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PUBLIC POLICIES AND THE CHALLENGE OF ACCESS TO TREATMENT FOR PATIENTS WITH RARE DISEASES

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ABSTRACT
Introduction: Rare diseases are characterized by their low prevalence in the population, but they present significant challenges for patients, families, and the health system. These individuals often face considerable barriers in accessing specialized medical care, including difficulties in accessing appropriate treatments and surgical procedures, when indicated. Objective: to describe the challenges encountered and the implementation of public policies to guarantee access to surgeries for patients with rare diseases. Method: This is a literature review carried out in the electronic databases of Latin American and Caribbean Literature on Health Sciences (LILACS), Medical Literature Analysis and Retrieval System Online (MEDLINE/PubMed) and Scientific Electronic Library Online (SciELO). The inclusion criteria were complete articles, with text available online, published between 2014 and 2024. The search for studies was carried out between the months of May and July 2024. Results: Regarding the main results, it was found that patients with rare diseases face considerable difficulties in accessing treatments, especially those of a surgical nature. One of the main obstacles is the shortage of specialized professionals, which, combined with the high cost of treatments, makes access difficult. Conclusion: The complexity of access to treatment and surgery for patients with rare diseases is a significant problem, exacerbated by the absence of clear clinical protocols and the high cost of treatments. Public policies aimed at rare diseases are fundamental and need to be expanded to ensure improvements in access to treatment.

Keywords: General surgery. Rare diseases. Human Rights.

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INTRODUCTION

Rare diseases, or also called orphan diseases, are health conditions that affect only a small part of the population. A rare disease is considered to be one that affects less than 65 out of every 1000 people. However, countries may adopt different criteria to classify rare diseases, considering epidemiological and clinical factors (Brazil, 2021).

When it comes to characteristics, rare diseases can be chronic, progressive, and can cause serious complications, compromising the quality of life of patients. They can also affect any part of the body, and may have genetic origins, although they can also be caused by environmental or infectious factors (Santos and Almeida, 2017).

Due to their low prevalence in the population, these diseases receive less attention in research, delaying the options for diagnosis and appropriate treatment (Santos and Almeida, 2017).

Access to the treatment of rare diseases has a history marked by significant challenges, because until the middle of the twentieth century, these conditions were neglected, due to the lack of medical and scientific knowledge. The theme began to gain momentum in the 1980s, when the United States enacted the *Orphan Drug Act*, a pioneering legislation that aimed to encourage research and development of drugs for rare diseases, offering benefits, such as exemption from fees and financial support for pharmaceutical companies (Santos and Oliveira, 2019).

Currently, there are several rare diseases identified, which can be classified based on their origin, symptoms, and affected body systems. Some examples are: genetic diseases (cystic fibrosis, Huntington's disease, Duchenne muscular dystrophy, etc.), autoimmune diseases (Sjögren's syndrome, systemic lupus erythematosus, etc.), neurological diseases (amyotrophic lateral sclerosis, Friedreich's ataxia, Rett syndrome, etc.), endocrine diseases (Klinefelter syndrome, Turner's syndrome, etc.), among others (Pereira et al., 2021).

According to the World Health Organization (WHO), there are approximately 6,000 to 8,000 different types of recognized rare diseases. This variety, associated with low prevalence, hinders the development of early and effective treatments and diagnosis strategies. Thus, the National Policy for Comprehensive Care for People with Rare Diseases in the SUS was an important milestone in the attempt to integrate and improve access for the part of the population affected by these diseases (Pereira et al., 2021).

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In this scenario, public policies represent a fundamental pillar for the promotion of well-being and for ensuring universal access to health services. Some studies show that in the absence of effective public policies, they affect the implementation of actions, impairing the quality of life of patients, early treatment, and increasing the suffering of this population (Alves, 2020).

In view of the above, the study had the following problem: What guarantees of access and public policies for the treatment of patients with rare diseases?element. It is believed that the study is of great importance, due to the increasing number of diagnoses and the complexity of treating these conditions.

The study is justified by the need to improve access to treatment and the urgency of the practices ensured by public policies, making them efficient and inclusive, ensuring coverage and support for these patients.

