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Public Policies for Rare Diseases in Brazil: Regulatory Path, Challenges in Research and Access to Orphan Drugs

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Abstract

Introduction: Rare diseases, which affect millions globally, are chronic and often genetic conditions, frequently manifesting in childhood. In Brazil, they are considered rare when they affect up to 65 in every 100,000 people. With around 7,000 types and impacting 5-6% of the world's population, including 13 million Brazilians, they require multidisciplinary management. High morbidity and mortality rates and the lack of a cure mean that treatment focuses on improving quality of life. **Objective:** To describe the construction and implementation of official ethical and health regulations on rare and ultra-rare diseases, with the purpose of identifying barriers and challenges in research development and access to orphan drugs in the Brazilian context. **Methods:** An exploratory study that adopted a qualitative approach, focusing on documentary analysis and narrative review of the literature. Official websites were visited and literature was reviewed, using qualitative and quantitative approaches to evaluate the National Policy for Comprehensive Care for People with Rare Diseases (PNAIPDR), ethical and health regulations, and access routes to orphan drugs in Brazil. **Results:** The study reveals that the implementation of the PNAIPDR was hampered by a lack of professionals, reference centers, and financial incentives. Health regulations accelerated the processing of orphan drugs, which boosted the number of studies on rare diseases, which is still in its infancy in Brazil, reflecting on effective access and the incorporation of new technologies into the SUS. Research ethics showed a commitment to protecting participants through CNS regulations, which led to the approval of Law No. 14,874/2024 in August 2024. **Conclusion:** Brazil has made progress in regulation, but the effective implementation of policies faces structural and financial challenges. Strengthening research infrastructure, streamlining the incorporation of drugs into the SUS, and exploring alternative financing options are crucial to ensuring equitable access to treatments for rare and ultra-rare diseases.

Keywords: rare disease, health policy, health systems, orphan drug production, access to essential medicines and health technologies.

Políticas públicas para doenças raras no Brasil: Caminho Regulatório, Desafios na Pesquisa e Acesso a Medicamentos órfãos

Resumo

Introdução: Doenças raras, que afetam milhões globalmente, são condições crônicas e muitas vezes genéticas, manifestando-se frequentemente na infância. No Brasil, são consideradas raras quando atingem até 65 em cada 100 mil pessoas. Com cerca de 7.000 tipos e impactando 5-6% da população mundial, incluindo 13 milhões de brasileiros, exigem manejo multidisciplinar. A alta morbimortalidade e a falta de cura tornam o tratamento focado em melhorar a qualidade de vida. **Objetivo:** Descrever a construção e implementação das normativas éticas e sanitárias oficiais sobre doenças raras e ultrarraras, com o propósito de identificar suas barreiras e desafios no desenvolvimento de pesquisa e no acesso a medicamentos órfãos o cenário brasileiro. **Métodos:** Estudo exploratório que adotou uma abordagem qualitativa, com foco na análise documental e revisão narrativa da literatura. Foram visitados sítios oficiais e revisão de literatura, utilizando abordagens qualitativas e quantitativas para avaliar a Política Nacional de Atenção Integral às Pessoas com Doenças Raras (PNAIPDR), regulamentações éticas e sanitárias, e as vias de acesso a medicamentos órfãos no Brasil. **Resultados:** O estudo revela que a PNAIPDR teve sua implementação dificultada pela falta de profissionais, centros de referência e incentivos financeiros. Que as regulamentações sanitárias aceleraram a tramitação para medicamentos órfãos, o que impulsionou o quantitativo de pesquisas com doenças raras, que ainda se apresenta incipiente para o Brasil, refletindo no acesso efetivo e na incorporação de novas tecnologias no SUS. A ética em pesquisa apresentou um compromisso com a proteção dos participantes por meio das normativas do CNS e que culminou na homologação em agosto de 2024 da Lei nº 14.874/2024. **Conclusão:** o Brasil progrediu na regulamentação, mas a efetiva implementação das políticas enfrenta desafios estruturais e financeiros. Fortalecer a infraestrutura de pesquisa, agilizar a incorporação

de medicamentos no SUS e explorar alternativas de financiamento são cruciais para garantir o acesso equitativo a tratamentos para doenças raras e ultrarraras.

