





## New problems of a new health system: the creation of a national public policy of rare diseases care in Brazil (1990s-2010s)

Nuevos problemas de un nuevo sistema de salud: la creación de una política pública nacional de atención de enfermedades raras en Brasil (1990-2014)

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**ABSTRACT** This study discusses actors and institution movements leading to the disclosure in 2014 of Resolution 199 by the Brazilian Ministry of Health, which establishes the National Policy for the Comprehensive Care of Persons with Rare Diseases. Taking as sources the mainstream newspapers, drafts law, and secondary literature on the subject, we begin our analysis in the early 1990s when the first patient associations were created in Brazil – mainly for claiming more funds for research on genetic diseases – and arrive at the late 2010s when negotiations for a national policy are taking place in the National Congress. Resolution 199 is part of an ongoing process and the path towards its disclosure and the complications that followed have given us elements to discuss contemporary aspects of the Brazilian public health. Based on the references of the history of the present time and the social studies of science, we argue that two aspects have been fundamental to creating a national policy: framing different illnesses within the terminology “rare diseases” and the construction of a public perception about the right of health which is guaranteed by the 1988 Brazilian Constitution.

**KEY WORDS** Rare Diseases; Public Policy; Comprehensive Health Care; Health System; Brazil.

**RESUMEN** En este trabajo se analizan los movimientos de actores e instituciones que llevaron a la promulgación, en 2014, de la Resolución 199 del Ministerio de Salud de Brasil, que establece la Política Nacional de Atención Integral a las Personas con Enfermedades Raras. Tomando como fuentes los principales periódicos, proyectos de ley y bibliografía secundaria sobre el tema, comenzamos nuestro análisis a principios de la década de 1990 con la creación de las primeras asociaciones de pacientes en Brasil, para reclamar fundamentalmente más fondos para la investigación de enfermedades genéticas, y llegamos a fines de la década de 2010 con las negociaciones para una política nacional. La Resolución 199 es parte de un proceso en curso, en el que el camino hacia la promulgación y las complicaciones posteriores nos dan elementos para discutir aspectos actuales de la salud pública brasileña. Sobre la base de la historia del tiempo presente y los estudios sociales de la ciencia, argumentamos que hay dos aspectos que han sido fundamentales para crear una política nacional: enmarcar diferentes enfermedades en la terminología “enfermedades raras” y la construcción de una percepción pública sobre el derecho a la salud, que se garantiza en la Constitución brasileña de 1988.

**KEY WORDS** Enfermedades Raras; Política Pública; Atención Integral de Salud; Sistema de Salud; Brasil.

## INTRODUCTION

In January 2014 a Resolution 199 of the Brazilian Ministry of Health instituted the National Policy for the Comprehensive Care of Persons with Rare Diseases [*Política Nacional de Atenção Integral às Pessoas com Doenças Raras*] which aims to guarantee health care for rare disease patients. Another goal mentioned in the Resolution is the decrease of morbimortality rates and secondary manifestations of such diseases, in addition to the improvement of life quality through health promotion, early diagnosis and treatment, reduction of disability and palliative care<sup>(1)</sup>. The creation of a specific legal regulation for this subject is the result of the historical framing of many illnesses in one broad spectrum and the mobilizations of many groups and institutions for social recognition of rare diseases as a public health issue.

The definition of a rare disease is volatile at some level. As we shall demonstrate, in the early 90s it referred to illnesses that caused degeneration or some kind of disability, mostly being framed as simply “chronic diseases” or “genetic disorders”. Since the beginning of the twentieth century, an epidemiologic approach has been used to define a pathology as rare disease. In the European Union, for example, a disease is considered rare if its incidence (number of cases per year) is equal or lower than one case in 2,000 people<sup>(2)</sup>. In the 2014 Resolution, the criteria were basically the same as indicated in Article 3: “For the purpose of this Resolution, a disease is considered rare when it affects up to 65 individuals per 100,000 people, in other words, 1.3 individuals per 2,000 people.”<sup>(1)</sup>

The very idea of creating a public policy for diseases that by definition targeted a restricted number of individuals represents a change of the public health epidemiologic way of thinking. How did groups and institutions negotiate with the authorities to create a consensus over the relevance of rare diseases for public health? This paper takes this historical problem to discuss the implementation of Resolution 199. We begin our analysis in the early 1990s when the first patient

associations were created in Brazil – mainly for claiming more funds for research on genetic diseases – and arrive at the late 2010s when negotiations towards a national policy are taking place in the National Congress.

After the creation of the Unified Health System [*Sistema Único de Saúde – SUS*] in the early 1990s – based on universal and integrated care – new demands were incorporated into the public health agenda through claims of medical societies, pharmaceutical companies, health technicians and civil society groups<sup>(3)</sup>. A greater capacity of actors and institutions outside the health field to mobilize and negotiate their agendas allowed to reframe those illnesses previously considered specific of philanthropy or the private sector.

