aposta
revista de ciencias sociales
ISSN 1696-7348

Application of the principles of universality and comprehensiveness in the treatment of rare diseases not supported by the Unified Health System (SUS) in Brazil

Aplicação dos princípios da universalidade e da integralidade no tratamento de doenças raras não amparadas pelo Sistema Único de Saúde (SUS) no Brasil

Aplicación de los principios de universalidad y exhaustividad en el tratamiento de enfermedades raras no apoyado por el Sistema Unificado de Salud (SUS) en Brasil

Angélica Silveira da Silva Moraes
Master of Laws (Tax Law)
Fundação Getulio Vargas, Brazil

Murillo Dias
Doctor in Business Administration
Rennes School of Business, France

Received in: 6/11/2026
Acepted in: 8/10/2026
DOI: 10.23900/ra.v24i118.1742

Citation format:
Moraes, A. S. da S., & Dias, M. (2026). Application of the principles of universality and comprehensiveness in the treatment of rare diseases not supported by the Unified Health System (SUS) in Brazil. *Aposta: Revista De Ciencias Sociales*, 24(118), e1742. https://doi.org/10.23900/ra.v24i118.1742

Abstract
Rare diseases are, by definition, pathologies that affect a small number of people but, together, profoundly impact a significant portion of the population and the health system. In Brazil, many of these diseases are not explicitly recognized in protocols or in the list of treatments offered by the SUS, which generates gaps in diagnosis, treatment, and social support. This research contributes to understanding how these gaps affect not only patients but also their families, the health system, and society in general. In addition, the study aims to highlight the ethical, social, and economic aspects that justify expanding support for these conditions, contributing to the debate on more equitable public policies aligned with the universal and comprehensive health defended by the Federal Constitution. By addressing this theme, the research seeks to collaborate with the strengthening of the right to health for all Brazilians, including those affected by rare diseases.
Keywords
Rare Diseases, Public Healthcare, Law, Diagnosis, Public Policies.

Resumo
As doenças raras são, por definição, patologias que afetam um número reduzido de pessoas, mas que, em conjunto, impactam profundamente uma parcela significativa da população e o sistema de saúde. No Brasil, muitas dessas doenças não são reconhecidas explicitamente em protocolos ou no rol de tratamentos ofertados pelo SUS, o que gera lacunas no diagnóstico, tratamento e suporte social. A relevância desta pesquisa reside em contribuir para o entendimento de como essas lacunas afetam não apenas os pacientes, mas também suas famílias, o sistema de saúde e a sociedade em geral. Além disso, o estudo visa destacar aspectos éticos, sociais e econômicos que justificam a ampliação do apoio a essas condições, contribuindo para o debate de políticas públicas mais equitativas, alinhadas

aposta
revista de ciencias sociales
ISSN 1696-7348

à saúde universal e integral defendida pela Constituição Federal. Ao abordar esta temática, a pesquisa busca colaborar com o fortalecimento do direito à saúde para todos os brasileiros, incluindo aqueles acometidos por doenças raras.
Palavras-chave
Doenças Raras, Saúde Pública, Direito, Diagnóstico, Políticas Públicas.

Resumen
Las enfermedades raras son, por definición, patologías que afectan a un pequeño número de personas, pero que, en conjunto, afectan profundamente a una parte significativa de la población y al sistema sanitario. En Brasil, muchas de estas enfermedades no están reconocidas explícitamente en los protocolos ni en la lista de tratamientos ofrecidos por el SUS, lo que genera lagunas en el diagnóstico, tratamiento y apoyo social. La relevancia de esta investigación radica en contribuir a la comprensión de cómo estas brechas afectan no solo a los pacientes, sino también a sus familias, al sistema sanitario y a la sociedad en general. Además, el estudio pretende destacar aspectos éticos, sociales y económicos que justifican la ampliación del apoyo a estas condiciones, contribuyendo al debate sobre políticas públicas más equitativas, alineadas con la salud universal e integral defendida por la Constitución Federal. Al abordar este tema, la investigación busca colaborar en el fortalecimiento del derecho a la salud para todos los brasileños, incluidos aquellos afectados por enfermedades raras.
Palabras clave
Enfermedades Raras, Sanidad Pública, Derecho, Diagnóstico, Políticas Públicas.

