“Many of us are rare”: the right to health and the moral economy of rare diseases activism in Brazil (1990-2020)

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ABSTRACT: This article discusses the different meanings attributed to the right to health in the mobilizations to guarantee care for rare disease patients in Brazil. Since the early 1990s, rare disease patient family associations have been putting agendas to the public authorities, with demands ranging from the recognition of diseases to the development of research and diagnostic tests. The trajectory of the mobilizations to guarantee care for rare disease patients is part of a broader process of social articulation around the possibilities and limits of the Brazilian National Health System. The right to health is at the center of this process, being the Subject of varied debates and interpretations in the political, health, and legal arenas, involving arguments guided by scientific evidence, legal principles, and moral values. We argue that these variations in the directions of the right to health have involved concomitant processes of strengthening networks of rare disease actors and institutions, and of maturing the health care system, both converging towards the establishment of a “moral economy of rare disease patients”. This moral economy is centered on the idea that the public health relevance of these diseases cannot be identified by epidemiological evidence, but rather by individual and family experience. We used a wide scope of documentation: texts from newspapers and magazines widely circulated in the country, legislative material, publications in specialized journals, and website materials from rare disease patient organizations in Brazil.

PALABRAS CLAVE: enfermedades raras, economía moral, derecho a la salud, activismo, Brasil.

KEYWORDS: rare diseases, moral economy, right to health, activism, Brazil.

1. Introduction(*)

On January 30, 2014, an internal ordinance of the Brazilian Ministry of Health established the National Policy for Comprehensive Care for People with Rare Diseases[1], which provided for the organization of everything from the offer of diagnostic tests and genetic counseling to the complex access to orphan drugs, high-cost drugs with low attractiveness to the pharmaceutical industry. The elaboration process of this decree involved a series of negotiations between actors of the political field, patient and family associations, medical societies, and entities representing the pharmaceutical industry, in articulations that made possible the transformation of an agenda of specific groups into a public policy[2].

Since the early 1990s, rare disease patient family associations have been putting agendas to the public authorities, with demands ranging from the recognition of genetic diseases such as amyotrophic lateral sclerosis and Gaucher disease to the development of research and diagnostic tests. With the maturation of the Unified Health System, created in 1990 and guided by the principles of universality, integrality, and equity in access to health care, groups mobilized around rare disease care, and their agendas changed, especially with the demand for access to medicines via the State, in respect to the right to health guaranteed by the Brazilian Constitution of 1988.

The Brazilian political-institutional scenario in recent years has made the effectiveness of the national policy of 2014 quite problematic, in the context of the dismantling of the health system and strengthening of the private sector based on neoliberal agendas. Thus, mobilizations for the care of rare disease patients remained relevant and necessary for interest groups, with the strengthening of networks created in previous decades and

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1. Brasil. *Portaria n.º 199, de 30 de janeiro de 2014*. Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras, aprova as Diretrizes para Atenção Integral às Pessoas com Doenças Raras no âmbito do Sistema Único de Saúde (SUS) e institui incentivos financeiros de custeio. Disponível em: https://bvsms.saude.gov.br/bvs/saudelegis/gm/2014/prt0199_30_01_2014.html
2. Luiz Alves Araújo Neto and Luiz Antonio Teixeira, "Nuevos problemas de un nuevo sistema de salud: la creación de una política pública nacional de atención de enfermedades raras en Brasil (1990-2014)," *Salud Colectiva* 16 (2020): e2210. doi: 10.18294/sc.2020.2210.

the articulation of the idea of "public responsibility"[3] in ensuring access to health care for ill people.

The trajectory of the mobilizations to guarantee care for rare disease patients is part of a broader process of social articulation around the possibilities and limits of the Brazilian National Health System. The right to health is at the center of this process, being the Subject of varied debates and interpretations in the political, health, and legal arenas, involving arguments guided by scientific evidence, legal principles, and moral values. The very definition of "rare diseases" carries a set of expectations and values quite particular and complex to the health field, involving issues such as ethics of care, state responsibility, and the relationship between the cost and effectiveness of health measures.

This article discusses the different meanings attributed to the right to health in the mobilizations to guarantee care for rare disease patients in Brazil. Starting from the 1990s, the moment of creation of the first associations dedicated to rare conditions in Brazil, and reaching up to 2020, six years after the publication of the national policy, we demarcate some main meanings attributed to the right to health in the debates about rare diseases: recognition of the relevance of genetic diseases; demand for research and development of diagnostic technologies; access to orphan drugs; guarantee of comprehensive care. We argue that these variations in the directions of the right to health have involved concomitant processes of strengthening networks of rare disease actors and institutions, and of maturing the health care system, both converging to the establishment of a "moral economy of rare disease patients". This moral economy is centered on the idea that the public health relevance of these diseases cannot be identified by epidemiological evidence, but rather by individual and family experience. In this way, public responsibility to patients with rare genetic conditions challenges the traditional framework of "statistical reasoning"[4] and "metrics"[5].

Our approach brings together references from the History of Medicine and the History of Public Health, especially on the participation of

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3. Martha Moreira *et al.*, "Quando ser raro se torna um valor: o ativismo político por direitos das pessoas com doenças raras no Sistema Único de Saúde," *Cadernos de Saúde Pública* 34, no. 1 (2018): e00058017 https://doi.org/10.1590/0102-311X00058017
4. Alain Desrosières, *La politique des grands nombres: une histoire de la raison statistique* (Paris: Decouvert, 1995).
5. Vincanne Adams, ed, *Metrics: What counts in Global Health* (London/Durham: Duke University Press, 1995).

interest groups in the organization of health systems, with readings from Anthropology and Sociology on the power of patients to set agendas for the health field and biomedicine, as well as in studies on the importance of this perspective in evidence-based decision making and policy planning[6]. To this end, we used a wide scope of documentation: texts from newspapers and magazines widely circulated in the country, legislative material, publications in specialized journals, and website materials from rare disease patient organizations in Brazil.