In this paper, we discuss this “reframing process” of rare diseases through the actions of patients’ activism and political negotiation to create laws for protecting people suffering from those conditions. We argue that this process involved some main points: the very definition of which diseases could be considered rare; the search for law devices to get access to specific medication (“orphan pharmaceuticals”), known as judicialization; and debates towards the SUS foundation principle limitations (health as a right of all and a State obligation).

## ABOUT THE RESEARCH WORK

Our analysis is grounded on primary sources such as newspaper articles and news; Brazilian drafts law, federal laws and resolutions; and secondary bibliography. We dialogued with two authors from the social studies of science and the history of medicine, Karin Knorr Cetina, who argues that scientific facts are products of negotiations among different “epistemic cultures”<sup>(4)</sup>, and Charles Rosenberg, who understands disease as a result of “framing” processes based on biological, cultural and social elements.<sup>(5)</sup>

We based our research on two daily journals, *O Globo* and *Folha de São Paulo*, both edited in the Southeastern region of Brazil

(Rio de Janeiro and São Paulo). We used them as a convenience sample, because both journals are among those with the largest circulation in the country, in addition to having their full archive accessible on the web and consistently approach science and health issues. However, we do know this methodological option implicates demarcating the analysis to one single region of a complex country such as Brazil. We searched for “rare diseases” and “genetic diseases” between 1990 and 2018 to constitute our body of analysis.

Incorporation of genetic tests and therapies have been topics of sociological and anthropological studies in the last few years, mainly focusing on how the associations of patients mobilize actors and institutions to make their demands for public policies and funding available. In Brazil, as we shall discuss, this topic is improved by the constitutional right to health on which the SUS is based on and imposes on public health management vital issues related to budget limitations and to the distributions of resources in a very complex and huge country such as Brazil. In this sense, our historical approach focuses on the changes and continuities of political and social movements for rare disease care, not engaging in a sociological analysis.

## ACKNOWLEDGING NEW PROBLEMS

### Rare diseases in the 1990s public scene

In the early 1990s Brazilian public health was in a deep transformation. In the previous decade, a movement coordinated by a variety of physicians and others health field workers and researchers – in what was called the “Movement for the Health Reform” – proposed that the State had to provide full care for the population. This basic principle was set forth in the 1988 Constitution and remarked a basic principle of the SUS,<sup>(6)</sup> which would organize health in a decentralized network structure. Until then, the coverage of health care was limited by many factors,

mostly connected to the care of formal commerce and the organized urban working class.<sup>(7,8)</sup>

The public and universal characteristic of the SUS guided a new arrangement in priority-setting, with greater participation of the civil society which opened a space for lobbies and articulations for mobilizing specific group agendas. Hence, civil society sectors and pharmaceutical groups articulated advocacy and activist actions to bring the new diseases to the public scene. Diseases of great social appeal and stigma – cancer, tuberculosis and AIDS – were the targets of the first advocacy and activist groups in Brazil, where a long tradition of philanthropic work was already established.<sup>(9,10)</sup> Those groups started to operate with their specific agendas related to the interests, beliefs, expectations and possibilities of the patients, families and allies.

As discussed by Steven Epstein, studying the role played by these groups in public health policies is fundamental because they show us how patients elaborate strategies for legitimating their positions before the scientific community and the State,<sup>(11)</sup> which allows to “change the game rules transforming the definition of which counts as credibility in scientific research.”<sup>(11)</sup> Such relationship between the advocacy groups and the scientific community has a vital role in incorporating new diseases into the public health agenda. In the 1990s many claims of laypeople about rare diseases were about funding research on conditions such as amyotrophic lateral sclerosis (ALS) or Down’s syndrome. In general, rare diseases were framed based on the following elements: chronic clinical conditions, associated disabilities and social stigma resulting in prejudice against ill people. Social discrimination was the main point discussed by those who placed themselves as “speakers” of the patients, organizing associations and going public to put their claims forward.

It is possible to trace some of these speakers through *O Globo* and *Folha de São Paulo*, two of the biggest newspapers in Brazil. In 1990s these newspapers documented the creation of patients’ associations for Down’s

syndrome, Gaucher's disease, Cushing's syndrome and ALS. In August and September 1992, *O Globo* published a series of articles about the movement of 60 people suffering from ALS – a chronic disease that causes muscle degeneration and patient's depletion – for creating an association which represents their claims in the public sphere.<sup>(12,13,14,15)</sup> The leading person in the Association for Patients of ALS Syndrome was Nilcéia da Conceição, whose husband, Elias, suffered from ALS and had already lost arm and leg movement. Nicéia claimed, according to the articles, that lack of public funding for research and medical technology, in addition to the prejudice against ALS patients due to their “deformations” were the major problems to face.