1. Introduction

The right to health, enshrined in article 196 of the Federal Constitution of 1988 as a right of all and a duty of the State (Brasil, 1988), represents one of the fundamental pillars of the Brazilian Democratic State of Law. Its implementation, however, faces structural challenges when confronted with pathologies that, by their very nature, escape ordinary standards of care — as is the case with rare diseases. The main objective of this work is to analyze the lack of application of the principles of universality and comprehensiveness in the treatment of rare diseases not supported by the Unified Health System (*Sistema Único de Saúde* - SUS), investigating the gaps between the Constitutional design of the health system and the concrete reality experienced by millions of Brazilians who are waiting for diagnosis and treatment, comparing with Portugal's healthcare system, as the units of analysis (Yin, 2004)once they share the institutional commitment to universal and comprehensive healthcare, to highlight differences and possible lessons for treatment.

This theme is justified by its social, legal, and human relevance (WHO, 2013). It is estimated that between 5,000 and 7,000 rare diseases are cataloged worldwide, affecting approximately 13 to 15 million Brazilians—a number higher than the population of many countries (SUS, 2026). Despite this expressiveness, these pathologies remain on the margins of structured public policies, either because of the low individual prevalence of each one, or because of the high costs of the so-called

orphan drugs. The recent inclusion of exome genetic sequencing in the SUS (2026) represents a significant advance, but it also reveals how far we still have to go. Investigating this reality contributes to the debate on more equitable public policies and to the strengthening of the right to health as a fundamental right for all.

The problem-situation that guides this research can be summarized in the following question: to what extent does the absence of incorporation of treatments for rare diseases by the SUS constitute a violation of the constitutional principles of universality and integrality, and what is the role of the Judiciary in correcting this omission? The central hypothesis is that the rigidity of the cost-effectiveness criteria adopted by CONITEC[1], combined with the slowness of the administrative processes, has generated a systematic exclusion of patients with rare diseases, transferring to the Judiciary the function of guaranteeing the existential minimum and the dignity of these patients.

As for the methodology, the present work adopts a qualitative, exploratory-descriptive approach, combining three complementary strands. The first is bibliographic and doctrinal research, analyzing works by authors such as José Afonso da Silva, Ingo Wolfgang Sarlet, Ana Paula de Barcellos, Maria Sylvia Zanella Di Pietro, and Fernando Facury Scaff, among others. The second is jurisprudential research, examining precedents of the Federal Supreme Court (Themes 6, 500, and 793) and the Superior Court of Justice (Theme 106), as well as paradigmatic decisions such as the Zolgensma case. The third is comparative research, analyzing the Portuguese model for treating rare diseases through the National Health Service (SNS) and the Exceptional Use Authorization (EUA), as a counterpoint to the Brazilian reality.

The general objective of this work is to analyze the tension between the constitutional principles of universality and integrality of the SUS and the exclusion of treatments for rare diseases, and to evaluate the role of judicialization as a mechanism for realizing these rights. As specific objectives, it is intended to: (i) examine the constitutional and legal framework of the right to health and the structure of the SUS; (ii) investigate the specific challenges of rare diseases and orphan drugs, including the tension between the reserve of the possible and the existential minimum; (iii) analyze the jurisprudence of the higher courts on the provision of non-incorporated treatments; (iv) to understand the phenomenon of the judicialization of health and its impacts; and (v) compare the

[1] The acronym CONITEC stands for National Commission for the Incorporation of Technologies in the Unified Health System (*Comissão Nacional de Incorporação de Tecnologias no SUS*). CONITEC is the National Commission for the Incorporation of Technologies in SUS, created by Law nº 12.401/2011 and regulated by Decree nº 7.646/2011. Its role is to advise the Ministry of Health by assessing scientific evidence, cost-effectiveness, and budget impact before deciding which technologies are adopted in Brazil's public healthcare system.

Brazilian experience with the Portuguese model, extracting lessons that can improve national public policy.

2. Theoretical Framework

2.1 Right to Health and the Unified Health System (SUS)

Health is a subjective public right and, therefore, the object of State intervention, since the State must guarantee it to safeguard the physical and mental integrity of its citizens (Brasil, 1988). The guarantee of health as a public right is, therefore, a matter of life for the State, which must decide whether or not to make available the most advanced technologies or whether to intervene in a given case. Where the most advanced technologies or State intervention are excluded, there is, in the end, denial of life itself in the case of rare diseases (Brasil, 1988).