Therefore, the general objective of this study was to describe the challenges encountered and the implementation of public policies to guarantee access to surgeries for patients with rare diseases

METHOD

TYPE OF STUDY

This is an integrative review, with the objective of gathering and synthesizing the results of scientific publications related to patients with rare diseases who require a surgical approach.

The integrative review consists of the broadest methodological approach to reviews, configuring itself as a type of review that brings together the analysis of multiple published studies, of different methodologies, with the purpose of obtaining a deep understanding of a given phenomenon, based on previous studies. (Souza, Silva and Carvalho, 2010).

Combining data from the theoretical and empirical literature, incorporating a range of purposes, contributing to generate a consistent and understandable panorama for: definition of concepts, review of theories and evidence, discussion about research methods and results, pointing out knowledge gaps that need to be filled with the realization of new studies (Souza; Silva; Carvalho, 2010).

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STAGES OF INTEGRATIVE REVIEW

The present review was conducted in stages, consisting of the adaptation of the Preferred Reporting Items for Systematic Reviews and Meta Analyses (PRISMA) recommendations. These are: the identification of the theme and selection of the research question for the elaboration of the integrative review; establishment of criteria for inclusion and exclusion of studies; definition of the information to be extracted from the selected studies, in order to allow the categorization of the findings; evaluation of included studies; interpretation of the results; and preparation of the presentation of the review/synthesis of knowledge (Prisma, 2018).

To guide the review, the following question was formulated: What guarantees of access and public policies for the treatment of patients with rare diseases?

All scientific productions that had, in their content, rare diseases associated with surgeries were considered. Articles published without time restriction in English, Portuguese or Spanish were included. The exclusion criteria adopted were theses, dissertations, review articles, and those repeated in the databases explored.

The search for studies was carried out between May and July 2024 in the electronic databases of Latin American and Caribbean Literature on Health Sciences (LILACS), Medical Literature Analysis and Retrieval System Online (MEDLINE/PubMed) and Scientific Electronic Library Online (SciELO).

The search strategy used in the databases used the following descriptors in English, recognized by the Health Sciences Descriptors (DeCS) and Mesh systems: (General surgery) AND (Rare Diseases) AND (Human Rights OR Right to Health) and (General surgery) AND (Rare Diseases).

The search strategies used are shown in the table below:

Chart 1 – Search strategy: Integrative Review

DATABASES SEARCH STRATEGY	SEARCH STRATEGY
LILACS / PubMed / SciELO	(General surgery) AND (Rare Diseases) AND (Human Rights OR Right to Health)
LILACS / PubMed / SciELO	(General surgery) AND (Rare Diseases)

Source: Prepared by the author (2024).

Selection of studies

Initially, the identified studies were evaluated through the analysis of the titles, discarding duplicate searches in the databases, in addition to those that had no implication with the objectives of this review. Then, the abstracts were read, selecting those that did

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not meet the inclusion criteria defined in this research. Finally, through the full reading, the studies that dialogued with the objectives of the research undertaken here were selected, so that it culminated in the final sample included in this integrative review.

All studies identified through the search strategy were initially evaluated through the analysis of titles and abstracts, following the eligibility criteria. Those who met the criteria to answer the question of this research were selected.

The selection of studies was first carried out by a single reviewer, and the doubts regarding them sought to be resolved with a second evaluator.

Extraction and Analysis of Data/Content

To characterize the articles included in the review, the following information was extracted: title, year, place of the study and relevant information related to the theme.

To compile and present the results about the difficulties, strategies and propositions, the technique of Discursive Textual Analysis proposed by Moraes and Galiazzi (2006) was used.

RESULTS

A total of 87 publications were found in the databases, of which 13 from LILACS, 68 from MEDLINE/PubMed and 6 from SciELO were from the database. After deleting the duplicate studies in the database, a total of 86 publications were published to be evaluated, following the defined parameters. Of these, 17 studies demonstrated that they met the inclusion criteria, as well as having the necessary elements to answer the proposed guiding question. The entire selection process that resulted in their inclusion is presented in the following flowchart (Figure 1) and results (Table 1).