According to the press, the ALS Syndrome Patients Association was successfully opened at the Deolindo Couto Institute of Neurology, in Rio de Janeiro city.<sup>(16)</sup> In 1998, another ALS institution was created in São Paulo, the Brazilian Association of Amyotrophic Lateral Sclerosis, directed by neurologist Acary Souza Bulle Oliveira. This institution, otherwise, adopted a profile of advocacy and scientific society, participating in the first epidemiological study on ALS in Brazil and organizing specialized congresses.<sup>(17)</sup>

Although with different profiles, both ALS patients' associations were representative of how rare diseases were approached in 1990s. In general, the patients' groups gathered in associations had in common the unfamiliarity with the disease and claims for funding on research. Furthermore, most of these associations had physicians in their boards or were installed in medical institutions. Such connections between patients and doctors constituted a specific type of activism characterized by laypeople's expectations and scientific knowledge, both articulated in a collaborative model of action or “hybrid collective model.”<sup>(18)</sup> Despite the similarities in the way those groups acted and were organized, there was not a notion that different degenerative rare diseases could be put together as a group, and no claims for health care were made; the agenda was focused on specific groups and

centered on research, only on rare occasions approaching the care problem.

There are evidences in our sources of only two specialized health care services for rare diseases: the National Network of Public Utility in Medicine, at Clementino Fraga Filho University Hospital, at Rio de Janeiro,<sup>(19)</sup> and a service at Dr. Boldrini Children's Center, in Campinas<sup>(20)</sup>. In public health policies, at the end of the decade, the debates on the Ministry of Health's funding for the acquisition of orphan medications started to gain more space in public health and the medical field, mainly due to the production of antiretroviral cocktails for HIV patients<sup>(21)</sup>. “Orphan medications” are those that are not of great commercial interest to pharmaceutical companies, either because of their high cost of production or the relatively low reach in the consumer health market. Given the low interest of companies in producing these drugs, the State plays a fundamental role in guaranteeing access to them by the patients, making them the target of lawsuits and claims for greater investments by the Ministry of Health.

### Rare diseases in the development of clinical genetics

The development of concerns about rare disease care is strictly connected with the history of clinical genetics, once the acknowledgment and the understanding of the natural history of those diseases depended on genetic diagnostic tools.<sup>(22)</sup> In Brazil, the genetic field has a long-way tradition, especially related to eugenics thoughts and practices, but clinical genetics has a more specific path that refers to the creation of the Brazilian Society of Clinical Genetics (SBG) in 1986 by a group of physicians concerned with the application of new genetic technologies and semiology to bedside medical practice.<sup>(23)</sup> However, in the early 1990s the SBG did not have much participation in the public debates on rare diseases.

This scenario changed after the mapping of the human genome in the mid-1990s and

the accession of the Brazilian genetics field to the Universal Declaration on the Human Genome and Human Rights, elaborated by UNESCO in 1997, which endorsed researches such as cloning and the use of new diagnostic tests.<sup>(24)</sup> In 1999, the Brazilian Society of Assisted Reproduction (SBRA) organized the Reproductive Medicine in the Twenty-First Century Congress, whose main topic was the possibilities of human genome researches and the ethical issues related to cloning. This event united specialists from many fields and institutions of Brazilian biomedical research, suggesting a bigger concern on the topic at the turn of the 21st century. One of the researchers' arguments for more funding was the concerns with respect to congenital diseases, as suggested by SBRA's president Edson Borges Junior:

We intend to correct possible genetic alterations in the embryo so as to avoid that the unborn baby develops a future disease after being implanted in the woman. For that reason, we want to discuss the legislation. Preventing a disease is a gain for all of us, however, the law does not authorize that.<sup>(25)</sup>

Even though a policy for rare and genetic diseases care was not on plate for the specialists, it is important to notice how the repercussions of the human genome brought genetic research and its diagnostic value to the public sphere. Therefore, two parallel movements were taking place in Brazil at the end of the century: on the one hand, diseases associated with disabilities and genetic etiology gained more space on the press through activist mobilizations claiming more research on the topic; on the other hand, the development of clinical genetics brought the debate towards genetic diseases and the value of an early diagnosis. At the dawn of the twenty-first century the public perception that health was a constitutional right led to more mobilizations and pressure on the government for a policy directed to rare diseases. Epidemiological studies on rare diseases were also fundamental in reframing

those conditions as a group of problems that share low-incidence rates but demanded costly care.<sup>(26)</sup>