This, however, is by no means a given, for there is an ongoing struggle between individual needs and available funds. Justice Gilmar Mendes, in his opinion in STA 175/CE (Brasil, 2010), observed that the intervention of the judiciary does not amount to an invasion of the government's area of action, for it is an essential tool of utmost importance for the correction of the State's failures and for the fulfillment of the health promises enshrined in the Constitution.

This duty of the State to guarantee the right to health is founded upon the ethical principle of human dignity. According to Sarlet (2017), human dignity demands that the State guarantee the minimum material conditions for a decent life. In health, the State's omission or lack of resources is not sufficient to deny fundamental rights to life and physical integrity.

Finally, according to Silva (2014); Sarlet (2017); and Mendes (2020) consider that the fundamental right to health must be an imperative of survival, which must be respected by all means of obstacle (including mere bureaucratic ones), to safeguard the essential core of the right to right to life of patients with rare diseases, through an even more incisive constitutional protection against claims of scarcity of resources.

2.2 Feature and Structure of the SUS

The institutionalization of the Unified Health System (SUS) represents the greatest social achievement in Brazilian history, made possible by its enactment (Brasil, 1990). The creation of this system marked the definitive transition from the old model managed by INAMPS — National Institute of Medical Assistance of Social Security (*Instituto Nacional de Assistência Médica da Previdência Social*), which conditioned access to health to social security contributions and formal employment relationships — to a model of universal, democratic and free access.

The structure of the SUS is guided by administrative decentralization and the hierarchization of services. From the perspective of Administrative Law, Maria Sylvia Zanella Di Pietro (2020) teaches that the SUS operates under a public-law regime, in which the Public Administration has an inescapable duty to ensure the continuity and efficiency of the service provided. For the author, network organization and decentralization are instruments to achieve the necessary capillarity, but they cannot serve as bureaucratic obstacles that prevent citizens from accessing essential treatment.

However, the materialization of this structure often clashes with budget management and the concept of the "reserve of the possible". At this point, Fernando Facury Scaff's doctrine analyzes the system from the perspective of Financial Law, arguing that the budget should be read in the light of the Constitution, and not the other way around. The financial viability of the system and the limits of the state's duty are often subject to judicial scrutiny. The jurisprudence of the Federal Supreme Court (STF) consolidated, through Topic 793 (Brasil, 2010), the joint and several liability of the federated entities (Union, States and Municipalities) in the provision of health services. This understanding ensures that the Federal Government, which has greater budgetary capacity, is held responsible for the cost of high-cost treatments, preventing the financial collapse of smaller entities.

In addition, the Superior Court of Justice (STJ), through Topic 106 (Superior Court of Justice, 2018), established cumulative requirements for the State to be required to provide medicines not incorporated in the RENAME - National List of Essential Medicines (*Relação Nacional de Medicamentos Essenciais*), being required: (a) Proof, by means of a substantiated medical report, of the indispensability or necessity of the medication; (b) The patient's financial inability to bear the cost of the treatment; and (c) Existence of registration of the drug with ANVISA[2].

2.3 The Creation of CONITEC and the Legal Framework for the Incorporation of Technologies in the SUS

The incorporation of health technologies within the scope of the Unified Health System (SUS) underwent a profound institutional transformation with the enactment of *Law No. 12,401, of April 28, 2011*, which amended Law No. 8,080/1990 to create the National Commission for the Incorporation of Technologies in the SUS (CONITEC). Before CONITEC's creation, decisions about which drugs,

[2] ANVISA is the Brazilian Health Regulatory Agency (National Health Surveillance Agency), the federal authority responsible for regulating and supervising health-related products and services in Brazil. Much like the FDA in the United States or the EMA in Europe, ANVISA oversees the approval and registration of medicines, vaccines, medical devices, and food products, ensuring their safety, efficacy, and quality before they reach the public. It plays a crucial role in protecting public health. It works closely with CONITEC, since only technologies registered with ANVISA can be considered for incorporation into the Unified Health System (SUS).

procedures, and equipment the SUS would offer were made in a fragmented manner by different instances, without unified criteria based on scientific evidence and cost-effectiveness analysis.

CONITEC, supported by the Department of Management and Incorporation of Health Technologies (DGITS), advises the Ministry of Health on decisions about the incorporation, exclusion, or alteration of technologies in the SUS. The decision-making process is guided by three fundamental pillars: efficacy and safety (scientific evidence), economic analysis (cost-effectiveness), and budgetary impact. The commission includes representatives from various sectors, including SUS managers, specialists, medical societies, and, innovatively, members of civil society and the National Health Council, giving the process democratic legitimacy.