Palavras-chave: doenças raras, política de saúde, sistemas de saúde, produção de medicamentos órfãos, acesso a medicamentos essenciais e tecnologias de saúde.

Introduction

Rare diseases have increasingly been recognized as a global public health issue, including in Brazil. Although these conditions individually affect a small portion of the population, they collectively impact millions of people worldwide, generating a significant public health burden. According to the World Health Organization (WHO), in Brazil, a disease is considered rare if it affects up to 65 people per 100,000 inhabitants, that is, approximately 1.3 cases per 2,000 individuals. Most rare diseases are of genetic origin, involving alterations in genes or chromosomes, and predominantly manifest in childhood, accounting for approximately 75% of cases. In addition, some have infectious, allergic, degenerative, toxic, or proliferative causes (Figure 1).

Globally, it is estimated that more than 400 million people live with a rare disease, corresponding to 5–6% of the world population, with approximately 7,000 distinct conditions. In Brazil, the Ministry of Health estimates that around 13 million people are affected by these conditions. Most of these diseases are chronic, progressive, and disabling, with high morbidity and mortality. As cures are rare, clinical management requires a multidisciplinary approach, involving various medical specialties and complementary therapies, aiming to alleviate symptoms, slow disease progression, and improve patients' quality of life.

In developed countries, data indicate that approximately 30% of patients with rare diseases die during childhood, and about 25.7% of these diseases are fatal before the age of five. Only 37.5% of these patients are able to lead a life considered normal.[1] Although few equivalent data exist for developing countries, evidence points to a potentially more severe situation due to diagnostic difficulties, limited access to treatments, and higher overall rates of infant mortality. Organizations such as the WHO and Rare Diseases International (RDI) emphasize the importance of understanding the impact of rare diseases in resource-limited settings.

Within rare diseases, ultra-rare diseases stand out, defined in Brazil by CNS Resolution No. 563/2017 as those with an incidence of one case per 50,000 inhabitants or fewer. These diseases often originate from "de novo" genetic mutations that arise spontaneously in important regions of the genome and are identified through genetic sequencing.[2]

Despite advances in knowledge, there remains a scarcity of scientific evidence on rare diseases, and many cases lack specific treatments. Nevertheless, there are care strategies and interventions aimed at improving quality of life and extending patient survival. Orphan drugs—those intended for the treatment or diagnosis of these diseases—play a fundamental role, but their access in Brazil is marked by challenges. The Brazilian Unified Health System (SUS) seeks to ensure comprehensive care for these patients, yet it faces obstacles such as high drug costs, the absence of specific clinical protocols, and inconsistent public funding, which hinder equitable access.[3,4]

In Brazil, the commercialization of medicines depends on registration with the National Health Surveillance Agency (ANVISA), which certifies safety, quality, and efficacy. After regulatory approval, the drug price must be authorized by the Chamber for the Regulation of the Pharmaceutical Market (CMED).[5] Additionally, research on rare diseases plays a crucial role in the development of new technologies and treatments. In Brazil, such research is regulated by bodies such as ANVISA, responsible for overseeing clinical trials, and the National Research Ethics Committee (CONEP), which reviews and approves protocols to protect participants.

The incorporation of new medicines into the Brazilian Unified Health System (SUS) goes through the National Commission for the Incorporation of Technologies in SUS (CONITEC), which evaluates efficacy, safety, and cost-effectiveness, and develops a Clinical Protocol and Therapeutic Guidelines (PCDT) for each approved medicine. However, this process is slow and does not meet patients' urgent needs, which has driven the judicialization of access to orphan drugs.

The development of drugs for rare diseases faces specific challenges: low prevalence hinders recruitment for robust clinical trials; phenotypic and genotypic heterogeneity complicates therapy development; the scarcity of preclinical data, biomarkers, and clinical outcomes complicates efficacy evaluation; and the lack of historical precedents limits understanding of disease progression and treatment response. All of these factors make the process costly and complex.[6]

The National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR), established in 2014 by the Ministry of Health, guarantees the right to timely access to appropriate diagnoses and treatments according to the needs of each patient. However, access pathways to orphan drugs in Brazil are diverse and generate different realities for patients, highlighting disparities and barriers within the system (Figure 2).