### **The right to health and claim for a rare disease care policy in the 2000s**

At the turn of the twenty-first century the public understanding of the constitutional right to health was the basis for judicial actions, patients' claims and the proposal of public policies directed to rare disease care. Still under the repercussions of the genomic researches and the development of new diagnostic technologies, the Brazilian government promulgated a law and a Resolution (respectively in 2000 and 2001) about priority attendance to disabled people and the implementation of a national screening program for phenylketonuria, congenital hypothyroidism, sickle cell diseases and cystic fibrosis.<sup>(27,28)</sup>

The elaboration of a screening program for such diseases through the "heel test" was considered an achievement of the public health system and for the recognition of these illnesses as a public health issue.<sup>(26)</sup> Even though the Resolution initiated the dissemination of genetic tests, activists and scientific community's demands for funding and the creation of more structures and policies for rare disease care exposed the limitation of the SUS at that time.

The 13th Brazilian Congress of Clinical Genetics was held in Aguas de Lindoia (São Paulo state) in April 2001, in which the practical implications of new researches in the genetics field were discussed. According to the president of the Brazilian Society of Clinical Genetics, Carlos Ruchaud, more public funding was necessary for research and the acquisition of diagnostic technologies for genetic diseases such as Gaucher's. Another highlighted point was the demand for the acquisition of orphan medications by the federal government, which was guaranteed by the right to health; although Ruchaud acknowledged the difficulty of maintaining a buying policy for those drugs, suggesting

partnerships with universities and pharmaceutical companies.<sup>(29)</sup>

Such difficulties were related to the priority-setting models for public health policies in Brazil, where since the early 1920s have based decision-making in the idea of targeting diseases with greater population impact, be it in a sociopolitical sense<sup>(3)</sup> or an epidemiological one. Additionally, Brazilian public health has a long tradition of actions directed to specific diseases based on a “campaign model”, which means articulating vertical strategies in a well-defined period of time<sup>(7)</sup>. Diseases which did not have such an appeal tended to be reframed by physicians and laypeople based on socioeconomic and rhetorical arguments, as it happened with cancer and cardiac diseases.<sup>(30,31)</sup> In this sense, creating directed policies for conditions that would not fit in the long tradition of public health concerns is a challenge for the rare disease care movement that demanded the creation of a rationale that could articulate the right to health to the patients’ needs.

A debate involving São Paulo state Secretary of Health Alberto Hideki Kanamura and the president of the Paulista Association of Gaucher’s Disease Patients [*Associação Paulista dos Portadores da Doença de Gaucher*], Pedro Carlos Stelian illustrates the controversy over public health coverage and budget limitations. In an opinion section of *Folha de São Paulo*, Kanamura argued that the constitutional definition of health as a “right of all and duty of the State” was unrealistic to the Brazilian society and should be relativized, despite being a “social conquest”. According to him:

In a country where people still die of malnutrition, lack of potable water or of pure ignorance towards primary sanitary concepts, it is hard not to question the decisions which direct the health budget to treat rare diseases, when the same resources could benefit thousands who live disease as a rule. [...] Spending money in large scale treatments is a decision that can’t be taken without evaluating the cost/benefit relationship and

a proper analysis of the cost/effectivity relationship.<sup>(32)</sup>

Kanamura’s ponderations referred to recent judicial decisions in favor of patients who claimed for the coverage of the public cost of their treatments by the SUS based on the constitutional right to health.<sup>(33,34)</sup> Kanamura claimed that the judicial system did not take into account the budget limitations of the health system and that spending so much money in the acquisition of medication and diagnostic technologies for specific rare diseases would affect other actions. Furthermore, the “right to health” principle allowed non-legitimated procedures to be experimented using public resources. He proposed a review of the constitutional text, anticipating the bankruptcy of the health system if there were no changes to the current structure.

Weeks later, Pedro Stelian wrote an opinion article in response to Kanamura’s critique to the “right to health”. His analysis was based on a “patient’s perspective” and made clear the difference between his point of view and the Secretary’s. Stelian criticized the management’s perspective and defended the constitutional text and its implications to the public administration. Kanamura’s dilemma was false, Stelian argued, for rare and common disease health care did not consist in a dichotomy. It was the health managers’ obligation to use resources in such a way that no patients were “forgotten”:

There is no point in medicine providing advances in disease treatments if they cannot reach patients. If the cost is high, it is up to the public health managers not to abandon patients – be they majority or minority – but seek alternatives to reduce costs. Otherwise, we slip into the prioritization of what is politically more interesting to the managers. After all, does the issue of caring for what is rare matter less because it yields less votes? Is a rare disease carrier the same, too, as a rare vote?<sup>(32)</sup>

Such debate demonstrates some critical aspects to the constitution of rare and genetic

diseases in public health problems. First, it is interesting to observe how different social arenas frame diseases and health issues from specific perspectives and expectations and how this process of the meaning of illness affects the functioning of public health. It is not the socio-historical analysis to evaluate the validity of the arguments of the actors, but to observe that the managers' perspective, guided by aspects of population epidemiology and by principles of public administration, is quite different from the patients' concerns, directed towards the individual experience of the disease.