2. Right to health in Brazil: exclusion, universalization, and limits

A discussion on the history of the right to health in Brazil should take into account the different expectations regarding the role of the state in the provision of health services over time, especially during the 20th century. Conceptually, the use of the notion of right refers to the context of mobilization for the re-democratization of the country in the 1970s and 1980s, in a strict association between democracy and the health conditions of a population[7]. In previous periods, the offer of services by the State resulted from other interpretations about its functions and obligations, in a structure divided (from the 1930s on) between public health —responsible for campaigns to combat specific diseases, sanitary education, and other sanitation actions— and social security —in charge of offering medical services to contributing workers[8]. In addition, philanthropic and charitable institutions provided care to the poor population and those suffering from chronic diseases, such as tuberculosis and cancer[9].

One of the central points discussed by the Brazilian health field in the 1970s and 1980s, in the context of the Sanitary Reform Movement, was

6. Steven Epstein, "The construction of lay expertise: AIDS Activism and the Forging of Credibility in the Reform of Clinical Trials," *Science, Technology, and Human Values* 20, no. 4 (1995): 408-437, doi: https://doi.org/10.1177/016224399502000402; João Biehl and Adriana Petryna, "Peopling global health," *Saúde e Sociedade* 23, no. 2 (2014): 376-289, https://doi.org/10.1590/S0104-12902014000200003
7. José Luís Fiori, Hésio Cordeiro and Reinaldo Guimarães, "A questão democrática na área da saúde," *Saúde em Debate* 9 (1980): 11-3.
8. Cristina Fonseca, *Saúde na Era Vargas: dualidade institucional de um bem público* (Rio de Janeiro: Editora Fiocruz, 2007).
9. Luiz Teixeira and Cristina Fonseca, *De doença desconhecida a problema de saúde pública: o INCA e o controle do câncer no Brasil* (Rio de Janeiro: Ministério da Saúde, 2007).

the exclusionary character of the health structure in the country and the limitations of the system centered on access to medical care through social security, in addition to remarkable cases of corruption in the system of contracting services by the State[10]. Together with other highly relevant political and ideological processes, such as criticism of the biomedical model, the institutionalization of the field of Collective Health, and the debates on expanding health care, among others, this reform movement enabled the creation of the Unified Health System (SUS) and the definition of health as a social right in the 1988 Constitution[11].

Although the constitutional guarantee of the right to health and the creation of SUS represented great social achievements in the history of Brazil, its implementation process and the interpretations of this process have raised different questions in the health field over the last thirty years. A clear diagnosis points to limitations in the effectiveness of the system's principles —universalization, integrality, and equity— which have made it difficult, in different ways, to guarantee the right to health[12]. A structural aspect is the intensification of neoliberalism as a platform of capitalism in the late twentieth century and early twenty-first century, placing various obstacles to public investment in social protection and welfare policies, a scenario that had specific contours in Latin America due to the privatizing character of the dictatorships that existed between the 1960s and 1990s[13].

Besides the limitations of the system, there are tensions regarding the articulation of the right to health in terms of public policies and service supply. This is the case of the binomial universalization and focalization of health policies, referenced in discussions about access to high-cost medicines and the budget constraints of the health system[14]. In short, focalization consists of

10. Carlos Gentile de Melo, "Medicina Previdenciária," in *Saúde e medicina no Brasil: contribuição para um debate,* ed. Reinaldo Guimarães (Rio de Janeiro: Editora Graal, 1978), 175-181.
11. Luiz Antonio Teixeira and Carlos Henrique Paiva, "Saúde e reforma sanitária entre o autoritarismo e a democracia," in *História da Saúde no Brasil,* ed. Luiz Antonio Teixeira, Tânia Pimenta, Gilberto Hochman (São Paulo: Editora Huictec, 2018), 430-463.
12. Luis Fernandes de Souza *et al.,* "Os desafios atuais da luta pelo direito universal à saúde no Brasil," *Ciência & Saúde Coletiva* 24, no. 8 (2019): 2783-92, https://doi.org/10.1590/1413-81232018248.34462018.
13. Sonia Fleury, "Iniquidades nas políticas de saúde: o caso da América Latina," *Revista de Saúde Pública* 29 no. 3 (1995): 243-250.
14. Deivison Faustino, "The universalization of rights and the promotion of equity: the case of the health of the black population," *Ciência & Saúde Coletiva* 22, no. 12 (2017): 3831-40, doi: 10.1590/1413-812320172212.25292017; Regina Marsiglia, Cássio Silveira and Nivaldo Junior,

the idea that public resources for social protection should be destined for the most vulnerable segments of the population, particularly the poorest, while universalization sustains the State's responsibility in guaranteeing care to all individuals, regardless of social categorization[15]. Other discussions involve the development of specific policies for groups most affected by different types of social inequity, such as the black population, as well as the actions of specific interest groups, such as the participation of the LGBT movement in the formulation of responses to the HIV/AIDS epidemic[16].

In this sense, rare disease activism's mobilizations around the right to health are part of a larger scenario of efforts to enforce the constitutional text of 1988, with distinct and sometimes conflicting interpretations of what health as a right means. Breast cancer advocacy, for example, interpreted the right to health mainly to claim access to technology, often conflicting with the Ministry of Health and National Cancer Institute's guidelines for early detection and screening[17]. These variations also involve distinct expectations of specific actors about health, medicine, and the role of the state. Observing the actions of patients' organizations and their allies in the political, pharmaceutical, and scientific fields, we deal with perspectives quite distinct from those of public health technicians and managers[18]. The demarcation of the specific position of rare disease interest groups is fundamental to understanding their moral economy and the meanings attributed to the right to health.

"Social policies: inequity, universality and focalization in health within Brazil," *Saúde e Sociedade* 14, no. 2 (2005): 69-76, https://doi.org/10.1590/S0104-12902005000200008.

15. Amélia Cohn, "O SUS e o Direito à Saúde: universalização e focalização nas políticas de saúde," in *Saúde e Democracia: história e perspectivas do SUS,* ed. Nísia Trindade Lima *et al.* (Rio de Janeiro: Ed. Fiocruz, 2005), 385-405.
16. Eliza Vianna and Dilene Nascimento, "'Produz-se um contradiscurso': O Grupo Pela Vidda RJ e SP na luta contra a AIDS," in *Quando a História encontra a Saúde*, ed. Ricardo Batista, Christiane Souza and Maria Elisa da Silva (São Paulo: Ed. Hucitec, 2020), 143-65.
17. Vanessa Lana and Luiz Teixeira, "Breast cancer and civil society: actions of FEMAMA in the temporal regulation for diagnosis and treatment of the disease in Brazil," *Caminhos da História* 26, no. 2 (2021): 112-35, doi: 10.38049//issn.2317-0875v26n2p.112-135.
18. Luiz Teixeira and Luiz Alves Araújo Neto, "Still controversial: early detection and screening for breast cancer in Brazil, 1950s-2010s," *Medical History* 64, no. 1 (2020): 52-70, doi:10.1017/mdh.2019.76 .