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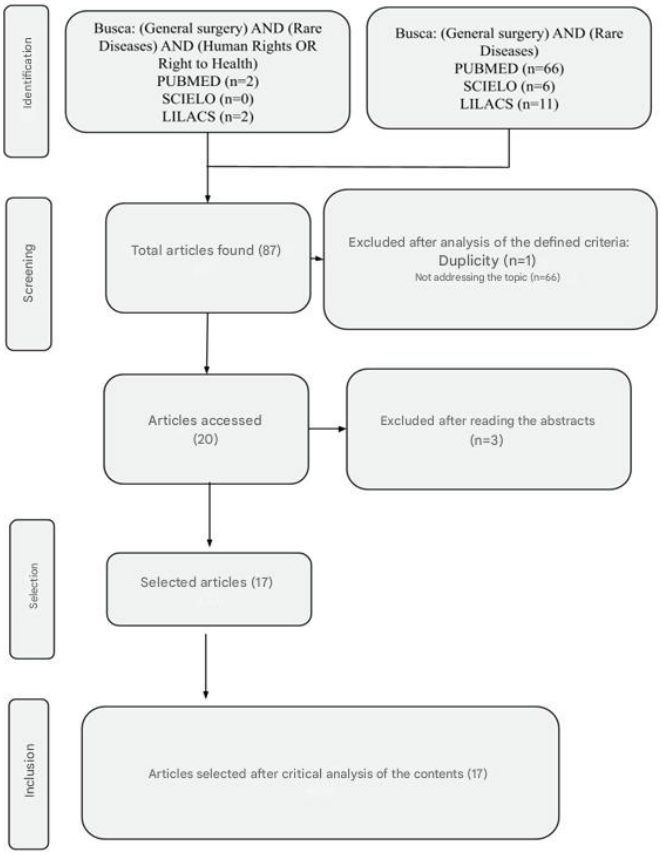
Figure 1 - Flowchart of the strategy for searching and selecting articles



Source: Prepared by the author (2024).

Below is a description of the studies found with their respective locations and years of publication, so it is observed that most of the studies were written in the years 2023, with only two written in the year 2014, 2018, 2020 and 2022.

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Chart 2 - Description of the studies found with their respective years of publication and place of publication.

AUTHOR	TITLE	YEAR/DATABASE	LOCATION OF STUDY	RELATIONSHIP WITH THE THEME
Samson SL, Nachtigall LB, Fleseriu M, Jensterle M, Manning PJ, Elenkova A, et al.	Durable biochemical response and safety with oral octreotide capsules in acromegaly	2022/PUBMED	USA	The article deals with acromegaly, a rare disease characterized by excess growth hormone due to a benign tumor in the pituitary gland, with the standard treatment being surgical removal of the pituitary tumor as a primary option to control the disease.
Schumacher FR, Schmit SL, Jiao S, Edlund CK, Wang H, Zhang B, et al.	Genome-wide association study of colorectal cancer identifies six new susceptibility loci	2015	USA	The main focus is on colorectal cancer genetics and identifying new susceptibility sites through genomic studies. It treats scientific advances in improving treatment.
Hirai F, Yamanaka T, Taguchi K, Daga H, Ono A, Tanaka K, et al.	A multicenter phase II study of carboplatin and paclitaxel for advanced thymic carcinoma: WJOG4207L	2015	Japan	Thymoma is a rare tumor originating from the epithelial cells of the thymus, associated with myasthenia gravis. Advances in treatment and diagnosis are essential.
Ann-Cathrin Koschker, Bodo Warrings, Morbach C, Seyfried F, Jung P, Dischinger U, et al.	Effect of bariatric surgery on cardio-psycho-metabolic outcomes in severe obesity: A randomized controlled trial	2023	Germany	The study involves the evaluation of super-obese patients, allowing access to treatment until surgery.
Hirschfield GM, Shiffman ML, Aliya Gulamhusein, Kowdley KV, Vierling JM, Levy C, et al	Seladelpar efficacy and safety at 3 months in patients with primary biliary cholangitis: ENHANCE, a phase 3, randomized, placebo-controlled study	2023	Not informed	Primary Biliary Cholangitis is a rare, chronic, autoimmune liver disease that causes destruction of the bile ducts. Initial treatment with ursodeoxycholic acid may be insufficient, requiring complementation with experimental therapies and surgeries. However, access to effective treatments remains a significant challenge for PBC patients, highlighting the ongoing need for research and development of new therapeutic options.
Gouveia VA, Fernandes BFS, Frota ERC, Cunninghan MCQ and S,	Clinical and Laboratory Characteristics of Cryptococcosis in Patients Admitted to the Emergency	2018	Brazil	Cryptococcosis is a rare systemic mycosis caused, mainly affecting immunosuppressed patients. Treatment requires accurate diagnosis through tests such