However (and here the second point is posed), the different conceptions of a problem originated from a shared epistemic culture,<sup>(4)</sup> that is, the same configuration of a scientific fact. Rare diseases were seen in the 1990s as specific entities that did not constitute a group; further discussions about the limitations of the health system focused on attending all the conditions with a low epidemiological incidence in the same umbrella, considering them as part of the same problem to the public administration.

Gradually, the very articles published in the press have come to use the term "rare diseases" more than the specific terminologies. In 2005, in the context of articles commenting on judicialization as a mechanism for compliance with the State's obligation to finance health, *Folha de São Paulo* reported the case of a woman suffering from Heck's disease and her difficulties in accessing medications and periodic exams. The episode was used to exemplify the broader problem of people with rare diseases, especially regarding the acquisition of orphan drugs.<sup>(35)</sup>

As a result of the mobilizations in the public scene, another group of actors gained prominence: the private industries and laboratories. In 2007, an article in the newspaper *O Globo* interviewed Rogério Vivaldi, an endocrinologist of the Genzyme laboratory, the main name in the genetic research market in the country. In the doctor's view, the problem of care for people with rare diseases in Brazil was greater access to orphan drugs. According to him, the Ministry of

Health should expand the list of drugs with exceptional imports: "The list of exceptional drugs must be updated [...]. For every dollar invested in research products or donations in Brazil, Genzyme spends US \$ 200,000 with import taxes and other taxes."<sup>(36)</sup>

A key difference in the relationship between public health and patients, and industry and patients is that, to a certain extent, the agendas placed by the market are closer to the patients' expectations, concerned about the possibility of using (consuming) medicines for the treatment of diseases. In this way, pharmaceutical companies and laboratories have gained greater prominence in the articulations for the attention of the carriers of rare and genetic diseases than the public health itself. In some cases, the state was presented as an antagonist of a narrative in which patients sought treatment, but bumped into government bureaucracy.

In 2009, a new debate gained space in the print media. Between April and May, the Federal Superior Court (FSC) held hearings to establish jurisprudence in cases of health judicialization, especially with regard to the access to orphan drugs. The first position presented by *Folha de São Paulo* was that of the then Health Minister, José Gomes Temporão, who raised concerns about the pressure to incorporate procedures without the proper technical consideration of the sector. For Temporão, it was necessary to establish clearer parameters for the inclusion of technologies and drugs in the scope of state financing, in order not to distort the essence of the health system and lose investments in strategic areas.<sup>(37)</sup>

In a later edition of the newspaper, Marcos Bosi Ferraz, a professor at the Faculty of Medicine of the Federal University of São Paulo and a member of the Brazilian Academy of Medicine, presented a different position than that of the Health Minister. For Ferraz, it was necessary to establish priorities for the actions of the sector considering the following criteria: "more important diseases, more frequent, more serious, with more suffering and greater chance of prevention"<sup>(38)</sup>. In this sense, while claiming that disease

carriers could not be left unprotected, he stated that it was necessary for the government to recognize its limitations and to assume “in a clear and transparent way some ‘no’.”<sup>(38)</sup>

On the same page, another dimension of the debate was presented through an article written by two lawyers, Andrea Salazar and Karina Grou. The authors approached the question from a closer angle to the point of view of patients, arguing that the judiciary’s action would guarantee the fulfillment of the right to access the weakest link in the scale, the users of the health system. For Salazar and Grou, the intervention of the FSC in matters of access to orphan drugs did not consist in a breakdown of the sovereignty of the executive power, but rather the essence of the balance of the three powers.<sup>(38)</sup>

The inclusion of more actors and groups in the discussion on the access to orphan drugs and care for people with rare and genetic diseases allowed to reposition the issue in the public agenda. If, in the early 1990s, the patients mobilized almost in isolation from the claim for investment and research; in an interval of almost twenty years the rare and genetic diseases were reconfigured as a subject of great interest to the health system. This shift went directly through the public perception of the right to health and the strengthening of mobilizations by pressing the State to increase funding for the purchase of orphan drugs.

In the decade of 2010, another arena entered the dispute, also with specific agendas: politics. The assimilation of patient and pharmaceutical guidelines by political actors made the discussion of rare and genetic diseases broader, proposing projects for a national policy of care.