3. The right to recognition: rare diseases and activism

The first form of mobilization for the care of rare disease patients was the activist groups created by patients' families in the early 1990s[19]. These associations, created in large urban centers of the country, mainly in Rio de Janeiro and São Paulo, focused on specific diseases, such as amyotrophic lateral sclerosis, Gaucher's disease, and Down syndrome (then qualified as a disease). Although there were common aspects in the characterization of these diseases, especially regarding the impairment of motor functions and their "rareness" - still without quantitative precision - the first associations did not think of a collective definition for rare conditions. Despite this, the similarities of the agendas and of the very diseases for which there were mobilizations make an overall analysis possible.

These first groups emphasized legal counseling and support for people with degenerative and genetic diseases and their families, mobilizing their inclusion in studies for the development of genetic tests, such as the development of a PCR (polymerase chain reaction) test for the diagnosis of familial hypercholesterolemia by the University of São Paulo, in 1994[20], and guaranteeing care in hospital institutions, such as the Hospital Clementino Fraga Filho, linked to the Federal University of Rio de Janeiro, which created a service for the care of people with rare diseases in 1991[21].

In the semantics of the groups forming in the 1990s, the right to health for rare disease patients was primarily a right to the recognition of their condition, both in its individual and collective dimensions[22]. On the individual level, the struggle for recognition dealt with the difficulty of diagnosing rare genetic and degenerative diseases, and with the discrimination and stigmatization of people suffering from them[23]. Stigmatization took different forms, both by reducing rare disease patients to the identity of disabled (and pejorative variations such as "retarded") and by discrimination between groups of

19. Araújo Neto and Teixeira, "Nuevos problemas de un nuevo sistema de salud", 3-5.
20. Ana Lúcia Azevedo, "Teste genético detecta doença rara," *O Globo*, December 25, 1994.
21. Folha de São Paulo, "Hospital do Rio cria serviço de atendimento a doenças raras," *Folha de São Paulo*, November 28, 1991.
22. O Globo, "Vítimas de ELA tentam se organizar," *O Globo*, October 4, 1992; O Globo. "Uma mobilização para salvar vidas," *O Globo*, September 1 1992.
23. O Globo, "Em defesa dos deficientes," *O Globo*, October 18 1992.

people with degenerative diseases, such as prejudice against patients with amyotrophic lateral sclerosis (ALS) by people with multiple sclerosis[24].

In the collective dimension, the struggle for recognition sought, above all, to guarantee care for people who had no space in healthcare networks, mainly because of the lack of structure to deal with genetic and complex conditions. Reacting to the denial of their illness and the silencing of the experiences of individuals with rare diseases when compared with other situations of greater epidemiological impact, the groups questioned quantitative-based evaluation parameters and valued subjectivity and qualitative evidence[25]. This rhetorical formulation was central to the conformation of a moral economy for rare disease interest groups, a set of political and moral values that guides patient advocacy.

We mobilize the concept of moral economy in dialogue with the readings of Fassin[26] and Palomera and Vetta[27], thinking about how feelings, political views, and moral values are articulated by groups to deal with specific problems, such as the moral economy of children with HIV[28] or the moral economy of epidemiologists[29]. These values are dynamic and bring into interaction the expectations of actors and their actions in concrete and complex contexts. In the case of rare diseases, the central point is to highlight the limits of frameworks such as Evidence-based medicine (EBM) and cost-benefit assessments in the organization of healthcare. Rare diseases challenge EBM methods and theoretical frameworks by making it difficult to organize clinical trials and produce data in terms of the general patterns of knowledge production in EBM. Even though patient organizations gave shape to a "politics of numbers" through the concept of rareness, framing

24. O Globo, "Alerta contra doença rara," *O Globo,* August 24, 1992.
25. Ítala Paris de Souza *et al.,* "Doenças genéticas raras com abordagem qualitativa: revisão integrativa da literatura nacional e internacional," *Ciência & Saúde Coletiva* 24, no. 10 (2019): 3683-3700, DOI: 10.1590/1413-812320182410.17822019.
26. Didier Fassin, "As economias morais revisitadas," *Revista Brasileira de Sociologia da Emoção* 18, no. 53, (2019): 27-54.
27. Jaime Palomera and Theodora Vetta, "Moral economy: rethinking a radical concept," *Anthropological Theory* 16, no. 4 (2016), https://doi.org/10.1177/1463499616678097
28. Didier Fassin, "Children as Victims: The moral economy of childhood in the times of AIDS," in: *When People Come First: Critical Studies in Global Health,* ed. João Biehl; Adriana Petryna (Princeton: University of Princeton Press, 2013), 109-30.
29. Luiz Alves Araújo Neto. "O 'papel transformador' de uma profissão: a economia moral dos epidemiologistas no Brasil (1970-2000)," *Revista NUPEM* 13, no. 29 (2021): 133-54. DOI: https://doi.org/10.33871/nupem.2021.13.29.133-154.

rare conditions in epidemiological reason, there are several limitations to the extent one can work with Evidence-based medicine to study these illnesses[30].

This critique mobilizes two distinct mechanisms of understanding rare illnesses: generalization and singularization. First, the construction of a "full picture view" represented in the 2010s by the slogan "Many of us are rare" adopted by the Parliamentary Mixed Front for the Comprehensive Care of People with Rare Diseases, arguing that the "rare" status does not mean little epidemiological impact and burden in the lives of patients and their families[31]. In the process of generalizing rare diseases, it points to the relevance of less noticeable problems through metrics based on epidemiological surveys, randomized clinical trials, and meta-analyses.