The principles of universality and comprehensiveness, present in PNAIPDR, obligate the State to provide special and non-discriminatory care to this population, using the best available scientific knowledge. Nevertheless, financial and structural constraints challenge the full implementation of these principles, considering the high cost of treatments and the complexity of public policies.

Access to orphan drugs in Brazil is a process marked by advances and setbacks, reflecting a global context in which the right to health is recognized as a fundamental human right. Although the Ministry of Health has increased investments in rare disease services, rising from BRL 23 million in 2022 to BRL 27.3 million in 2023, structural barriers persist, limiting broad and equitable access to innovative treatments.[7]

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This scenario highlights the urgent need to deepen understanding of the impact of Brazilian ethical and health regulations—from the National Policy for Pharmaceutical Assistance (PNAF, 2007) to the General Law on Research with Human Beings (Law No. 14,874/2024) — on research development and access to orphan drugs. The insufficiency of integrated data and analyses regarding the connections, gaps, and consequences of these regulations in the practical healthcare context represents a significant obstacle.

Thus, it is essential to fill this gap in order to identify solutions that optimize scientific research and ensure fair access to treatments, contributing to improved quality of life for patients with rare diseases in Brazil.

Accordingly, the main objective of this study is to explore and describe the process of development and implementation of official ethical and health regulations related to rare and ultra-rare diseases in Brazil, from the National Policy for Pharmaceutical Assistance (PNAF, 2007) to the General Law on Research with Human Beings (Law No. 14,874/2024). The study aims to identify and discuss the implications, barriers, and challenges of these regulations for advancing research on rare diseases and access to orphan drugs in the country, contributing to debate and the formulation of more effective public policies.

Methods

This is an exploratory study that adopted a qualitative approach, focusing on document analysis and a narrative literature review. The study did not employ a quantitative method, as its primary objective was to describe the nuances present in regulations and in the experiences related to the challenges of research and access to orphan drugs in Brazil, reflecting a more exploratory and interpretative nature. The choice of a qualitative method stemmed from the interest in describing nuances and interpreting the interconnections and gaps in public policies, rather than quantifying data.

The document analysis involved an in-depth examination of official documents in force between 2007 and 2024, covering laws, policies, and resolutions relevant to rare and ultra-rare diseases. This period was strategically selected to trace the evolution of public policies from the creation of the National Policy for Pharmaceutical Assistance (PNAF) in 2007 to Law No. 14,874/2024. Documents were selected from the official portals of the Ministry of Health and the National Health Surveillance Agency (ANVISA), totaling 10 regulations, including the National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR) (2014) and resolutions from the National Health Council (Figure 3).

The following documents were included

- National Policy for Pharmaceutical Assistance (PNAF) (2007);
- National Policy for Clinical Genetics Care in SUS (PNAIGC) (2008);
- Law No. 12,401/2011 (CONITEC);
- CNS Resolution 466/2012;
- CNS Resolution 563/2016;
- National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR) (2014);
- Annex XXXVIII of Consolidation Ordinance No. 2 of 2017;
- Consolidation Ordinances No. 6 and No. 2, both dated September 28, 2017;
- General Law on Research with Human Beings (Law No. 14,874/2024).

The analyzed documents addressed central themes such as rare and ultra-rare diseases, access to medicines for these conditions, strategies for research and technological development, public health, and government-funded care for this specific population. The selection also considered keywords such as “access to medicines,” “research incentives,” “vulnerability,” “health promotion,” and “quality of life,” to highlight relevant passages and identify patterns.

The qualitative analysis followed the principles of Content Analysis according to Bardin, involving the stages of pre-analysis, material exploration, and result processing. During exploration, meaning units related to problems, planned actions, declared intentions, and policy gaps were noted, enabling the creation of emergent thematic categories aligned with the axes of the aforementioned keywords. Although the methodological text did not predefine these categories, they were constructed from the material itself.