### **Rare diseases in the political arena: the genesis of a national care policy**

In 2011, federal deputy Marçal Filho of the Brazilian Democratic Movement [*Movimento Democrático Brasileiro – MDB*] presented to Congress the first draft law specifically aimed at patients with rare diseases. In the project

presented to the Chamber of Deputies, the State’s obligation to provide prescribed drugs for the treatment of rare and serious diseases, based on the premise of the universal right to health,<sup>(39)</sup> is reaffirmed.

Although it was important and urgent to establish parameters to facilitate the purchase of orphan drugs for patients with rare diseases, the draft law did not provide a clear qualification of what conditions would be contemplated, without mentioning epidemiological data or even referring to classifications of international health agencies, such as the WHO. According to Article 3 of the draft law, the dispensation (guidance, prescription, and supply) would be based on medical reports. Thus, it would be up to the medical profession, not the public health, to define which diseases would be classified as “rare and severe”, and the Ministry of Health should establish “the rules regarding the administrative procedures to be observed to receive the medical prescription of the analysis of the reports and for dispensing the drug directly to the patient.”<sup>(39)</sup>

That same year, two bills aimed at patients with rare diseases were submitted to the Chamber of Deputies. Mara Gabrielli, a deputy elected by the state of São Paulo and affiliated to the Brazilian Social Democracy Party (*Partido da Social Democracia Brasileira – PSDB*), submitted draft law No. 1656, proposing that patients with neuromuscular diseases with motor paralysis should receive priority from the SUS to obtain drugs and equipment necessary for treatment.<sup>(40)</sup> Gabrielli’s project was different from Marçal Filho’s project, and articulated epidemiological and clinical data from national and international agencies about neuromuscular diseases, pointing out the central justification for the approval of the law in the occurrence of respiratory complications in the patients.

The main difference of Gabrielli’s project, however, lies in the explicitness of the allies involved in proposing the law. At the end of the legislative text, she said:

with the support of internationally renowned associations involved in the treatment

of these diseases, such as the Brazilian Association of Amyotrophic Lateral Sclerosis – ABRELA, the Brazilian Association of Muscular Dystrophy – ABDIM and the Brazilian Institute of Sleep.<sup>”(40)</sup>

In addition, according to the author of the project, public consultations were carried out with neuromotor disease patients, in order to guarantee the participation of the actors most interested in the process. Described as a tool for scientific and social legitimation of the text, the connections presented by the author also allowed us to visualize the movement in progress since the previous decade: actors from different arenas establishing dialogues in order to transform rare diseases into a public health agenda.

The third project of 2011 was submitted by the deputy of Rio de Janeiro, Jean Wyllys, of the Socialism and Freedom Party [*Partido Socialismo e Liberdade* – PSOL]. More comprehensive than the other two, Wyllys’ text is directed to the treatment of patients with rare diseases by the SUS, including medical care, the purchase of orphan drugs, the provision of specific equipment, and the “adequacy of home care units to people with rare diseases.”<sup>”(41)</sup> The main novelty in Jean Wyllys’ text was the first comprehensive definition of rare diseases based on epidemiological and clinical data. The Article 2 of the draft law states that “a person with a rare disease is considered to be a person with a debilitating and / or incapacitating condition whose prevalence in each 100,000 thousand inhabitants corresponds to 65 cases.”<sup>”(41)</sup>

The definition of rare disease using prevalence data was based on the guidelines of the World Health Organization and the experience of care policies of the European Union. The European concept of rare disease, according to the project, highlighted the pathological transversality of the term, involving genetic, degenerative, autoimmune, infectious and oncological conditions.<sup>”(41)</sup> This broader characterization allowed the design of health care actions at several levels, as some diseases would have their control centered on primary care, others on high

complexity, etc. Another important aspect of the framework presented in Jean Wyllys’ project concerns the impact of rare diseases on infant mortality, which is the second leading cause of death in this group.

In addition, the proposal referred to other legislation enacted in previous years by the Brazilian government. The main highlight was given to Federal Decree No. 6949 of August 25, 2009, which integrated the Convention on the Rights of Persons with Disabilities into the Brazilian legal system. The Convention, signed in New York in 2007, aimed to promote, protect and guarantee the full and equal enjoyment of human rights by persons with disabilities. The Brazilian legislation of 2009 addressed two articles of the document signed in 2007, “Health” (Article 25) and “Empowerment and Rehabilitation” (Article 26).<sup>”(42)</sup>

The three projects submitted in 2011 represented distinct concerns and expectations in the debate on care for rare disease patients. Marçal Filho’s text was directly linked to the scenario of judicial mobilizations that marked the 2000s, as an attempt to satisfactorily respond to the topic of judicialization by adding more actors and institutions to the discussion. Gabrielli’s proposal, in turn, was aligned with the articulations of patient associations, operating since the 1990s and seeking to increase the strength of their demands for research and access to orphan drugs. Wyllys’ project ultimately represented a broader health care plan, probably linked to a deputy mandate platform, which legislates with emphasis on the agendas of the groups considered minorities.