The singularization, in turn, highlights the particularities of the illness due to a specific rare condition, such as muscle degeneration resulting from amyotrophic lateral sclerosis, or even the intense fatigue resulting from chronic fatigue syndrome. The definition of a distinctive character for an illness is central to the demarcation of a disease as a specific entity, with an important impact on the interaction between doctor and patient, on the legitimization of the search for care in health services, on the bureaucratic organization of patient care, and the self-knowledge of the sick person[32]. In addition, the singularization of rare diseases has grounded the demands of associations for the development of specific drugs by the pharmaceutical industry, despite the low commercial interest in producing drugs whose target audience is limited and the cost is very high.

The demarcation of general and unique aspects of rare diseases by associations demanded alliances with medical societies and other interest groups, such as political segments. In terms of reaching out to experts, the Brazilian Society of Clinical Genetics represented an important ally to rare disease activism, as seen in the XIII Brazilian Congress of Clinical Genetics, held in Águas de São Pedro (São Paulo) in 2001. Unlike HIV/AIDS activism,

30. Vololona Rabeharisoa *et al.* "From 'politics of numbers' to 'politics of singularisation': Patients' activism and engagement in research on rare diseases in France and Portugal," *BioSocieties*, 9, no. 2 (2014): 1-24, doi: 10.1057/biosoc.2014.4.
31. See: "Estatuto", accessed October 25, 2022, https://muitossomosraros.com.br/frente-parlamentar/estatuto/,
32. Charles Rosenberg, "The Tyranny of Diagnosis: Specific Entities and Individual Experience," *The Milbank Quarterly* 80, no. 2 (2002): 237-260, doi: 10.1111/1468-0009.t01-1-00003.

which was very active in building up a "lay expertise"[33] to mobilize its agendas in Brazil, disseminating studies in its bulletins and advising HIV-positive people on their relationship with doctors[34], rare disease activism followed what Rabeharisoa and colleagues call a "delegation model"[35], where patients take care of the emotional and social aspects and delegate the medical aspects to the experts.

In this way, the right to recognition for rare diseases is one of the meanings attributed to the right to health established by the 1988 constitution, as part of a broader movement to promote the agendas of specific groups through the activism of patients and their families. In the following decades, other aspects of the right to health have been strongly mobilized by activist groups, but the agenda of recognition remains central to the agenda of rare disease activism, emphasizing the individual and family experience of illness to activate the notion of public responsibility for the care of patients with rare conditions.

4. Orphan drugs, access to treatment, and judicialization of health care

A turning point in the history of rare diseases in Brazil was the mobilization seeking access to orphan drugs through state resources. Since the 2000s, the search for high-cost drugs for the treatment of genetic and rare diseases has become the main demand of patient organizations and has occupied the public scene, on the pages of newspapers, in debates involving actors from the political and health fields, and in events organized by medical societies and entities representing the pharmaceutical industry[36]. In discussions on the theme, the right to health appears as a central element both for defenders of the purchase of orphan drugs by the state and for its critics. The sense

33. Steven Epstein, "The construction of lay expertise: AIDS activism and the forging of credibility in the reform of clinical trials," *Science, Technology and Human Values* 20, no. 4, (1995): 408-37, https://doi.org/10.1177/016224399502000402
34. Eliza da Silva Vianna and Dilene do Nascimento, "Produz-se um contradiscurso: o Grupo Pela Vidda RJ e SP na luta contra a aids," in *Quando a História encontra a Saúde,* ed. Ricardo Batista, Christiane Souza and Maria Elisa Silva (São Paulo: Editora Hucitec, 2020), 143-165.
35. Vololona Rabeharisoa *et al.,* "The dynamics of causes and conditions: the rareness of diseases in French and Portuguese patients' organizations' engagement in Research," *Papiers du Research du CSI*. (Paris: Centre de Sociologie de l'Innovation, 2012), https://shs.hal.science/halshs-00702088
36. Araújo Neto and Teixeira. "Nuevos problemas de un nuevo sistema de salud," 6.

attributed to the right, in these situations, is specific and synonymous with access to technologies and forms of biomedical care, which puts tension on the broader sense of health (a "positive conception"[37]) on which the Brazilian constitution of 1988 is based.

One of the first records of rare disease activism actions on getting access to orphan drugs was the availability by the Ministry of Health, in 2002, of treatment for osteogenesis imperfecta —a genetic disease that severely weakens bones and, at the time, affected 12 thousand to 18 thousand people in Brazil[38]— with disodium pamidronate. The treatment began to be offered by the Department of Medical Genetics of the Fernandes Figueira Institute of the Oswaldo Cruz Foundation in Rio de Janeiro, with mediation by the Brazilian Association of Osteogenesis Imperfecta, which would refer patients for free treatment[39]. This type of articulation exemplifies the "delegation model" commented on above, with activist groups connecting sick people to reference centers for treatment.

However, the landscape for making available high-cost drugs for genetic diseases has become increasingly complex over the decade, especially after the results of mapping the human genome and patenting it, making the market for biomedical development and innovation increasingly restricted and significantly increasing the prices of new drugs[40]. Allied with this, the healthcare system was confronted with a growing number of gaps regarding the care of patients with rare diseases, such as incurable metabolic disorders[41] or chronic pain syndrome[42]. These groups, although receiving support from patient organizations and medical institutions, were not covered by the therapeutic protocols of the Unified Health System (SUS) nor were able to access medications for the management of their diseases.

In this context, mobilizations for the care of rare disease patients have gradually entered the legal arena, as an "alternative way to access health

37. Kenneth Camargo Jr. "As armadilhas da 'concepção positiva de saúde'," *Physis: Revista de Saúde Coletiva* 17, no. 1 (2007): 63-76, https://doi.org/10.1590/S0103-73312007000100005.
38. Robert Jansen, "Ministério da Saúde oferece terapia gratuita para doença rara nos ossos," *O Globo,* April 26, 2002.
39. Jansen, "Ministério da Saúde oferece terapia," 7.
40. Marise Basso Amaral; Sergio Rego. Doenças raras na agenda da inovação em saúde: avanços e desafios na fibrose cística. *Cadernos de Saúde Pública*, 36 (12), (2020) doi: 10.1590/0102-311X00115720
41. Fabiane Leite, "País não ampara doenças como a de Jhéck," *Folha de São Paulo*, September 3, 2005, 11.
42. Drauzio Varella, "A genética da dor," *Folha de São Paulo*," December 9, 2006, 16.

care"[43] and a specific form of negotiation around the right to health - judicialization. The recourse to justice to guarantee access to treatment has been the target of several reflections in the health field, especially due to the intensification of the judiciary's action in different spheres of Brazilian public life, something that has been called the "judicialization of life"[44]. One of the most sensitive and recurrent points of discussion is the controversy over the role of judicialization in the functioning of the state apparatus. Is it a way to improve public policies? Or would it be opening a series of specific cases that make the State's action unfeasible[45]? We are interested, within the scope of historical analysis, in observing how judicialization has composed an important path in the mobilization for the care of patients with rare diseases, strengthening the idea of public responsibility, and establishing a relevant interpretation of the right to health.