In addition to document analysis, a narrative review of recent scientific literature (2020–2024) was conducted, searching databases such as MEDLINE/PubMed with descriptors related to legislation, public policies, rare diseases, and access to orphan drugs. This review aimed to situate the documentary findings within the contemporary context of scientific knowledge, allowing comparison and interpretation of the data in light of current theories and debates.

The combination of these two sources—official documents and scientific literature—enabled a richer interpretative analysis, which went beyond merely describing the regulations and sought to understand their practical implications, identify barriers and challenges faced, and uncover gaps in Brazilian public policies. Thus, the study highlighted the complexity of the subject, revealing the need for more integrated and effective policies to ensure access to and the development of research on orphan drugs in Brazil.

Ethical Considerations

As this research involves publicly available documents, approval by a Research Ethics Committee (CEP) may be waived, in accordance with National Health Council (CNS) Resolutions 466/2012 and 510/2016, which regulate research involving human beings. Nevertheless, it is essential to ensure the integrity of the documents and the accuracy of the cited information, avoiding any alterations or decontextualized interpretations. All sources were duly referenced.

Figure 1. Rare Diseases in Numbers

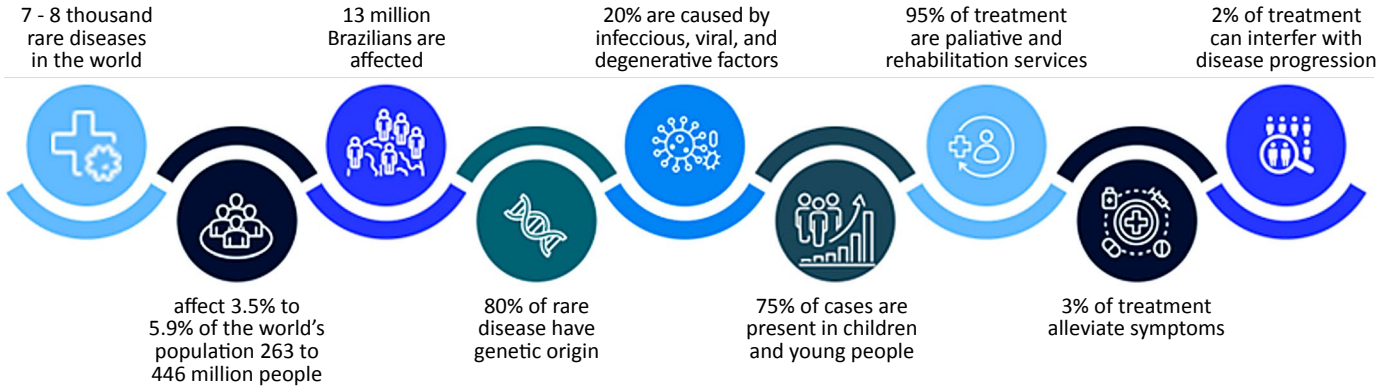


Figure 2. The PNAIPDR and Its Updates

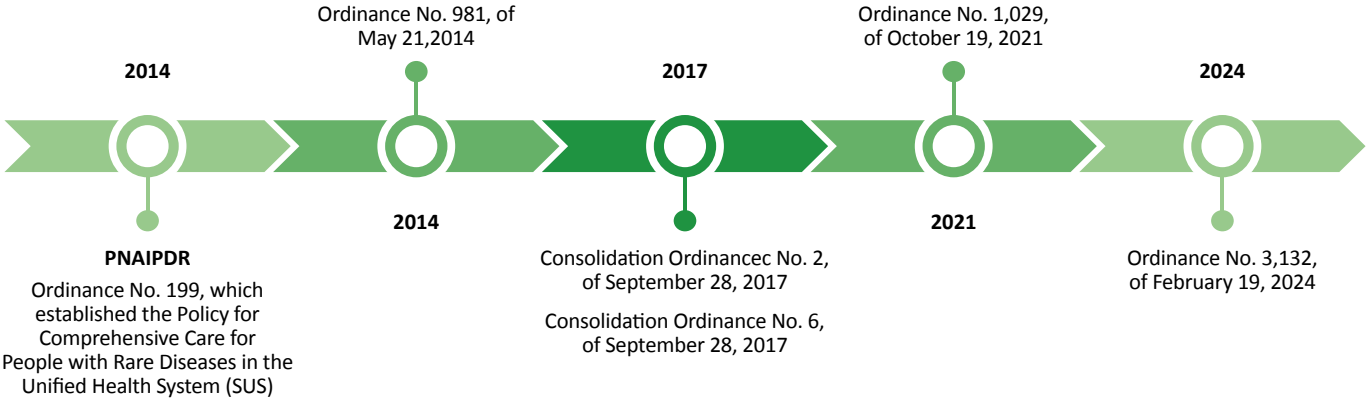
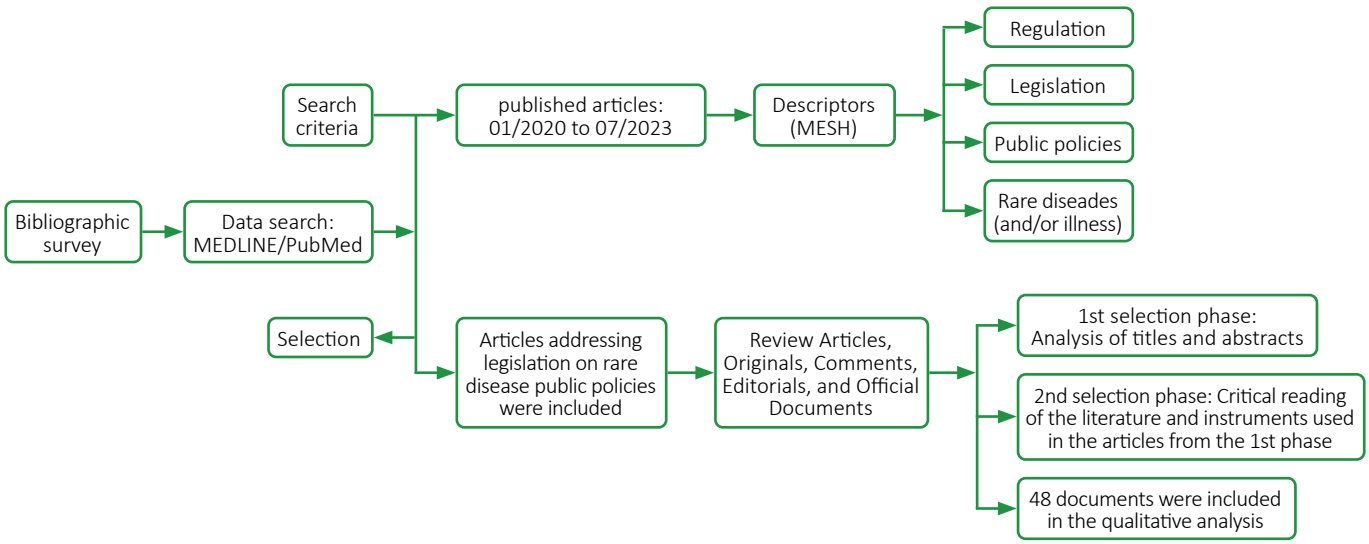


Figure 3. Flowchart of the Literature Survey



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Results

This study aimed to describe the process of development and implementation of ethical and health regulations for rare and ultra-rare diseases in Brazil, from the framework of the National Policy for Pharmaceutical Assistance (PNAF – 2007) to the recent General Law on Research with Human Beings (Law No. 14,874/2024), identifying challenges for research development and access to orphan drugs in the country.

Document analysis and the narrative literature review revealed a regulatory trajectory culminating in the National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR), established in 2014. However, this policy was preceded by important initiatives, such as the PNAF of 2007 (CNS Resolution No. 338/2004), considered a landmark for expanding the budget for medicines targeting genetic diseases, many of which are classified as rare (approximately 80% have a genetic origin).

Another advance was the National Policy for Clinical Genetics Care in SUS (PNAIGC) in 2009, which established a foundation for genetic diagnosis and treatment, essential for the care of rare and ultra-rare diseases. These instruments created the conditions for the development of specific policies and technologies targeting these conditions.