The submission of the three projects represented the beginning, not the end, of negotiations and mobilizations in the political arena. Internalized in the government’s decision-making agenda and with a well-defined visibility flow, the projects waited for a window of opportunity. The three of them went through evaluations and changes of text in the National Congress, not receiving approval of their final texts. Nevertheless, they served as the political basis for proposing legislation specifically aimed at patients with rare diseases. In addition, the debates on the

public stage, with the press as an important vehicle for positioning, have gradually been shifted towards the demand for a comprehensive care policy, extrapolating the issue of orphan drugs and the development of research.

In the activism arena, an important development concerned the National Policy on Comprehensive Care in Clinical Genetics,<sup>(43)</sup> approved in 2009, which proposed the creation of a health care network for genetic diseases, from primary health care to specialized care in clinical genetics. In 2011, an encounter in Cuiabá gathered many associations to discuss the implementation of the “genetic Resolution” and the problem of genetic diseases care. In parallel to the Congress program, the first Meeting of Rare Disease Patients’ Associations was held. The goal was to make room for associations to exchange experiences, and to discuss their problems and achievements. This meeting represents an important precedent for the creation of a national policy for rare disease care.<sup>(44)</sup>

In April 2012, representatives of the Ministry of Health, medical societies and patient associations met in Brasília to discuss the creation of a national policy for the care of patients with rare diseases. The president of the Brazilian Society of Medical Genetics, Marcial Francis Galera, believed that the new project could be based on the National Policy on Comprehensive Care in Clinical Genetics. According to Galera, the use of the 2009 text would be advisable because 80% of the rare diseases have a genetic etiology, and place the same emphasis on the demands for other types of rare conditions, emphasizing the early diagnosis.

Conflicts about the implementation of a national policy for rare diseases care involved, among other points, the matter of financing health care. As we mentioned in the early 2000s, the “cost-effectiveness” argument constantly confronted the “right to health” principle that mobilizes Brazilian health system. Compromising large portions of the Ministry of Health’s budget, some politicians and technicians claimed, (forecasting an increase of R\$ 300 million only with the purchase of orphan

drugs) was an obstacle to the proposal’s effectiveness.<sup>(45)</sup>

The groups interested in the creation of the new national policy added more and more allies in the attempt to approve the measure in the National Congress. Since 2012, new actors with great visibility have entered this arena. Former soccer player and federal deputy Romario has actively engaged in the cause of rare and genetic diseases, partly because of his own personal motivation (his daughter is a carrier of Down’s Syndrome). Along with Jean Wyllys, Marçal Filho, Mara Gabrielli and Senator Eduardo Suplicy of the Workers’ Party (*Partido dos Trabalhadores – PT*), he has become a relevant voice in the dissemination of the agendas of the actors and institutions dedicated to the creation of a policy of comprehensive care.

In the scientific field, research in the field of genetics and public health increasingly emphasized the importance of the issue for the Brazilian society, highlighting the impact of rare conditions in large cities and the polarized epidemiological transition. In this dimension, the Fernandes Figueira Institute, a unit of the Oswaldo Cruz Foundation, through its Department of Medical Genetics, plays an important role in the production of research and acting in the public sphere, in order to give visibility and credibility to the speech of politicians, patients and pharmaceutical companies.<sup>(23)</sup>

In March 2013, Interfarma, an association of all pharmaceutical companies in the country, organized a seminar on rare diseases in Rio de Janeiro, bringing together members of the scientific community, government regulatory agencies, the Legislative Branch and patient associations.<sup>(46)</sup> In addition, it contracted a research on the subject by the American company IMS Health, in order to show the impact that the groups’ proposal would have on Brazilian public health and the well-being of the target population.

The study commissioned by Interfarma resulted in a report, published in 2013, titled “Rare Diseases: Contributions to a National Policy”. In the paper, it is argued that the attention to rare conditions would have

two dimensions: the treatment and purchase of orphan drugs. The report considered early detection as part of the therapeutic process, not the prevention of the disease. In this sense, the need to reverse the late diagnosis was considered the central point of the proposal developed by Interfarma.<sup>(47)</sup> Finally, the national policy should be based, according to the document, on three dimensions: organization of assistance; adoption of registration mechanisms; incorporation of orphan drugs into the SUS.<sup>(47)</sup>

These spheres of action would have the public service as a point of support, but would also rely on the performance of private initiative, such as the pharmaceutical industry itself. Citing the European experience, the document concludes that the implementation of the new policy could change the scenario of care for people with rare diseases, particularly regarding access to medicines:

Based on the group of rare diseases that have more organized data – mucopolysaccharidosis I, II and VI, Gaucher, Niemann-Pick type C, pulmonary arterial hypertension, Fabry, hereditary angioedema, acromegaly and familial amyloid polyneuropathy – Interfarma predicts that the implementation of an official program could benefit more than 5,000 patients with medication. This estimate represents an increase of 75% in relation to the population served today.<sup>(47)</sup>

The document prepared by Interfarma expressed, in general, the conjunction of expectations of different social arenas on the issue of care for people with rare diseases. The perception that the principle of the universal right to health would legally imply access to orphan drugs by the patients made this agenda central to national policy advocates. At this point, it is interesting to note how an “epistemic culture” about rare diseases was formulated as groups of different expectations, beliefs, and interests organized a common understanding of a scientific fact (“carriers of rare conditions”). The sharing of this agenda gave more

strength and allowed the working group to elaborate a proposal for the care of the sick.

Still in 2013, senator Vital do Rego, of the Brazilian Democratic Movement Party [*Partido do Movimento Democrático Brasileiro* – PMDB], submitted drafts law No. 530, which provided for the establishment of the National Policy for Rare Diseases, focusing on the creation of a differentiated regime for the purchase of orphan drugs through the SUS<sup>(48)</sup>. Another aspect raised by the project submitted by Rêgo concerns the expansion of assistance to patients with rare diseases in Brazil, initially established by Resolution No. 81 of January 20, 2009, which established the National Policy for Comprehensive Care in Clinical Genetics. According to the text, the Resolution did not contemplate diseases of infectious, inflammatory and autoimmune origin. This would lead to some serious problems for the patients with these diseases: lack of clinical protocols, lack of trained professionals, lack of investments in research and difficulty accessing medications.

The project of Vital do Rego was discussed in the National Congress, with slower referrals than the groups involved in the cause demanded. The pressure from the actors and institutions led the Ministry of Health to implement the policy at the ministerial level, through Resolution No. 199, dated January 30, 2014, which established the National Policy for the Comprehensive Care of Persons with Rare Diseases, approved the Guidelines for Comprehensive Care for Persons with Rare Diseases under the SUS and instituted financial incentives.<sup>(1)</sup>

The document provided attention to patients with rare diseases in two axes: the first one directed to congenital anomalies, intellectual deficiency and innate errors of metabolism; while the latter dealt with infectious, inflammatory, and autoimmune conditions. He also organized the care line, defining what was appropriate for each level of care and how the network would be structured. One of the highlights of the concierge was the implementation of Specialized Care Services for Rare Diseases, which would have its own budget for the composition of its teams,

made up of nurses, nursing technicians, medical doctors, geneticists, neurologists, pediatricians, general clinicians, psychologist, nutritionist and social worker.

## FINAL CONSIDERATIONS

After Resolution No. 199, rare diseases care did not improve as much as expected. Since 2015, Brazilian politics and public policy has entered into a crisis status that impacted directly the SUS and its actions. A milestone to this scenario is the approval of an amendment to the Constitution establishing a limit of budget increase for areas such as health, education and public security.<sup>(49)</sup> In general, the public health sector is suffering the discontinuity of funding and programs devoted to specific problems such as HIV, rare diseases, among other.

In 2017, a sub-commission of rare diseases was created in the Brazilian Senate to discuss the implementation of the National Rare Diseases Care Policy, which was considered very difficult due to the political and economic scenario. This sub-commission worked for a year listening in public hearings to associations and representatives of the Executive Branch, an alternative draft law circulating within the commission itself circulated in the group, but from the understanding that this project was vague, did not detail important issues, not representing any advance, the

movement was to support the project 56/2016 that reaffirmed Resolution 199,<sup>(50)</sup> understanding that it was already on a walk and thus, also respected the work that had been done in the Chamber of Deputies.

Rare disease care is a very interesting example of how new agendas are raised based on a reformulation of the public health system. The openness proposed by the SUS in decision-making of public policy and the "right to health" principle were fundamental for many groups gathering their specific concerns and interests and taking them to the public sphere. However, if the new system allows these sorts of mobilizations, it also imposes new problems on health management and funding, demanding a closer relationship between public health experts, political field, activists, press and scientific community.

The history of rare diseases care traces a very different picture in the construction of the public health agenda, in which health happens to be also the subject of smaller groups. The nationalist discourse gives way to the process of medicalization, revealing problems of urban layers that have an entry into the health market, claiming their right of access to medicines necessary for therapeutic and palliative treatment. In fact, the 2014 Resolution was a starting point for new mobilizations and negotiations concerning the attention to those individuals with rare conditions, both within the political field and in daily life.

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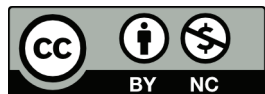
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