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Christo PP, Gomez RS, et al.	Room Hospital das Clínicas, Universidade Federal de Minas Gerais, Tertiary Reference of the Unified Health System: Retrospective Analysis from 2000 to 2013			as culture and may include surgery to remove lesions. Access to appropriate treatment is crucial, especially in patients with immunosuppressive conditions, to reduce mortality associated with the disease.
Trapnell BC, Inoue Y, Bonella F, Morgan C, Jouneau S, Bendstrup E, et al.	Inhaled Molgramostim Therapy in Autoimmune Pulmonary Alveolar Proteinosis	2020	Not informed	This study evaluated the use of medications in patients with alveolar autoimmune proteinosis PAP (aPAP). The results showed that the experimental medication improved lung function. Study indicates that improved access to treatment improves quality of life.
Christophersen IE, Rienstra M, Roselli C, Yin X, Geelhoed B, Barnard J, et al.	Large-scale analyses of common and rare variants identify 12 new loci associated with atrial fibrillation	2014	USA	The research identified new genetic sites associated with atrial fibrillation, significantly expanding knowledge about its molecular bases. The research highlights potential targets for new pharmacological and surgical treatments.
Deppen SA, Blume JD, Kensinger CD, Morgan AM, Aldrich MC, Massion PP, et al.	Accuracy of FDG-PET to diagnose lung cancer in areas with infectious lung disease: A meta-analysis	2014	Japan	The main focus of the text is on the use of positron emission tomography (PET) with fluorodeoxyglucose (FDG) for the diagnosis of pulmonary nodules and on the accuracy of this method in specific contexts, enabling treatment as early as possible.
Lozano de Ávila CA, Martínez Estrada GA, Clason E Ramos, Duque Atencio V, Maza Manjarrez AP.	Clinical-pathological characterization and management of patients with chronic granulomatous mastitis in a center of high complexity	2021		Chronic Granulomatous Mastitis (CMM), a benign inflammatory condition of the breast. Management varies between antibiotic therapy, corticosteroids, observation, and, less frequently, surgery. The ideal treatment is not defined, being guided by the clinical presentation and preference of the treating physician.
Rahal E M, Moreno V M, Villa N J.	Mucormycosis rhino-orbito-cerebral: Experiencia en doce años	2008	Chile	Rhino-orbitocerebral mucormycosis is a rare and potentially lethal opportunistic infection caused by fungi. It mainly affects immunosuppressed patients. Treatment mainly involves