One aspect of the problem that cannot be ignored concerns the relationship between public and private in the health sector in Brazil. Although the Unified Health System is responsible for providing services at all levels of care, the strength of the private system —the legacy of decades of a system segmented between public health and social security medicine[46]— has increased in the last thirty years, with a considerable presence of health insurance companies. However, in the 2000s, Brazilian insurance companies did not offer coverage for any services related to genetic conditions, which increased the demand from rare disease interest groups to update the list of drugs provided by the Ministry of Health[47]. In 2007, treatments for some rare conditions had already been authorized by the National Health Surveillance Agency (Anvisa), the regulatory agency created in 1999 and responsible for releasing the use of technologies and medications in the country. This was the case for treatments for Gaucher and Fabry diseases and type 1 mucopolysaccharidosis (MPS1); however, the supply by health secretariats

43. João Biehl and Adriana Petryna, "Tratamentos jurídicos: os mercados terapêuticos e a judicialização do direito à saúde," *História, Ciências, Saúde – Manguinhos* 23, no.1 (2016): 173-92, https://doi.org/10.1590/S0104-59702016000100011.
44. Camilla Felix Barbosa de Oliveira and Leila Maria Torraca de Brito, "Judicialização da vida na contemporaneidade," *Psicologia: Ciência e Profissão* 33 (2013): 78-89.
45. Lenir Santos, "Judicialização da saúde: as teses do STF," *Saúde em Debate* 45, no. 130 (2021): 807-18, https://doi.org/10.1590/0103-1104202113018
46. "O mix público-privado na saúde".
47. Antônio Marinho, "Em defesa de poucos," *O Globo*, June 24, 2007.

was irregular and insurance companies did not reimburse policyholders for the costs[48].

It did not take long for the judicialization path to gain space in the public arena, especially pointing out the budgetary burden represented by the supply of expensive medicines to "public health-dependent Brazilians"[49]. Narratives pointing to the vulnerable condition of low-income people, carriers of genetic diseases, and "court clients"[50] that ended up becoming hostages or incidental lobbyists of the pharmaceutical industry's interests occupied the pages of newspapers. It is important to note how, in these reports, the use of the health system is not considered an exercise of rights, but rather a representation of the precarious conditions of people who needed help from the state. This reading brings us back to a neoliberal vision of law and the role of the State, the focalization perspective commented on in the previous section, in which the right to health would be guaranteed to poorer people[51]. The problem, in the case of orphan drugs, is that this guarantee involved high amounts, compromising the budget balance.

In 2009, the debate over the judicialization of health care occupied the plenary of Brazil's Supreme Court (STF). Between April and May, the court held hearings to establish jurisprudence in health judicialization cases, especially regarding access to orphan drugs. The then health minister José Gomes Temporão wrote an article for the Folha de São Paulo newspaper, a widely circulated national periodical, expressing his concern with the pressure to incorporate procedures without due technical consideration in the sector. For Temporão, it was necessary to establish clearer parameters for the inclusion of technologies and drugs in the scope of state funding, so as not to distort the essence of the health system and undermine investments in strategic areas.

> It is impressive and worrisome how the pressure for incorporating experimental procedures, products not registered in the country, and technologies without a strong consensus among specialists, which involve thousands of judicial developments, has distorted the image of SUS management, incorrectly treated as a hinderer of access to procedures and medications.

48. Marinho, "Em defesa de poucos," 6.
49. Fabio Fabrini, "Saúde paga caro por remédios que Justiça manda fornecer," *O Globo*, November 15, 2009.
50. Ruben Berta, "A saúde nos tribunais," *O Globo*, July 24, 2016.
51. Faustino, "The universalization of rights".

> Transferring to the SUS the responsibility for care provided outside its operational norms can generate consequences such as deregulation of access to care, loss of integrality, and reduction of control and evaluation of the care provided[52].

Later, Marcos Bosi Ferraz, professor of the School of Medicine at the Federal University of São Paulo (UNIFESP) and member of the Brazilian Academy of Medicine, presented a more open and radical position than the health minister. For Ferraz, it was necessary to establish priorities for the sector's actions taking into consideration the following criteria: "more important diseases, more frequent, more serious, with more suffering, a greater chance of prevention"[53]. In this sense, although he claimed that people with diseases could not be helpless, he stated that the government needed to recognize its limitations and "clearly and transparently say no sometimes"[54].

On the same page of the newspaper where Ferraz expressed his position, lawyers Andrea Salazar and Karina Grou defended judicialization as a way to balance inequalities, arguing that the actions of the judiciary would guarantee the fulfillment of the right to access to the weakest link in the scales, the users of the health system. For Salazar and Grou, the intervention of the STF in matters of access to orphan drugs did not consist in a breach of the sovereignty of the executive branch, but rather, the essence of the balance of the three powers as established by the Brazilian constitution of 1988[55].