Decree No. 7,646/2011 regulated Law No. 12,401/2011 (Brasil, 2011) and established the procedural flow, setting maximum deadlines of 180 days (extendable for another 90) for the conclusion of the analyses. This timeframe, although reasonable for conventional technologies, is challenging in the context of rare diseases, where patient urgency often conflicts with bureaucratic slowness.

2.4 Rare Diseases and the Challenges of Access to Treatments

Rare diseases in Brazil are a confluence of medicine, law, and the economy, shaping public health policies and programs. Of the estimated 5,000 to 7,000 diseases that affect people worldwide and are considered rare (up to 65 individuals per 100,000 population), 13-15 million people are affected in Brazil, most of whom do not receive timely and adequate treatment. To provide comprehensive care for people with rare diseases, the Ministry of Health implemented the National Policy for Comprehensive Care for People with Rare Diseases (Portaria nº 199/2014), later consolidated in Portaria nº 2/2017. The care framework for people with rare diseases is organized around two axes: genetic conditions and non-genetic conditions. Iriart et al. (2023) described the “diagnostic labyrinth” that patients with rare diseases experience through repeated contacts with health services, misdiagnoses, and delays in diagnosis that negatively affect their health and favor disease progression.

Recent breakthroughs for patients with rare diseases in Brazil came with the implementation of free exome sequencing by the Unified Health System (SUS). Genetic tests can identify mutations responsible for diseases, and recent studies showed that exome sequencing can identify 85% of mutations in rare diseases.

The main difficulty in treating rare diseases is the cost of some drugs, called pharmaceutical products for the treatment of rare diseases, or "orphan" in English. These drugs are difficult for

pharmaceutical companies to market because the number of patients with the disease is very small, and the cost of research and development for a new drug is often very high. Once the state grants registration of the drug, the government can decide whether the treatment should be provided to the patient, based on cost-effectiveness criteria. In 2022, CONITEC (National Council for the Incorporation of Technologies) established cost-effectiveness limits for disease treatment, which are applied worldwide, such as by the NICE (National Institute for Health and Clinical Excellence) in the United Kingdom. However, for high-cost drugs, long-term treatment and definitive "cure", patients are often excluded from treatment for cost reasons, and the only way left for the patient is to judicialize the treatment, as reported by Iriart et al. (2023).

Therefore, the situations presented above raise another question, fundamentally constitutional. In effect, the State always resorts to the “reserve of the possible” argument, i.e., to the limits imposed by its social spending capacity. However, it is possible to argue, based on social rights, that there are limits to what can be spent on health, and that the “existential minimum” must be immediately protected. To that end, a series of essential conditions for a human life to be considered to be fully developed are reserved. As mentioned, Barcellos (2011) first introduced this concept, followed by Sarmento (2016). The main condition for a series of others to be protected is that they are immediately indispensable, regardless of the limitations of the public budget. Therefore, to protect them, it is necessary to resort to judicial intervention when existing public policies are insufficient to incorporate the necessary technologies.

Brazil has recently advanced significantly in several key areas of rare diseases, but many challenges remain, and most must be addressed through fairer public policy for these patients. Thus, we conclude that recent advances also point to significant current challenges, and that much work remains needed in service delivery, diagnostics, inclusion of orphan drugs, and the judicialization of access to essential treatment.

3. Methodology

This research is qualitative and exploratory-descriptive in nature and was developed as a descriptive multiple case study. As Yin (2004) states, case study research is the most appropriate method for investigating complex social phenomena in their context. This study analyzed two health systems: the Brazilian Unified Health System (SUS) and the Portuguese National Health Service (SNS). In both systems, we analyzed and compared the treatment of rare diseases.

A multiple case study research design was chosen because it allows within-case analysis (similarities and differences within each context) and between-case analysis (similarities and

differences between contexts), enabling analytical generalization (Yin, 2004). A descriptive study is also appropriate because this is the first descriptive study to describe in depth the problems that patients with rare diseases face in two different countries.

To validate the results, we collected data using three strategies. First, we conducted a documentary analysis of the legal framework, public policies, and official reports of both countries. Second, we conducted a jurisprudential analysis of decisions by the Supreme Court of Brazil (STF) and the Superior Court of Justice (STJ) related to access to treatment for rare diseases. Finally, a bibliographical review of the main doctrinal contributions of constitutional and administrative law to the studied topic was conducted.