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				debridement surgery to remove necrotic tissue.
Malheiros JA, Oliveira CMD, Arantes Junior AA, Santos JSS, Gusmão SNS.	Prevalence And Management Of Refractory Meralgia Paresthetica In Lumbar Spine Surgery: 5 Years Of Experience	2020		Meralgia paresthetica is a compressive neuropathy, being observed in 22% of cases after lumbar spine surgery by subsequent approach. Initial treatment is conservative, with local measures and medications for neuropathic pain. Refractory cases can be treated with corticosteroid and anesthetic injections into the inguinal ligament region. Persistent cases may require decompression of the femoral cutaneous nerve or retroperitoneal neurectomy.
Wu P, Zhang Y, Zhang Y, Wang S, Fan Z.	Progress in the surgical treatment of sacrococcygeal pilonidal sinus: a review	2023	China	Pilonidal cyst is considered a rare condition, and treatment involves several surgical approaches, including techniques. Access to optimal treatment remains a controversial topic and varies by region, and there is a lack of consensus on the best surgical approach.
Phillips J, Subedi D, Lewis SC, Keerie C, Cronin O, Porteous M, et al.	Randomised trial of genetic testing and targeted intervention to prevent the development and progression of Paget's disease of bone	2023	Scotland	The study on zoledronic acid to prevent Paget's bone disease illustrates many of the challenges and considerations involved in accessing treatment in rare diseases. The research demonstrates the importance of focus and international cooperation are key to overcoming these challenges and ensuring that all patients, regardless of the rarity of their condition, have access to effective and safe treatments.
Zotes-Valdivia H, Alejandro Martínez-Arias M, Manuel Mier-Odriozola J, Morales-Gómez J, Joffre-Aliaga A.	Surgical treatment of pulmonary aspergilloma: 10 years of experience at the National Institute of Respiratory Diseases	2015	Mexico	Pulmonary aspergilloma is a rare condition caused by the growth of fungi (Aspergillus) in pre-existing pulmonary cavities, often associated with previous pulmonary tuberculosis, with surgery being the treatment of choice. Access to this treatment is essential for quality of life.
Liao C, Castonguay CE, Heilbron	Association of Essential Tremor	2022	Canada	One of the most common movement disorders, however, is bilateral and symmetrical

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K, Veikko Vuokila, Medeiros M, Houle G, et al.	With Novel Risk Loci			kinetic or postural tremors, which can reduce quality of life and impair daily functions. Studies in the form of treatment
Gao Y, Gao H, Wang G, Yin L, Xu W, Peng Y, et al.	A meta-analysis of Prognostic factor of Pancreatic neuroendocrine Neoplasms	2018	China	Pancreatic neuroendocrine neoplasms (pNENs) are rare and heterogeneous diseases of the pancreas, and their treatment is surgical resection. Access to and improvements in imaging techniques have increased the detection of pNENs, improving survival and quality of life.

DISCUSSION

The studies addressed emphasize the importance of access to innovative and effective treatments, expanding to various medical conditions, especially in rare diseases. To this end, continuous research and development of new therapies is needed that are crucial for improving patients' quality of life and clinical outcomes.

Personalization of treatments and international collaboration are important, and it is essential to face the challenges of access to health care. However, access to this personalization of treatments and surgical interventions for rare diseases is a challenge that impacts clinical outcomes and patients' quality of life. Recent studies reveal that several factors contribute to this difficulty, namely: the scarcity of specialized centers, late diagnosis, and inequalities in the health system.

According to the study by Davis et al., (2021), patients affected by rare diseases face some barriers in treatment, such as challenges in accessing surgical care, with the lack of specialized centers being the main obstacle. Thus, the absence of professionals trained in rare diseases leads to a long wait for treatment, aggravating patients' conditions and limiting treatment options.

Concomitantly, the study by Smith et al., (2020) argues that the education and improvement of health professionals is crucial to improve the recognition and management of these diseases, where training programs can reduce the time to diagnosis and facilitate access to appropriate surgical care.

Even so, Johnson and Liu (2021) pointed out the importance of early diagnosis for success in rare disease surgeries, that is, interventions being performed at early stages can increasingly improve outcomes and reduce mortality.

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Another contributing factor to difficulty in accessing the treatment of rare diseases is regional inequalities in access to health services that are evident, resulting in significant disparities in treatment (Davis et al., 2021).

Technological innovations, on the other hand, as they are minimally invasive approaches, have great potential to improve access and prognostic outcomes for patients. These techniques reduce complications and speed up recovery, facilitating the postoperative period for patients who already face additional challenges (Davis et al., 2021).

Confirming the results of a study by Hirai et al. (2016), which addresses that in colorectal cancer, the relevance of genetic advances in personalized medicine for rare diseases is highlighted, given that the discovery of new markers can lead to the development of prevention strategies and more appropriate treatment, reducing the incidence and lethality of the disease.