These positions represented different sides of the debate on the right to health in Brazil and its guarantees and limits for the care of rare disease patients. The idea that the guarantee of the right should be conditioned to certain parameters of functionality and effectiveness of the health system, as pointed out by Temporão, was a counterpoint to the perspective of patients, who mobilized the constitutional text to access medicines. It is important to note that this apparent opposition between public health and patient activism concerns quite distinct sets of expectations, values, and views on medicine and health among state technicians and patient organizations. As Aronowitz notes, the negotiations between the different standpoints of

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52. José Gomes Temporão, "O SUS e o direito à coletividade," *Folha de São Paulo*, April 26, 2009, 3.
53. Marcos Bosi Ferraz, "O STF e os dilemas da saúde," *Folha de São Paulo*, May 9, 2009, 3
54. Ferraz, "O STF e os dilemas da saúde," 3.
55. Andrea Salazar and Karina Grou, "As verdadeiras causas e consequências," *Folha de São Paulo*, May 9 de maio, 2009, 3.

groups participating in health planning and decision-making are suggestive of the complexity of the management process in the sector, which is not limited to scientific evidence-driven practices[56].

The argument that the health system should prioritize more prevalent problems that have a greater burden on the population and services also appears frequently in the claim that it would not be possible to provide high-cost medications. This logic operates with the limitations of public health financing, emphasizing a preference for the provision of more urgent services to the detriment of others with less epidemiological impact. Certainly, this cost-benefit-oriented positioning of public policy is the main target of criticism of rare disease activism, since it calls into question the very legitimacy of care for people with these diseases[57].

In the path of judicialization, the right to health has increasingly become synonymous with access to medicines, highlighting the relevance of the pharmaceutical industry as an ally of rare disease activism and putting greater pressure on the regulation and the incorporation of medicines into the National List of Essential Medicines (RENAME). In 2011, the National Commission for Incorporation of Technologies into the Unified Health System (CONITEC) was created to advise the Ministry of Health on the incorporation, exclusion, and alteration of health technologies, as well as to prepare clinical protocols and therapeutic guidelines. The committee is composed of representatives from professional bodies, such as the Federal Council of Pharmacy and the Federal Council of Medicine, public managers, such as the National Council of State Health Secretariats, and regulatory agencies, such as the National Health Surveillance Agency (Anvisa).

The process of incorporating medicines by Conitec involves the submission of an application for inclusion, the holding of public consultations and research evaluations, the convening of a plenary meeting of the committee, and the publication of protocols and guidelines in the official journals of the Brazilian government. These steps demand time and complex procedures, which makes the commission's performance a constant concern

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56. Robert Aronowitz, "To screen or not to screen: what is the question?," *Journal of General Internal Medicine* 10, no. 5 (1995):295-7, doi: 10.1007/BF02599892.
57. Raquel Boy and Fermin Roland Schramn, "Bioética da proteção e tratamento de doenças genéticas raras no Brasil: o caso das doenças de depósito lisossomal," *Cadernos de Saúde Pública* 25, no. 6 (2009): 1276-84; Márcia Pinto *et al.,* "Cuidado complexo, custo elevado e perda de renda: o que não é raro para as famílias de crianças e adolescentes com condições de saúde raras," *Cadernos de Saúde Pública* 35, no. 9 (2019): 1-13, doi: 10.1590/0102-311X00180218.

for activist groups since the speed of access to medicines is a central issue for sick people. In addition, the successful incorporation of a specific drug to the list of the health system is no guarantee of regular supply by the services. Thus, the judicialization agenda to obtain treatment for rare diseases has maintained an important place on the agenda of rare disease activism in the 2010s, in part due to the absence of specific legislation for orphan drugs, such as those created in the United States, Japan, Australia, and the European Union in the 1980s and 1990s. The tensions in access to medications through the courts pose fundamental ethical debates in the field of health, referring to the notion of public responsibility and the principle of protection as guidelines of the state's obligation to provide health services to the population[58].

On the heels of discussions about the right to treatment for people living with rare conditions, broader propositions for care took shape in the 2010s, following the notion of comprehensive care. Increasingly, demands for a national policy on rare diseases gained ground among activist groups and resonated with actors in the political field. However, judicialization continued to be a vital topic in the activism agenda mostly because of the persistent problems in getting access to medicines through the legal system.

5. From advocacy to public policy: interest groups and the national policy on rare diseases

The idea of comprehensive care has great relevance in the semantics of the Brazilian health field, representing one of the principles that guide the health system —integrality— and one of the main challenges of its operation, either by the structure of services or the imperative of medical specialization and its impact on the organization of care. The definition of comprehensive care as one of the key points of the Unified Health System goes back to the sanitary reform process itself and to the criticism of the model guided by hospital medicine, assigning to the State the role of acting on the social determinants and conditioning factors of health until the guarantee of the provision of

58. Boy and Schramn, "Bioética da proteção".

services of the highest complexity in health, such as organ transplants in hospitals and the supply of medicines with high development costs[59].

In the context of rare diseases, the 2009 National Policy of Comprehensive Care in Clinical Genetics presented the first direction toward integrality. This first national policy emphasized genetic counseling, but proposed actions towards comprehensive care, such as the organization of levels of care, the identification of the determinants and conditioning factors of genetic diseases, the definition of criteria for the operation of genetic services, the encouragement of research, and the qualification of care[60]. The effectiveness of this policy, however, has encountered several limitations, from the difficulty in establishing medical genetics services in the more than 5 thousand Brazilian municipalities to the qualification of professionals to perform genetic counseling, anel dilemmas in the bioethical field[61].

Despite its limitations, the national policy on clinical genetics brought an important point to rare disease activism and its allies in the medical field and the pharmaceutical industry: the demand for comprehensive care for patients with rare conditions, expanding the agenda regarding orphan drugs. From the outset, the development of legislation for rare diseases demanded articulation with actors in the political field and a clearer definition of the diseases that would fall under the definition of rareness. Among the actors, some names gained prominence in the early 2010s as allies of the activist groups, such as Mara Gabrilli, Jean Wyllys, and former soccer player Romário, representatives of different positions in the political arena[62].