The enactment of Law No. 12,401/2011, which amended Law No. 8,080/1990, was another significant milestone, establishing the National Commission for the Incorporation of Technologies in SUS (CONITEC), responsible for evaluating and incorporating health technologies. CONITEC is also responsible for the preparation and updating of Clinical Protocols and Therapeutic Guidelines (PCDTs), being essential for standardizing care. As of 2025, there are 27 PCDTs focused on rare diseases: 2 under development, 9 being updated, 3 under review, and 13 already approved (Table 1).

The scenario prior to PNAIPDR was characterized by fragmented healthcare, lack of clear guidelines, and restrictions on access to diagnosis and treatment, which compromised the generation of scientific knowledge. The PNAIPDR has contributed significantly by establishing care guidelines, organizing the healthcare network, instituting specialized services, and defining flows for technology incorporation.

However, the policy still faces challenges regarding updates in the face of rapid scientific and technological advancements. Although revisions are underway—such as the expansion of the list of eligible diseases and the updating of PCDTs—the pace of change does not keep up with innovations and patients' emerging needs, creating gaps, particularly in the field of research.

The absence of structured mechanisms to promote research is one of the main weaknesses of the PNAIPDR. The policy does not provide specific funding for studies on rare and ultra-rare diseases, forcing researchers to rely on general calls for proposals that do not always consider the particularities of this field. This limitation compromises knowledge generation and the development of innovative therapies, hindering the creation of an environment conducive to clinical research and innovation.

Another critical issue is the lack of a robust national registry system for patients with rare diseases. Without qualified and interoperable data, it becomes difficult to conduct population-based epidemiological studies, recruit patients for clinical trials, or monitor interventions on a large scale. The absence of a national registry undermines both surveillance and the evaluation of the effectiveness of health policies and technologies.

Table 1. Rare Diseases with Approved PCDT(14)

Rare Diseases
Acromegaly
Autoimmune Hemolytic Anemia
Angioedema associated with C1 esterase inhibitor deficiency (C1-INH)
Spinal Muscular Atrophy 5q Type I and II
Primary Biliary Cholangitis
Biotinidase Deficiency
Growth Hormone Deficiency – Hypopituitarism
Dermatopolymyositis and Polymyositis
Diabetes Insipidus
Dystonias and Hemifacial Spasm
Addison's Disease
Crohn's Disease
Gaucher's Disease
Paget's Disease
Pompe Disease
Wilson's Disease
Sickle Cell Disease (Sickle Cell Anemia)
Amyotrophic Lateral Sclerosis
Multiple Sclerosis
Systemic Sclerosis (Scleroderma)
Ankylosing Spondylitis
Phenylketonuria (PKU)
Cystic Fibrosis
Paroxysmal Nocturnal Hemoglobinuria
Autoimmune Hepatitis
Hidradenitis Suppurativa
Congenital Adrenal Hyperplasia
Pulmonary Arterial Hypertension
Hypoparathyroidism
Congenital Hypothyroidism
Classical Homocystinuria
Hereditary Ichthyoses
Primary Immunodeficiency with Antibody Defects
Exocrine Pancreatic Insufficiency
Lymphangioleiomyomatosis
Neuronal Ceroid Lipofuscinosis type 2
Systemic Lupus Erythematosus (SLE)
Miastenia Gravis
Mucopolysaccharidosis type I
Mucopolysaccharidosis type II
Mucopolysaccharidosis type VII
Mucopolysaccharidosis type IV A
Mucopolysaccharidosis type VI
Osteogenesis Imperfecta
Familial Amyloid Polyneuropathy

Source: Ministry of Health- Approved PCDTS (14)

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The lack of a specific national research plan also limits the definition of priorities and interinstitutional coordination. Without clear guidelines for funding and conducting research on rare diseases, efforts remain scattered, hindering consistent advances in the field.