Technological advances also permeate the standard treatment, which may involve treatments at the level of those who cannot undergo surgery, as discussed by Samson et al. (2022), who demonstrate that the administration of oral octreotide capsules provides durable biochemical responses and safety in the treatment of acromegaly, and traditionally the standard treatment involves surgical removal of the pituitary tumor.

The multidisciplinary approach also becomes fundamental for the patient experience and optimization of postoperative outcomes, as collaboration between surgeons and rare disease specialists becomes essential for treatment efficiency (Anderson et al., 2022).

As in the study by Rahal et al. (2021), which reinforces the need for a multidisciplinary approach, since, for effectiveness in the management of chronic granulomatous mastitis, there is a need for the approach to be personalized and interdisciplinary, requiring a treatment adapted to the individual characteristics of the patient.

In Brazil, when it comes to public policies aimed at rare diseases, the objective is to improve the quality of life of patients, promote early diagnosis and ensure access to effective treatment, as provided for in the National Policy for Comprehensive Care for People with Rare Diseases, which provides for the articulation between the levels of the Unified Health System (SUS) and the inclusion of these diseases in the list of diseases treatable and monitored by the public system (Brazil, 2014).

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In addition, this policy aims to integrate services and ensure access to diagnosis and treatment, including complex surgeries. Other relevant actions include: implementation of reference centers and specialized in rare diseases and definition of standards for standardized treatments, facilitating the necessary interventions (Brasil, 2014).

The SUS includes the treatment of rare diseases, through programs such as the availability of drugs for the treatment of rare diseases, which are carried out through specific therapeutic protocols and the exceptional drugs program. It also promotes the incorporation of medicines, through the National Commission for the Incorporation of Technologies in the SUS (CONITEC), which plays a central role in the analysis and incorporation of treatments (Brasil, 2014).

Another focus of public policies within the scope of the SUS is awareness and professional training, since these diseases can be difficult to identify without adequate training. However, as previously addressed by the Davis et al. study (2021), the reality is the mismatch of trained professionals, making it difficult to access adequate treatment (Brasil, 2014).

In the European Union (EU), rare disease policy has been a priority since the 2000s, when a recommendation was adopted that encourages member countries to develop national plans for rare diseases. Another support created was the *European Reference Networks* (ERNs), which are networks that connect specialists from different countries, with the aim of providing support to patients and doctors, as well as incentives for the development of drugs for the treatment of rare diseases (European Commission, 2021).

Currently, France is one of the countries that leads public policies on rare diseases. In 2004, the country implemented specific national plans to improve the diagnosis and treatment of rare diseases, which include financial support for surgical interventions and complex care. Another novelty was the competence centers that work together with the reference centers, with the aim of facilitating access to specialized treatments (France, 2018).

In Canada, the approach adopted is based on partnerships with provinces and federals, which are province-specific programs with federal partnerships to support treatments, with cooperation with university hospitals. Still, the *Canadian Organization for Rare Disorders (CORD)* was the creator of the strategy for rare diseases, which aims at early detection, access to treatment, and financial support (CORD, 2015).

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Public policies for surgical and non-surgical treatment of rare diseases vary by country, but they all share the goal of ensuring adequate access to specialized medical care. However, significant challenges still persist, even in countries with well-developed strategies, that is, even with the existence of public policies, there are still obstacles that impact clinical outcomes and quality of life of patients.

CONCLUSION

The study revealed the importance of access to effective treatment for rare diseases, which is a challenge faced by patients around the world. Access to personalized treatments and multidisciplinary approaches have proven to be fundamental to optimize the time to clinical results, but still, the obstacles remain significant.

The public policy programs of each country can include everything from public funding to the creation of specialized centers that ensure high-quality care. These programs play a crucial role in ensuring access to treatments for rare diseases, promoting early diagnosis, and ensuring treatment.

Finally, the experience of other countries shows that even with well-structured policies, there are still considerable challenges, such as the lack of trained professionals and regional inequalities in access to care. Continuous effort is needed to improve access to specialized medical care, ensure the training of trained professionals and innovation in treatments.

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