In 2012, a group led by Gabrilli created the Parliamentary Front for Combating Rare Diseases, important for the formulation of specific laws and policies, with strength also in the articulation with civil society through the slogan "Many of us are rare". We emphasize here the advent of social networks and their use as platforms for groups of patients and families so that they

59. Jairnilson Paim, "Modelos de atenção à saúde no Brasil," in *Políticas e sistema de saúde no Brasil,* ed. Lígia Giovanella *et al.* (Rio de Janeiro: Editora Fiocruz, 2009), 547-73.
60. Brasil. *Portaria n.º 81, de 20 de janeiro de 2009*. Institui, no âmbito do Sistema Único de Saúde (SUS), a Política Nacional de Atenção Integral em Genética Clínica. Brasília. Disponível em: http://dtr2001.saude.gov.br/sas/PORTARIAS/Port2009/GM/GM-81.htm
61. Maria Concepción Nóvoa and Teresinha Fróes Burnham, "Desafios para a universalização da genética clínica: o caso brasileiro", *Rev Panam Salud Publica* 29, no. 1 (2011):61-8.
62. Araújo Neto and Teixeira, "Nuevos problemas de un nuevo sistema de salud,".

can share experiences and mobilize their agendas[63]. Precisely for groups marked by not being composed of so many people, the virtual resource has made it possible to bring together actors and agendas, being a tool of great value to rare disease activism[64]. The Many of Us Are Rare platform is one of the websites that provides space for patient organizations, linked to the parliamentary front and the Brazilian Society of Medical Genetics. According to its website, the platform interacts with 13 million rare disease patients in Brazil, disseminating news, events, and patient testimonials[65]. Bringing together a greater diversity of rare conditions, virtual activism reinforces the importance of singularizing these diseases, as it strengthens the construction of identity against the backdrop of large numbers and epidemiological metrics, mobilizing the political and moral values established in previous decades by activist groups.

However, mobilization in the political arena, especially in policymaking, has demanded greater clarity in the generalization of rare diseases, with the establishment of criteria that bring together so many diverse conditions. In a bill authored by the then federal congressman Jean Wyllys in 2011, it was defined that "a person with a rare disease is one affected by a debilitating and/or incapacitating pathology whose prevalence per 100,000 inhabitants corresponds to 65 cases"[66].

The definition of rare disease through prevalence data was based on the World Health Organization guidelines and the European Union's experience with the topic. The European concept of rare disease, according to the project, highlighted the pathological transversality of the term, involving conditions of genetic, degenerative, autoimmune, infectious, and oncological etiology. This broader characterization allowed the design of healthcare actions at different levels since some diseases would have their control centered in primary care, others in high complexity, etc. Another important aspect of

63. Danielle Fialho da Silva, "Redes de solidariedade do sentir e do resistir: associativismo dos pacientes com fibromialgia e síndrome pós-pólio nos meios digitais (2004-2021)" (PhD diss. Casa de Oswaldo Cruz/Fiocruz, 2021).
64. Maria Angélica de Faria Domingues de Lima, "As associações de pacientes com doenças raras e as mídias sociais" (PhD diss. Instituto Fernandes Figueira/Fiocruz, 2018).
65. https://muitossomosraros.com.br.
66. Jean Wyllys. *Projeto de Lei No. 2669, de 09 de novembro de 2011* [Internet]. 2011 [citado 10 feb 2019]. Available in: https://tinyurl.com/wvqm6ct.

the framework presented in the project concerns the impact of rare diseases on infant mortality, indicated as the second cause of death in this group[67].

In sum, the two movements of singularization and generalization were strategic in the demands for a comprehensive care policy, allowing for negotiations both in the realm of the experience of illness and public responsibility (the individual right to health) and in the political dimension and state administration through the population impact of rare diseases (the collective right to health). Negotiations in the political arena involved discussions about funding, the organization of care, and the participation of the private sector in the provision of services[68]. In 2014, Ordinance 199 of the Brazilian Ministry of Health established the National Policy for Comprehensive Care for People with Rare Diseases, providing for the care of rare disease patients along two axes: the first targeted congenital anomalies, intellectual disability, and inborn errors of metabolism; while the second dealt with infectious, inflammatory, and autoimmune conditions[69]. It also organized the structure of care, defining the responsibilities of each level of care and the network. One of the highlights of the ordinance was the implementation of the Specialized Care Services for Rare Diseases, which would have their budget for the composition of their teams, composed of a nurse, a nursing technician, a physician in charge, a geneticist, a neurologist, a pediatrician, a general practitioner, a psychologist, a nutritionist, and a social worker.

Although it is a turning point in the history of rare disease activism in Brazil, "Ordinance 199", as it became commonly known by interest groups, has not been translated into the intended transformations in ensuring access to comprehensive care for patients. Although there have been advances, such as the registration of seven health facilities as reference centers in the care of rare diseases in 2016[70], the general scenario continued to be marked by difficulty in access to treatment and limitations in the diagnostic and counseling network. On the orphan drugs, state purchases through judicialization represented extremely significant expenses for the Ministry

67. Ibid.
68. Araújo Neto and Teixeira, "Nuevos problemas de un nuevo sistema de salud," 9-10.
69. Brasil. *Portaria n. 199, de 30 de janeiro de 2014*. Institui a Política Nacional de Atenção Integral às Pessoas com Doenças Raras, aprova as Diretrizes para Atenção Integral às Pessoas com Doenças Raras no âmbito do Sistema Único de Saúde (SUS) e institui incentivos financeiros de custeio.
70. Moreira *et al.*, "Quando ser raro se torna um valor," 1.

of Health, considerably higher compared to drugs registered by health surveillance and incorporated into the national list of medications[71].

This scenario, in part, is due to Brazil's political and institutional instability since 2015, especially with the impeachment of President Dilma Rousseff in 2016. Since then, neoliberal platforms to dismantle the public machine and reduce state funding in social protection areas have taken priority in Brazilian politics, with emphasis on Constitutional Amendment 95 of 2016, known as the "spending cap amendment", which imposes severe limits on public investment in areas such as health and education[72].

In this context, patient organizations mobilize both to ensure access to treatment through their networking and judicialization[73] and to affirm the importance of family experience with rare conditions[74] and the need for interventions that consider the complexity of these diseases[75]. Thus, the formulation of specific legislation has not changed the moral economy of rare disease activism; on the contrary, it has reinforced the criticism of the rationality of metrics in decision-making and health planning and the role of associative work as a form of mobilization for the agendas of vulnerable groups and/or those neglected by public policies.