Slow incorporation of new diagnostic and therapeutic technologies is another identified weakness. Although the PNAIPDR establishes guidelines for organization and care, the delay in adopting orphan drugs and advanced therapies within SUS restricts patient access and limits the conduction of real-world evidence (RWE) studies. Such studies are essential to demonstrate safety and efficacy in national contexts and to guide clinical and regulatory decision-making.

Therefore, despite the normative and institutional advances observed since 2007, structural and operational barriers persist, limiting scientific development, evidence generation, and equitable access to innovative therapies for rare and ultra-rare diseases in Brazil. Strengthening the PNAIPDR, with a focus on research, data, and innovation, is essential to overcome these challenges.

Discussion

Public policies for rare diseases, both in the United States and Europe, originated from pressure exerted by patient organizations and families affected by specific conditions. In the United States, during the 1970s, the National Organization for Rare Disorders (NORD), a private organization, lobbied the government to create specific laws for the development of orphan drugs, which culminated in the Orphan Drug Act. This Act established incentives for orphan drug research, including subsidies for clinical studies, market exclusivity, and increased patent protection for the compounds.[8]

Although the National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR) includes shared guidelines to address deficiencies in specialized healthcare services through professional incentives and adjustments, both diagnosis and therapy require the implementation of the directives guiding the Brazilian Unified Health System (SUS). Nevertheless, critical gaps remain in meeting the multifaceted needs of patients.[9]

The SUS does not have a specific pharmaceutical care policy for rare diseases, and the construction of such a policy encounters bioethical challenges involving issues such as equity, limited resources, and the principle of "reserva do possível" (the limitation of what is feasible)[10]. Despite this, the demand for judicially mandated treatment (TRE) is increasing, sometimes conflicting with the National Medicines Policy.

The central question here is whether the government should financially stimulate research for the development of drugs for rare diseases. Critics argue that, due to their rarity, proving treatment efficacy is more difficult, and creating special funds for rare diseases could violate the principle of equity in the healthcare system. However, experts agree that research on rare diseases often reveals pathological mechanisms common to other conditions and represents important scientific advances.[11]

Perhaps the answer to this question can once again be drawn from recent history. The United States, through the Orphan Drug Act, created special government funding lines for orphan drug research and allowed differential management of clinical trials, leading to faster approval of these drugs.

The results of this program are evident in the numbers: between 1972 and 1982, the U.S. approved ten treatments for rare diseases, while from 1983 to 2006, this number rose to 282, highlighting the importance of such subsidies.[10]

The PNAIPDR, in particular, lacks a robust framework for funding and incentivizing research and development of innovative therapies. This omission perpetuates dependence on foreign technologies and limits access to essential therapeutic options. Although existing policies emphasize the organization of the healthcare system and care pathways, the absence of strategic investment in translational research and the development of national biotechnologies compromises the autonomy and sustainability of the healthcare system in the face of these complex pathologies.

A recent survey and characterization of research protocols on rare diseases involving human subjects, submitted to the CEP/Conep system through Plataforma Brasil between 2013 and 2023, revealed a landscape with striking results that directly reflect the weaknesses of the PNAIPDR in promoting translational research on rare and ultra-rare diseases in Brazil. The studies showed a predominance of single-center designs, suggesting limited potential for research collaboration, and nearly all protocols originated from national institutions. The temporal distribution of studies indicated a progressive increase from 2013 (58 studies) to 2019 (160 studies), followed by a decline in 2020 and 2021, and a subsequent recovery to 176 studies in 2023.

Analysis of funding sources showed a predominance of publicly funded studies (67.87%), followed by privately funded studies (30.82%), and a small proportion of mixed funding (public/private, 1.31%). The result that most reflected the challenges and barriers in research and access to orphan drugs concerned the study design and type: there was a significant predominance of observational studies (87.78%), in contrast with a minority (12.22%) of interventional/experimental studies with a primary clinical purpose (according to the WHO). It is important to note that the "interventional/experimental" category in Plataforma Brasil encompasses a wide variety of study designs, characterized by different types of intervention (e.g., experimental, comparator, active, device, placebo, dose-escalation, including even no-intervention groups) and diverse intervention modalities (such as drugs/medicines/vaccines, medical devices, biological therapies, surgical procedures, radiation, behavioral approaches, gene therapies, and dietary supplementation). The results also revealed the distribution of interventional/experimental studies across development phases: Phase 1 (19.50%), Phase 2 (6.92%), Phase 3 (22.01%), Phase 4 (5.03%), Phase 1/2 (2.52%), Phase 2/3 (3.77%), a representative "Other" category (30.19%), and a portion of studies not classified by phase (10.06%).[12]