4. Judicialization and the Cost of the Absence of Public Policies

The judicialization of health reveals itself as a symptom of a deep structural flaw: the insufficiency of public policies and the slowness of administrative processes. Studies by Iriart et al. (2023) show that patients are startled by uncertainty.

4.1 Doctrinal and Critical View on the Judicialization of Health

The state's omission to fund orphan drugs under the argument of reserving the possible is criticized by Scaff (2020), who argues that it is precisely where the market fails that the SUS should act with greater vigor.

4.2 Judicialization as a Reflection of the Delay in the Incorporation of Technologies

The gap between patients' needs and the time required for CONITEC evaluation has created a growing phenomenon of judicialization. Between 2012 and 2023, CONITEC analyzed hundreds of incorporation requests, and studies published in the journal Health Technology Assessment in the Brazilian National Health System (PMC, 2024) indicate that a significant portion of recommendations for exclusion or non-incorporation concerned rare-disease technologies.

In this context, Topic 106 of the STJ establishes cumulative requirements that allow the Judiciary to determine the supply of medicines not incorporated by CONITEC: (a) proof of the indispensability of the medicine; (b) the patient's lack of financial capacity; and (c) existence of registration with ANVISA. Consolidated jurisprudence recognizes, therefore, that the absence of incorporation by CONITEC is not an absolute obstacle to access, especially when the right to life and the dignity of the human person are at stake.

The relationship between CONITEC and the right to health of rare disease patients is analyzed by Ingo Wolfgang Sarlet (2017, p.112) argues that administrative bodies, such as CONITEC, cannot replace the essential core of the fundamental right with purely economic criteria. For Sarlet (2017), technical evaluation is welcome and necessary, but it cannot override the existential minimum when the patient's life is at imminent risk.

In addition, Di Pietro (2020) offers the basis for questioning the legality of any omissions by CONITEC. If the commission exceeds the legal deadlines of 180/270 days without a reasoned decision, or if the motivation for the decision disregards the patient's individual clinical condition, there is room for judicial control. The theory of "losing a chance" also applies here: each day of delay in incorporating an effective treatment represents a lost chance to avoid irreversible sequelae or death itself. Furthermore, when the patient, in the tireless search for effective treatment, encounters barriers to exercising his rights, there is nothing left but judicialization as the last way to guarantee the minimum of dignity.

5. Results and Discussions

5.1 Analysis of Experiences in the Treatment of Rare Diseases: a Comparative Study Between Brazil and Portugal

The choice for this comparison is based on the profound influence that the Portuguese constitutional doctrine exerts on the Brazilian legal system, especially with regard to the protection of social rights and the realization of the dignity of the human person. The analysis seeks to identify how different administrative and judicial models respond to the challenge of very high-cost therapies for rare diseases.

5.2 The Zolgensma Case and the Paradigmatic Shift in Brazil

The analysis of the efficacy of the right to health for patients with rare diseases in Brazil finds a fundamental milestone in the case of the drug Zolgensma (*Onasemnogene abeparvovec*), indicated for the treatment of Spinal Muscular Atrophy (SMA) Type 1. This drug, considered one of the most expensive in the world, synthesizes the tension between biotechnological innovation and the financial sustainability of the Unified Health System (SUS).

Historically, access to this gene therapy was obtained exclusively through the courts. The judicialization of Zolgensma in Brazil began with a historic injunction issued in October 2020 by Minister Napoleão Nunes Maia Filho of the Superior Court of Justice (STJ) (STJ, 2020). At the time, the magistrate determined that the Union should deposit approximately R$ 6.7 million to complement

the acquisition of the medicine for a child, basing the decision on the urgency of the treatment and the protection of life. Another high-profile case was that of the child Antonella Ferreira Salistiano da Silva (Case No. 5007714-02.2021.4.03.6100, pending in the TRF3), which exemplifies the complexity of high-cost demands processed in the federal courts.

The magnitude of this phenomenon was quantified by the census study by Kretzschmar et al. (2024), published in the Journal of Public Health. The research identified 136 lawsuits involving Zolgensma against the Ministry of Health, revealing that in 113 cases (83%) the decision favored the patient. The financial impact of these decisions reached the figure of 'R$ 944.8 million' to the public coffers, showing that judicialization has become the main means of access before official incorporation.

