rather than a redesign of the system itself.

3.2. Payment model and cross-departmental collaboration

Current health insurance operates on an annual settlement basis—spending and revenues are calculated for the current year, and next year is a separate account. However, rare diseases require long-term, cross-year protection, especially for gene therapies, which follow a one-time investment, long-term benefit model. The cost is concentrated now, while the benefits are spread over decades. Using an annual budget logic to manage cross-cycle costs cannot properly balance the financial burden.

Rare disease patients also need more than just drugs. They need early diagnosis, long-term rehabilitation, psychological support, education, employment, and many other kinds of help. However, health authorities handle health matters, insurance authorities handle insurance, and civil affairs authorities handle social welfare—each works in its own area, with little coordination. As a result, substantial resources are spent, but outcomes remain far from satisfactory [8, 9].

4. Bearing the cost of enhancing resilience: a different path

Given the above difficulties, it is necessary to move beyond a narrow focus on cost control and consider how the healthcare system can become more resilient in the face of long-term pressures such as rare diseases. Academics call this ability systemic resilience. In 2022, Emami and colleagues examined health system resilience from the perspective of rare diseases and defined it as the ability

DOI: 10.54254/2753-7064/2026.38116

of a system to absorb, adapt, or transform in response to shocks in order to maintain core functions [10]. Other studies have suggested that investing in rare disease programs can strengthen the resilience of the entire health system by developing professional expertise networks, patient and caregiver networks, and more integrated multidisciplinary care [8, 9]. Specifically, three problems can be addressed as follows:

First, if the government can know in advance which rare diseases are likely to appear, roughly how many patients there will be, and how much money will be needed, it can prepare ahead of time rather than be caught unprepared. This requires establishing a national, unified rare disease registration and monitoring system.

Second, it is a low-risk option. The entire burden should not be placed solely on health insurance. A special rare disease guarantee fund can be established, supplemented by commercial insurance, charitable assistance, and social donations, to create a multi-layered pool of funds. If one pool runs low, another can still help, so the whole system does not collapse.

Third, for every expensive drug, health insurance agencies and pharmaceutical companies can negotiate—for example, pay-for-performance (pay more if it works well, pay less or get a refund if it does not), or installment payments (spreading a million-dollar treatment cost over several years). This would reduce the cash flow pressure on health insurance. Together, these measures aim to move the healthcare system from passive cost bearing toward proactive planning, shared risk, and more flexible long-term financing.

5. Conclusion

Rare diseases have gradually shifted from being rare to being a common concern, which is a positive development brought about by medical progress. Investing in rare disease projects can enhance the resilience of the entire healthcare system by fostering multidisciplinary professional capabilities and improving medical quality. Specifically, addressing rare disease issues is itself upgrading the overall capacity of the medical system.

If existing methods are still used, the burden will only become more difficult. However, if this opportunity is to improve the institutional design of the healthcare economy, make it more flexible, multi-layered, and resilient, then not only rare disease patients will benefit, but society as a whole will also be better prepared for other public health emergencies. The shift from cost burden to systemic resilience is not just a change in wording, but people's mindset and behavior are transformed from passive resistance to active preparation.

References

[1] Angelis, A., Tordrup, D., & Kanavos, P. (2015). Socio-economic burden of rare diseases: A systematic review of cost of illness evidence. *Health Policy, 119*(7), 964–979. https://doi.org/10.1016/j.healthpol.2014.12.016

[2] Yang, G., Cintina, I., Pariser, A., Oehrlein, E., Sullivan, J., & Kennedy, A. (2022). The national economic burden of rare disease in the United States in 2019. *Orphanet Journal of Rare Diseases, 17*, 163. https://doi.org/10.1186/s13023-022-02299-5

[3] Schofield, D., Kraindler, J., Shrestha, R. N., Tan, O., West, S., Hart, N., Wall, L., Tan, L., Hewett, D., Villaret, L., Ma, A., Grigg, J. R., & Jamieson, R. V. (2026). The quality-of-life impacts and economic burden of X-linked retinitis pigmentosa caused by variants in RPGR. *Eye, 40*(9), 1302–1307. https://doi.org/10.1038/s41433-026-04532-y

[4] Schey, C., Milanova, T., & Hutchings, A. (2011). Estimating the budget impact of orphan medicines in Europe: 2010–2020. *Orphanet Journal of Rare Diseases, 6*, 62. https://doi.org/10.1186/1750-1172-6-62

[5] Angell, B., Wang, S., Gadsden, T., Moorthy, M., Malik, C., Barratt, J., Devuyst, O., Ulasi, I. I., Gale, D. P., Sengupta, A., Palagyi, A., Jha, V., & Jan, S. (2024). Scoping review of economic analyses of rare kidney diseases.

DOI: 10.54254/2753-7064/2026.38116

Kidney International Reports, 9(12), 3553–3569. https://doi.org/10.1016/j.ekir.2024.09.004

[6] Every Life Foundation for Rare Diseases. (2021). The national economic burden of rare diseases study. *Every Life Foundation for Rare Diseases*. https://everylifefoundation.org/burden-study/

[7] Chambers, J. D., Silver, M. C., Berklein, F. C., Cohen, J. T., & Neumann, P. J. (2020). Orphan drugs offer larger health gains but less favorable cost-effectiveness than non-orphan drugs. *Journal of General Internal Medicine, 35*(9), 2629–2636. https://doi.org/10.1007/s11606-020-05805-2

[8] Ferrelli, R. M., Gentile, A. E., De Santis, M., & Taruscio, D. (2017). Sustainable public health systems for rare diseases. *Annali dell'Istituto Superiore di Sanità, 53*(2), 170–175. https://doi.org/10.4415/ANN_17_02_16

[9] Hollick, R. J., James, W. R. G., Nicoll, A., Locock, L., Black, C., Dhaun, N., Egan, A. C., Fluck, N., Laidlaw, L., Lanyon, P. C., Little, M. A., Luqmani, R. A., Moir, L., McBain, M., & Basu, N. (2024). Identifying key health system components associated with improved outcomes to inform the re-configuration of services for adults with rare autoimmune rheumatic diseases: A mixed-methods study. *The Lancet Rheumatology, 6*(6), e361–e373. https://doi.org/10.1016/S2665-9913(24)00082-1

[10] Emami, S. G., Lorenzoni, V., Marinello, D., Palla, I., & Turchetti, G. (2022). OP17 Analyzing health system resilience from the perspective of rare diseases. *Value in Health, 25*(12), S389. https://doi.org/10.1016/j.jval.2022.09.1933