In the legal arena, the enforcement of the right to health and access to orphan drugs by rare disease patients continued to be problematic as seen in the trial of two processes regarding the matter in the Federal Supreme Court (STF) between 2016 and 2019. The processes put into the discussion the State's obligation to provide high-cost medications non-registered by the National Surveillance Agency (Anvisa) and destined for people suffering from

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71. Rosângela Caetano *et al*. "O caso do eculizumabe: judicialização e compras pelo Ministério da Saúde," *Revista de Saúde Pública* 54, no. 22 (2020): 1-11, http://doi.org/10.11606/s1518-8787.2020054001693.
72. Fabio dos Santos and Daniel Feldman, *O Médico e o Monstro: uma leitura do progressismo latino-americano e seus opostos,*.(São Paulo: Editora Elefante, 2021).
73. Maria Angélica Lima, Ana Gilbert and Dafne Horovitz, "Redes de tratamento e as associações de pacientes com doenças raras," *Ciência & Saúde Coletiva* 23, no. 10 (2018): 3247-56, DOI: 10.1590/1413-812320182310.147620181.
74. Daniel Campos, Martha Moreira, Marcos Antonio do Nascimento, "Navegando em águas: notas de uma pesquisa com famílias de crianças e adolescentes vivendo com doenças raras," *Ciência & Saúde Coletiva* 25, no.2 (2020): 421-8, DOI: 10.1590/1413-81232020252.11542018
75. Márcia Pinto *et al.,* "Cuidado complexo, custo elevado e perda de renda: o que não é raro para as famílias de crianças e adolescentes com condições de saúde raras," *Cadernos de Saúde Pública* 35, no. 9 (2019): 1-13, doi: 10.1590/0102-311X00180218.

serious illnesses[76]. One of these processes was moved by Alcirene de Oliveira, a woman from Juiz de Fora who suffered from severe hyperparathyroidism and demanded access to the drug Mimpara (Cinacalcet) to treat the disease in 2009[77]. After many years of a contested process, the Supreme Court decided in favor of Alcirene, but she reported that she did not receive the medication even after repeated subpoenas from the Court. In June 2018, Alcirene died without access to Mimpara.

Between 2016 and 2019, these two processes fueled intense debates on the State's responsibility to provide high-cost medication to patients with serious illnesses who do not have the financial means to buy it. The many deliberations by the Supreme Court presented great concerns to rare disease activists since the outcome directly threatened the guarantee of health care to ill people. The increasing participation of social networks in the public debate was noticeable during these years, as seen in the creation of the "*My life has no price movement*" page on Facebook[78], which mobilized testimonies and statements of patients, families, politicians, and experts to pressure the Supreme Court.

In 2017, World Rare Disease Day took as its motto "We are all rare", to give value and visibility to the cause of rare disease patients. In a world where social rights are fragmented and weakened by the neoliberal agenda, the struggle for recognition, the founding agenda of patient and family organizations, remains central to rare disease activism and a necessary cause to guarantee the right to health. Increasingly, the family experience gains relevance in this process of recognition of the right to care for rare disease patients, strengthening the argument that people need to be included more in health planning and interventions[79]. Also, activists and scholars have been pointing out the relevance of an agenda for incorporating innovative technologies into healthcare by state action, since it would "allow the

76. Ana Luísa Casseb, "A legitimidade da concessão judicial de medicamentos de alto custo na tutela individual: aspectos jurídicos, filosóficos e políticos" (Master diss., Universidade Federal do Pará, 2018).
77. Roberta Oliveira and Palmira Ribeiro, "Mineira que lutava para receber remédio de alto custo do Estado morre após seis anos esperando decisão que ainda tramita no STF," *globo.com*, July 7, 2018. https://g1.globo.com/mg/zona-da-mata/noticia/mineira-que-lutava-para-receber-remedio-de-alto-custo-do-estado-morre-apos-seis-anos-esperando-decisao-que-ainda-tramita-no-stf.ghtml.
78. See https://www.facebook.com/movimentominhavidanaotempreco/.
79. João Biehl and Adriana Petryna, "Peopling Global Health," *Saúde e Sociedade* 23, no. 2 (2014): 376-89, https://doi.org/10.1590/S0104-12902014000200003

negotiation and consequent price reduction of drugs, regulation, and surveillance of their uses"[80].

6. Final considerations

The history of the right to health in Brazil has nuances and complexities that go far beyond the process of its inclusion in the constitutional charter of 1988. The concepts and objectives of the actors who formulated the Unified Health System have encountered, since its implementation, different obstacles to its realization, in a scenario marked by structural problems —specially inequality— and contextual problems (such as the emergence of neoliberalism as a platform for late capitalism). The mobilizations around the right to health by rare disease interest groups are representative of the complicated process of structuring the Brazilian health system and its tensions. In turn, they are also indicative of the greater power obtained by the activism of users of the system, increasingly imbued with the capacity to transform specific agendas into public agendas.

In this article, we address processes still little discussed by Brazilian historiography, investigating how different meanings for the broader notion of the right to health placed in the Brazilian constitution gained materiality through the action of actors and institutions. Moreover, we observe how, in the case of rare diseases, this polysemy of the right to health has also been linked to the development of actors' agendas, from the recognition of the conditions as relevant health problems to the demand for a nationwide comprehensive care policy. The activism of patients and their allies has been a subject of controversy in the health field, among other reasons, because it represents perspectives, interests, and values that are fundamentally different from those guiding public health.

In agreement with recent anthropological readings on the field of global health and its relations with specific contexts and realities[81], we believe that this difference in perspectives between patients and public health, although

80. Marise Basso Amaral and Sergio Rego, "Rare diseases on the agenda for innovation in health: progress and challenges with cystic fibrosis," *Cadernos de Saúde Pública* 36, no. 12 (2020): 1-14, doi: 10.1590/0102-311X00115720, p. 9.

81. João Biehl and Adriana Petryna, ed., *When People Come First: Critical Studies in Global Health* (Princeton: University of Princeton Press, 2013); Megan Vaughan, Kalui Adjaye-Gbewonyo and Marissa Mika, ed. *Epidemiological Change and Chronic Disease in Sub-Saharan Africa: social and historical perspectives* (London: University College London, 2021).

conflicting at various times, is a necessary path to the construction of a health system that is closer to guarantee the right to health and to actively include users in the operation of the health structure. This process, however, demands a deep knowledge of the social dynamics and structural aspects of Brazilian health problems, an important attribution of the human sciences in the field of collective health. ▮

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