The paradigmatic turning point occurred in 2022, when CONITEC recommended incorporating the drug under an unprecedented risk-sharing agreement, where payment is conditional on the clinical results observed. This movement culminated in the agreement ratified by the Federal Supreme Court in 2024, within the scope of Topic 1234, establishing that judicialization should be a last resort and prioritizing the expeditious administrative route. This is what Barroso (2007) defines as necessary judicialization, where the Judiciary intervenes to drive the Public Administration towards efficiency.

5.3 The Portuguese System and Comparative Analysis

The comparative research between Brazil and Portugal was carried out through a bibliographic and documentary analysis of academic and official sources from both countries (Lisboa & Rocha, 2024), examining structural differences between the two health systems and their respective patterns of judicialization. Additionally, Vidal (2019) analyzes the effectiveness of public policies for rare diseases in Portugal; de Silva et al. (2019) promote a comparative analysis of health policies between the two countries, which brings together systematized data on the Portuguese reality. On the normative level, Order No. 2584/2014, which established the National Strategy for Rare Diseases (ENDR), and the Decree-Law

No. 176/2006 (Statute of Medicines of Portugal), which is the basis for the Authorization of

Exceptional Use (EUA). The Portuguese doctrinal framework was based on the works of Miranda (2017) and Canotilho (2015), whose theses on the right to health as a right to benefits and the prohibition of social regression provided the theoretical basis for the comparison.

In Portugal, the National Health Service (SNS) deals with rare diseases under the aegis of the National Strategy for Rare Diseases (ENDR), established by Order No. 2584/2014. The Portuguese

management presents a relevant structural difference from the Brazilian one: the centrality and agility of the National Authority for Medicines and Health Products (INFARMED). While Brazil faces delays in updating the Clinical Protocols and Therapeutic Guidelines (PCDT), Portugal makes extensive use of the Exceptional Use Authorization (EUA).

From Miranda's (2017) perspective, the right to health is a right to benefits that objectively binds the State. This link is reinforced by the thesis of the Prohibition of Social Regression, defended by Canotilho (2015), which prevents the emptying of rights already conquered under the simple allegation of resource scarcity.

In summary, the comparison between Portugal and the new system of Topic 1234 in Brazil reveals that the main Brazilian bottleneck is not the lack of legislation, but the need for greater administrative efficiency. The lesson from the Portuguese experience is that protecting minorities with rare diseases is the definitive test of democracy, requiring that the reservation of the possible be interpreted in light of the existential minimum and the dignity of the human person.

6. Implications

This research explores implications across several areas where prior research has been referenced. Firstly, within the area of negotiation. Studies on the negotiation of physician shifts (Alevato & Dias, 2026) and on the management of infection costs (Saliba & Dias, 2025) provide frameworks for negotiating risk-sharing agreements to finance orphan drugs. Secondly, within the area of leadership. Studies on the management of home care (De Jesus & Dias, 2026) and transformational project leadership (Pan & Dias, 2024) demonstrate the importance of governance and organizational capacity to manage policies related to rare diseases. A third area of implication is understanding the Brazilian healthcare system (Pan & Dias, 2024) and the need to balance universality with fiscal sustainability. Other studies on negotiation and healthcare management (e.g., Dias, 2018) show how judicialization can be reframed as a failure of negotiation between stakeholders. By integrating negotiation theory, leadership studies, and systemic studies, health law and policy can reduce the need for judicialization, increase administrative efficiency, and promote equity in the treatment of rare diseases.

7. Conclusions

The present research demonstrated that the right to health of patients with rare diseases in Brazil operates in a zone of permanent tension between the maximum effectiveness of fundamental rights and the State's budgetary limitations. The analysis of the constitutional framework and the

structure of the SUS revealed that, although the system is designed for universality, CONITEC's administrative practice, guided by rigid cost-effectiveness thresholds, ends up excluding vital technologies and pushing citizens toward the judicial route.

It is concluded that judicialization, far from being a pathology of the system, acts as a mechanism for correcting administrative omissions. The Zolgensma case and the empirical data presented show that pressure from the Judiciary was the catalyst for adopting innovative management models, such as risk sharing. However, excessive dependence on the Judiciary generates uncertainty and inequality, since not all patients have the same access to justice.