Brazil faces a critical paradox: the constitutionally enshrined universality of health collides with the pragmatic limitations of the Unified Health System (SUS). While the PNAIPDR formally guarantees access to orphan drugs, diagnosis, and treatment, in practice it functions as a policy of restricted access. The high cost of pharmaceuticals and limited resources perpetuate inaccessibility for most patients. This discrepancy drives the pursuit of alternative pathways, such as judicialization of medicines and post-trial access in clinical studies.

The 130% increase in lawsuits for high-cost medicines is symptomatic of the failure to ensure equitable access through ordinary channels. Although the Interfederative Agreement seeks

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to establish criteria, judicialization diverts resources from broader public policies, creating challenges for SUS's financial sustainability and equity in care. Furthermore, judicialization, besides failing to guarantee access, exacerbates inequality in resource distribution, and post-trial access, limited under Law 14,874, proves insufficient for genetically inherited and progressive rare diseases, which require continuous, lifelong treatment.

Overcoming the complex challenge of rare diseases in Brazil requires a profound reformulation of public policies that goes beyond mere economic adjustments. Building an equitable and sustainable care network presupposes incorporating a robust bioethical perspective that prioritizes patient dignity and autonomy. Mere expansion of infrastructure and increased investment in research, while necessary, remain insufficient in the face of the historical neglect of the specificities of these pathologies within the financing model.

The PNAIPDR, in its current configuration, fails to realize the principle of equity, exposing the fragility of the State's commitment to comprehensive healthcare for patients with rare diseases.

Limitations

The qualitative approach, combining document analysis and narrative review, while appropriate for an exploratory study, presents limitations. The absence of primary data, such as interviews with key stakeholders, restricts understanding of practical experiences and implementation challenges. The interpretative analysis may be subjective in identifying gaps and connections. Furthermore, the lack of a rigorous systematic review and the absence of regional analyses limit theoretical depth and the understanding of local variations in policy implementation.

Conclusion

This investigation allowed for mapping the origin and evolution of the Brazilian regulatory framework on rare and ultra-rare diseases, highlighting three central points: weaknesses in public policies, diversity in access pathways to orphan drugs, and lack of incentives for technological development. This triad underscores the urgent need for review and strengthening from legal, ethical, social, and economic perspectives.

A notable deficiency is the absence of a robust legal instrument to ensure the integrity of the National Policy for Comprehensive Care of People with Rare Diseases (PNAIPDR), which remains an administrative ordinance without the force of federal law. Although SUS guarantees universality and comprehensiveness in principle, the documentary and literature analysis reveals a gap in the practical realization of these rights.

Brazil needs to advance in drug production, establish partnerships with associations and other stakeholders, and improve registration and incorporation processes within SUS. Optimizing regulatory timelines and encouraging international multicenter studies are also essential.

It was observed that not all drugs researched in Brazil are registered, hindering the incorporation of new technologies and access to innovative therapies. Reliance on judicialization to obtain these medicines, although common, is slow and complex, causing suffering for patients and their families.

To overcome these challenges, the creation of a specific law for rare diseases is recommended, with clear mechanisms for funding and monitoring; expansion of the care network and training of healthcare professionals; promotion of clinical research and technological innovation; acceleration of regulatory processes; expansion of therapeutic coverage; and greater participation of civil society and patient associations in the formulation and implementation of public policies.

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Collaborators

FAO, RES, and MRCGN contributed to the study design, data analysis and interpretation, as well as drafting the manuscript and critically revising the text. All authors approve the final version to be published and take responsibility for the content of the article, ensuring the accuracy and integrity of all its parts.

Declaration of conflict of interests

The authors declare that there are no conflicts of interest regarding this article.

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