The experience in Portugal offers a valuable lesson: administrative efficiency, materialized in mechanisms such as the Authorization for Exceptional Use (EUA), can reconcile therapeutic urgency with budgetary control, reducing the need for judicial intervention. For Brazil, the path points to the need to make CONITEC's criteria for rare diseases more flexible and to strengthen the administrative route, ensuring that the "reserve of the possible" never annihilates the "existential minimum" and the dignity of the human person.

8. Future Research

Future researchers are encouraged to go beyond the cost-effectiveness threshold currently used in many analyses and estimate the broader social costs of untreated rare diseases and their hospitalizations, as well as losses in productivity and human suffering. The budgetary analysis thus becomes compatible with Scaff (2020). A comparative analysis can be developed, including other institutions worldwide, for example, NICE in the UK, to identify the ethical principles integrated into different health care systems for resource allocation, such as the “existential minimum” (Barcellos, 2011; Sarmento, 2016).

Finally, the judicialization of health issues remains a subject that has not been studied in depth. As previously mentioned, the Brazilian judiciary (Barroso, 2007; Di Pietro, 2020) has established ways for patients to access drugs not yet incorporated into the universal health system (SUS), as well as to deny access to them. On the other hand, administrative Exceptional Use Authorizations already in place in Portugal (Vidal, 2019; Lisboa & Rocha, 2024) may be better suited to protect patients’ rights while ensuring the sustainability of health systems.

9. References

Alevato, R., Dias, M. (2026) Redesigning Remuneration Models for Physician Shifts: A Brazilian Healthcare Negotiation Case. *International Journal of Advance Research, 14*(2),1510-17, https://doi.org/10.5281/zenodo.18770037

Barcellos, A. P. (2011). *A eficácia jurídica dos princípios constitucionais: O princípio da dignidade da pessoa humana* (3ª ed.). Renovar.

Barroso, L. R. (2007). Da falta de efetividade à judicialização excessiva: Direito à saúde, fornecimento gratuito de medicamentos e parâmetros para a atuação judicial. *Revista Jurídica da Presidência, 9*(86).

Brasil. (1988). *Constituição da República Federativa do Brasil de 1988*. Presidência da República.

Brasil. (1990). *Lei nº 8.080, de 19 de setembro de 1990: Dispõe sobre as condições para a promoção, proteção e recuperação da saúde*.

Brasil. (2011a). *Lei nº 12.401, de 28 de abril de 2011: Dispõe sobre a assistência terapêutica e a incorporação de tecnologia em saúde no âmbito do SUS*.

Brasil. (2011b). *Decreto nº 7.646, de 21 de dezembro de 2011: Regulamenta a Lei nº 12.401/2011 e dispõe sobre a CONITEC*.

Brasil. Ministério da Saúde. (2014). *Portaria nº 199, de 30 de janeiro de 2014: Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras*.

Brasil. Ministério da Saúde. (2017). *Portaria de Consolidação nº 2, de 28 de setembro de 2017. Anexo IV*.

Brasil. Ministério da Saúde. (2022, setembro). *CONITEC aprova proposta de uso de limiares de custo-efetividade (LCE) nas decisões em saúde*.

Brasil. Ministério da Saúde. (2026). *Doenças raras: Exame no SUS reduz tempo de diagnóstico para 6 meses*. https://www.gov.br/saude/pt-br/assuntos/noticias/2026/fevereiro/ministerio-da-saude-inclui-exame-de-alta-tecnologia-no-sus-para-doencas-raras-espera-das-familias-por-diagnostico-reduz-de-7-anos-para-seis-meses (gov.br in Bing)

Brasil. Superior Tribunal de Justiça. (2018). *REsp 1.657.156/RJ. Relator: Min. Benedito Gonçalves. Primeira Seção. Julgado em 25/04/2018. Tema 106. DJe 04/05/2018*.

Brasil. Supremo Tribunal Federal. (2010). *STA 175 AgR/CE. Relator: Min. Gilmar Mendes. Tribunal Pleno. Julgado em 17/03/2010*.

Brasil. Supremo Tribunal Federal. (2019a). *RE 657.718/MG. Relator: Min. Marco Aurélio. Plenário. Julgado em 22/05/2019. Tema 500*.

Brasil. Supremo Tribunal Federal. (2019b). *RE 855.178/SE. Relator: Min. Luiz Fux. Plenário. Julgado em 23/05/2019. Tema 793*.

Brasil. Supremo Tribunal Federal. (2024). *RE 566.471/RN. Relator: Min. Marco Aurélio, Redator p/ acórdão: Min. Luís Roberto Barroso. Plenário. Julgado em 26/09/2024. Tema 6*.

Canotilho, J. J. G. (2015). *Direito constitucional e teoria da constituição* (7ª ed.). Almedina.

De Jesus, A.P., Dias, M. (2026). Strategic Negotiation for Leadership Development in Home Care Management. *IPHO-Journal of Advance Research in Medical & Health Science, 4*(2), 01–14. https://doi.org/10.5281/zenodo.18722243

Di Pietro, M. S. Z. (2020). *Direito administrativo* (33ª ed.). Forense.

Iriart, J. A. B., et al. (2023). Da busca pelo diagnóstico às incertezas do tratamento: Desafios do cuidado para as doenças genéticas raras no Brasil. *Ciência & Saúde Coletiva, 28*(4).

Kretzschmar, G., et al. (2024). Judicialização do Zolgensma no Brasil: Estudo censitário de 136 processos. *Revista de Saúde Pública, 58*.

Lisboa, L. V., & Rocha, D. D. P. M. (2024). Judicialização da saúde no Brasil e em Portugal: Uma análise comparativa. *Mediação, 19*(1), 41–58.

Mendes, G. F., & Branco, P. G. G. (2020). *Curso de direito constitucional* (15ª ed.). Saraiva.

Miranda, J. (2017). *Manual de direito constitucional* (Tomo IV, 6ª ed.). Coimbra Editora.

Nunes Júnior, V. S. (2021). *Direito à saúde e a reserva do possível*. Saraiva.

Pan, J. Dias, M. (2024) The Impact of Transformational Leadership on Project Management Success in the Brazilian Private Healthcare System. *Open Journal of Business and Management, 12*, 2553-2570. https://doi.org.10.4236/ojbm.2024.124132

Pan, J., Dias, M. (2024) Healthcare in Brazil: An Overview. *British Journal of Multidisciplinary and Advanced Studies: Health and Medical Sciences 5*(2),69-89, https://doi.org.10.37745/bjmas.2022.0475

Pinheiro, F. S., Borges, S. S., & Rodrigues, F. A. (2024). Avaliação de tecnologias em saúde no Sistema Único de Saúde: Perfil das recomendações de exclusão na Conitec, de 2012 a 2023. *Epidemiologia e Serviços de Saúde, 33*, e20240057.

Portugal. (2021). *Livro branco sobre doenças raras*. Aliança Portuguesa de Associações das Doenças Raras.

Saliba, G. & Dias, M. (2025) Health Cost Negotiation: Case Study of Surgical Site Infection. *GPH-International Journal of Biological & Medicine Science, 8*(8), 14-23. https://doi.org/10.5281/zenodo.17200069

Sarlet, I. W. (2017). *A eficácia dos direitos fundamentais* (13ª ed.). Livraria do Advogado.

Sarmento, D. (2016). A proteção judicial dos direitos sociais: Alguns parâmetros ético-jurídicos. In C. P. Souza Neto & D. Sarmento (Coords.), *Direitos sociais: Fundamentos, judicialização e direitos sociais em espécie* (2ª ed.). Lumen Juris.

Saunders, M., Lewis, P., & Thornhill, A. (2009). *Research methods for business students* (5th ed.). Pearson Education.

Scaff, F. F. (2020). *Direito financeiro* (7ª ed.). Saraiva.

Schwartz, I. V. D., et al. (2010). Medicamentos de alto custo para doenças raras no Brasil: O exemplo das doenças lisossômicas. *Ciência & Saúde Coletiva, 15*(supl. 3), 3443–3454.

Silva, J. A. (2014). *Curso de direito constitucional positivo* (37ª ed.). Malheiros.

Silva, M. A., et al. (2019). Doenças raras no Brasil e em Portugal: Uma análise comparada das políticas de saúde. *Cadernos de Saúde Pública, 35*(2).

Vidal, V. L. (2019). A efetividade das políticas públicas para doenças raras em Portugal. *Revista Portuguesa de Saúde Pública, 37*(1).

World Health Organization. (2013). *Priority medicines for Europe and the world: A public health approach to innovation. WHO background paper 6.19 – Rare diseases*. WHO.

Yin, R. K. (2004). *The case study anthology*. Sage.